ΑΔΑ: ΨΒΦ346Ψ Ζ2Ν-8ΥΘ
ΕΚΠΑ 27/04/2023
Α. Π.: Εισερχ. 39517

ΑΙΤΗΣΗ

ΕΠΩΝΥΜΟ: ΑΝΤΩΝΑΡΑΚΗΣ
ΟΝΟΜΑ: ΣΤΥΛΙΑΝΟΣ
ΠΑΤΡΩΝΥΜΟ: ΕΜΜΑΝΟΥΗΛ
ΗΜΕΡ.ΓΕΝΝΗΣΗΣ: 11 ΔΕΚ 1951.
ΙΔΙΟΤΗΤΑ: Ομότιμος Καθηγητής
Α.Δ.Τ./Αρ. Διαβατηρίου:..............................
E-mail: stylianos.antonarakis@unige.ch

Θέμα: *Υποψηφιότητα για τη θέση εξωτερικού μέλους του Συμβουλίου Διοίκησης του Εθνικού και Καποδιστριακού Πανεπιστημίου Αθηνών*

Τόπος/Ημερομηνία:
Γενεύη, 26.4.2023

Συνημμένα:

α) Αντίγραφο Δελτίου Αστυνομικής Ταυτότητας ή Αντίγραφο Διαβατηρίου.
β) Πλήρες βιογραφικό σημείωμα.
γ) Οποιαδήποτε έγγραφα ή στοιχεία κρίνει ο/η υποψήφιος/α ότι θα υποστηρίξει την υποψηφιότητά του/ης (προαιρετικό).
δ) Επιστολή εκδήλωσης ενδιαφέροντος για τη θέση του εξωτερικού μέλους .

ΠΡΟΣ
ΕΘΝΙΚΟ ΚΑΙ ΚΑΠΟΔΙΣΤΡΙΑΚΟ ΠΑΝΕΠΙΣΤΗΜΙΟ ΑΘΗΝΩΝ

Με την παρούσα αίτηση:

α) υποβάλλω υποψηφιότητα για τη θέση εξωτερικού μέλους του Συμβουλίου Διοίκησης του Εθνικού και Καποδιστριακού Πανεπιστημίου Αθηνών σύμφωνα με την ισχύουσα νομοθεσία στο πλαίσιο της με αριθμ. πρωτ. 17890/28-2-2023. διεθνούς πρόσκλησης για την ανάδειξη των εξωτερικών μελών του Συμβουλίου Διοίκησης του Ιδρύματος,

β) αποδέχομαι τους όρους συμμετοχής στην παρούσα και δηλώνω ότι γνωρίζω τις υποχρεώσεις που απορρέουν από την ιδιότητα του εξωτερικού μέλους σε περίπτωση εκλογής μου, καθώς και τις αρμοδιότητες που ασκεί το Συμβούλιο Διοίκησης του Ε.Κ.Π.Α. σύμφωνα με το άρθρο 14 του ν. 4957/2022,

γ) δηλώνω ότι συναινώ στη συλλογή και επεξεργασία των προσωπικών δεδομένων μου, όπως αυτά αναφέρονται στην παρούσα πρόταση και στα συνυποβαλλόμενα με αυτήν δικαιολογητικά αποκλειστικά για την αξιολόγηση της αίτησής μου στο πλαίσιο της διαδικασίας της παρούσας.

Σ.Ε. Αντωναράκης
Στυλιανός Αντωναράκης
Ο Αιτών / Η Αιτούσα
(ονοματεπώνυμο και ψηφιακή υπογραφή)

CURRICULUM VITAE

Stylianos Emmanuel **Antonarakis**, M.D., D.Sc.

Current Academic Appointments

- **Professor Emeritus (active), University of Geneva Medical School, Geneva Switzerland.**

Until September 30th, 2017

- **Professor of Medical Genetics, University of Geneva Medical School, Geneva Switzerland.**
- **Founding Chairman, Department of Genetic Medicine and Development, University of Geneva Medical School, Geneva Switzerland.**
- **Founder and first Director, Institute of Genetics and Genomics in Geneva, iGE3; University of Geneva.**
- **Director, Division of Genetic Medicine, University Hospitals of Geneva.**
- **Director, Center for Genetic Medicine, University Hospitals of Geneva.**
- **Invited Professor; Université Paris Descartes and Hôpital Universitaire Necker-Enfants Malades, Imagine Institute, Paris, France.**

Professional Address

Department of Genetic Medicine and Development
Centre Medical Universitaire - CMU 9
1 rue Michel Servet, 1211 Geneva 4 , Switzerland
Tel : 41-22-379-5707 or 8 Fax : 41-22-379-5706
Email: Stylianos.Antonarakis@unige.ch
http://www.hugo-international.org/HUGO-Past-Presidents
http://en.wikipedia.org/wiki/Stylianos_Antonarakis

Private Address

16 Rue Albert-Gos
1206 Geneva, Switzerland

Personal data

Birth **December 11, 1951** Athens, Greece
Family: Spouse: Grigoria Grigoriou, 1953
Children: Emmanuel, 1978; Gregory, 1979; Alexander, 1982; Christina, 1990.

Citizenships USA, Switzerland, & Greece

Education

Athens University School of Medicine, 1969-1975
M.D. degree (magna cum laude) 1975
Athens University School of Medicine
Doctoral Thesis (magna cum laude) 1983

Internship and Residency

Internal Medicine, 1976-1978
King Paul's University Hospital, Dept. of Medicine (Prof. G. Daikos)
Hellenic Air Forces Gen. Hospital, Dept. of Medicine (Dr. G. Psimenos)
Pediatrics, 1978-1980

Kozani General Hospital, Dept. of Pediatrics (Dr. P. Economopoulos)
Patras Children's Hospital, Dept. of Pediatrics (Prof. Th. Giogarakis)
Aghia Sophia University Children's Hospital, Dept. of Pediatrics (Prof. N. Matsaniotis)

Licenses to Practice Medicine

Athens, Greece 1975
State of Maryland, USA, 1984 License # D304
Switzerland, FMH Medical Genetics, License # 052057-51
Switzerland, FAMH Medical Genetics Diagnostic Laboratory

Board Certification

ECFMG Certificate: January 1975
FLEX Examination: January 1984 (State of Maryland)
Hellenic National Board of Pediatrics, 1980
American Boards of Medical Genetics Clinical Geneticist, 1984
Clinical Biochemical Geneticist, 1984
Molecular Geneticist, 1993
Swiss FMH in Medical Genetics (founding member), 2000
Swiss FAMH in Genetic Laboratory Analyses (founding member), 2002

Post-Doctoral Fellowship

The Johns Hopkins University School of Medicine
Department of Pediatrics, Genetics Unit (Jul 1980 - Mar 1983)
Professor Haig H. Kazazian, Jr., MD
Professor Victor A McKusick, MD

Academic Appointments

1982-83 Research Associate of Pediatrics, The Johns Hopkins Univ School of Medicine
1983-85 Assistant Professor of Pediatrics, The Johns Hopkins Univ School of Medicine
1985-91 Associate Professor of Pediatrics, The Johns Hopkins Univ School of Medicine
1990-91 Associate Professor of Medicine, The Johns Hopkins Univ School of Medicine
1986-94 Faculty, Human Genetics graduate Ph.D. program, The Johns Hopkins University School of Medicine
1989-96 Professor of Human Genetics, University of Crete Medical School.
1991-92 Deputy Director, Postdoctoral Fellowship Program in Human Genetics, JHUSM
1991-94 Professor of Pediatric Genetics, Center for Medical Genetics, The Johns Hopkins University School of Medicine.
1994-98 Professor of Pediatrics (Part-time appointment), Center for Medical Genetics, The Johns Hopkins University School of Medicine.
1991-94 Joint appointment (Professor) in the Department of Medicine, Center for Medical Genetics, The Johns Hopkins University School of Medicine.
1991-94 Joint appointment (Professor) in the Department of Biology, The Johns Hopkins University School of Arts and Sciences.
1992- Professor of Medical Genetics, University of Geneva Medical School.
1992-2017 Chief, Division of Medical Genetics, University of Geneva Medical School.
2004-2017 Chairman, Department of Genetic Medicine and Development, University of Geneva Medical School.
2003- Member (corresponding) of the Athens Academy of Arts and Sciences.
2011-17 Founder and first Director, Institute of Genetics and Genomics in Geneva, iGE3; University of Geneva.
2017- Honorary Member (elected; highest membership), Swiss Academy of Medical Sciences.

Hospital Appointments

1984-94 Medical Staff, The Johns Hopkins University Hospital.
1992-2017 Chief of Genetic Medicine, University Hospitals of Geneva.
2011-2017 Director, Genetic Medicine Center, University Hospitals of Geneva

Other Appointments

1989-93 Chairman, Consortium CEPH Linkage map for human chromosome 21
1991-93 Editor, Chromosome 21, Genome database and HUGO.
1993-96 Senior editor, Chromosome 21, Genome database and HUGO.
1992 Organizer; Chromosome 21 Third International Workshop, Baltimore, MD
1993- Editor, Mutation database for Mendelian Inheritance in Man (OMIM).
1995-00 Member, Scientific Program committee, European Society of Human Genetics
1996- Member, WHO Expert Advisory Panel on Human Genetics
1996- Chair, Human Mutation Nomenclature Committee; HUGO
1997-99 Member, Geneva University Medical School, Promotions Committee to Privat-Docents
1999 Organizer; European Society of Human Genetics meeting, Geneva, Switzerland
1999 Co-organizer; Chromosome 21 Eighth International Workshop, Rehovot, Israel
1999-present Chair, Geneva University Medical School, Promotions Committee to Privat-Docents
1999-03 Member, Geneva University Medical School, Committee for Academic Renewal
1997-03 Member, Geneva University Medical School, Committee for Research
2000-06 Elected member of the HUGO Council
2001-02 President, European Society of Human Genetics (President-Elect 2000-2001; vice-president 2002-2003)
2012-17 President of HUGO (Human Genome Organization) 2013-2017 (President Elect 2012-2013); 2017 President Emeritus
2000-07 Co-organizer; European School of Medical Genetics; Sestri Levante or Bertinoro, Italy.
2007-present Editor, Metabolic and Molecular basis of Inherited Disease (http://beta.ommbid.impelsys.com/)
2011-present Member, Bioethics committee of the Eastern Churches.
2016-17 Invited Professor; Université Paris Descartes and Hôpital Universitaire Necker-Enfants Malades, Paris, France
2018-present CoFounder and CEO, MediGenome, Swiss Institute of Genomic Medicine, Geneva, Switzerland (www.medigenome.ch).
2022-present CoFounder and Chairman of the steering committee, Greek Institute of Human Genomics (Ελληνικό Ινστιτούτο Γονιδιωματικής του Ανθρώπου ΕΙΓΑ), ITE, Athens, Greece.

Clinical Experience

- General Pediatrics
- Pediatric and Adult Medical Genetics
- Genetic Counseling
- DNA diagnostic laboratory analyses
- Molecular Cytogenetic analyses

Diagnostic laboratory Experience

- DNA diagnostic laboratory analyses
- Molecular Cytogenetic analyses

Research Interests (Past and Present):

The overall objective of my research is to understand how genetic variation causes phenotypic variation.
More specifically, the specific goals of the research in our laboratory (since 1982) were as follows:

- Molecular characterization of single gene disorders including the thalassemias, and Hemophilias.
- DNA polymorphisms in clinical medicine.
- Eukaryotic gene expression.
- Gene mapping using DNA polymorphisms in linkage analysis.
- Mechanisms involved in the break and reunion of chromosomes in ring formation.
- Molecular predisposition to Down syndrome, mechanism of non-disjunction.
- Molecular pathophysiology of Down syndrome.
- Apolipoprotein genes and premature coronary artery disease.
- Erythropoietin gene expression in transgenic mice.
- Cloning of unknown disease-related genes by positional cloning.
- Yeast artificial chromosome mapping.
- Insertional mutagenesis in human disorders (L1 repetitive element insertions).

- Human Genome Project; mapping and sequencing of human chromosome 21 and cloning of its genes.
- Mapping genes for multigenic disorders including schizophrenia.
- Mapping and cloning of genes responsible for developmental abnormalities.
- Gene therapy of hereditary disorders including hemophilia A.
- Molecular basis of hereditary deafness.
- Differential gene expression in aneuploidies.
- Molecular pathophysiology of disorders due to repeat expansion (e.g. 12mer repeat of CSTB in EPM1).
- Evolutionary history of the human genome.
- Functional analysis of Conserved Non-Coding DNA sequences.
- Genetic control of gene expression variation.
- Genetic analysis of somatic cell disorders
- Consanguinity to discover recessive alleles
- Single cell genomics
- Chromatin structure in aneuploidies

Laboratory Experience

- Isolation of DNA and RNA from various tissues
- Southern blot analysis
- Cloning on viral and plasmid vectors
- DNA sequencing
- Cell culture
- Linkage analysis of monogenic traits
- Linkage analysis of polygenic complex traits
- Linkage analysis of quantitative traits
- Association studies for complex traits
- Chromosome analysis, in situ hybridization
- DNA methodology for mutation detection
- Oligonucleotide hybridization
- Pulsed field gel electrophoresis
- Denaturing gradient gel electrophoresis
- Polymerase chain reaction
- Analysis of mutations in nucleic acids
- Transgenic and "knock-out" mice
- In situ hybridization
- Cellular and subcellular localization of proteins
- Cloning in yeast artificial chromosomes (YACs)
- Cloning by exon trapping, cDNA selection
- Protein interactions by Yeast two-hybrid
- Differential gene expression using SAGE
- Differential gene expression using microarrays
- SNP detection
- Gene expression variation using Real-Time quantitative PCR
- DNA genotyping with microarray technology
- Genomic Copy Number Variants with microarray technology
- Chromatin conformation capture
- High-throughput sequencing
- Chromatin immunoprecipitation (ChIP)
- Single cell genomics

Teaching Experience

- Clinical Pediatrics to Medical Students and Residents, JHU
- Clinical Biochemistry to Medical Students, JHU
- Human Genetics to Medical Students, JHU, UGMS 1983-2004
- Participation in the various activities of the Genetics Center of JHU (1981-1993) and UGMS (1993-date)
- Invited Speaker to meetings, symposia and workshops, including Gordon Conferences (since 1985) and Cold Spring Harbor Symp., Quant. Biology

- Faculty, Bar Harbor course for Mammalian Genetics, 1982-2022.
- Faculty, European School of Medical Genetics, 1993-2008 (co-organizer in 2000-2007)
- Faculty, Advanced Human Genetics Course, JHU, 1984-1990
- Faculty, Pathophysiology Course, JHU, 1983-1990
- Co-organizer, weekly genetics conference, 1984-1985, JHU
- Organizer, biweekly Medical Genetics Journal Club, 1992-date, UGMS
- Faculty, Human Genetics Course for Graduate Students, JHU, 1986-1993
- Director, Human Genetics Course for Graduate students, JHU, 1990-1992
- Director, Human Genetics Course for Graduate students, UniGE MedSch, 1993-2020
- Member; Human Genetics Course for Graduate students, UniGE NCCR graduate program 2002-date
- Faculty, Human Genetics Course for biology students, UniGE Science School 1995-date
- Member; Course to 2nd year Medical Students of UniGE MedSch on Human Reproduction (Problem-based learning) 1997-2019
- Organizer and member; Course to 1st year Medical Students of UniGE MedSch on Human Medical Genetics 2004-2017
- The Division of Medical Genetics of the University and Hospital of Geneva is one of the 4 centers in Switzerland for the training in Medical Genetics (this training was under the responsibility of S.E. Antonarakis)
- Faculty, Wellcome Trust School on Human Genomics, Sanger Institute (2009-2011)

Graduate Students[1], Postdoctoral Fellows[2], Students on laboratory rotations[3], Undergraduate students[4], MD thesis students[5]

1. Corinne Wong, M.D., Ph.D.[1]
2. Mary Kay McCormick, Ph.D.[1]
3. Andrew C. Warren, M.D., Ph.D.[2]
4. Hagop Youssoufian, M.D.[2]
5. John A. Ladias, M.D.[2]
6. Patricia Woods-Samuels, Ph.D.[1]
7. Gregg L. Semenza, M.D., Ph.D.[2]
8. Effrosini P. Economou, M.D.[2]
9. Hiroshi Inaba, Ph.D.[2]
10. Garry R. Cutting, M.D.[2]
11. Miyoko Higuchi, Ph.D.[2]
12. Susan Kass, Ph.D.[3]
13. Claudia R. Colyer, Ph.D.[3]
14. Lisbeth A. Guethlein, Ph.D.[3]
15. Michael B. Petersen, M.D., Ph.D.[2]
16. Andrew W. Bergen, Ph.D.[1]
17. Josef Coresh, M.D., Ph.D.[3]
18. Valerie L. Prenger, Ph.D.[3]
19. Felicity Collins, M.D.[2]
20. Michelle Makos, Ph.D.[3]
21. Marianna M. Kalaitsidaki, Ph.D.[2]
22. Melvin G. McInnis, M.D.[2]
23. Dimitris Avramopoulos, M.D., Ph.D.[1,2]
24. Matthew J. McGinniss, Ph.D.[2]
25. Nicola De Marchi, M.D.[2]
26. Sylvia Neuman, Ph.D.[2]
27. Haiming Chen, M.D.[2]
28. Jean-Louis Blouin, Ph.D.[2]
29. Will Lowther, Ph.D.[3]
30. Pierre Hutter, Ph.D.[2]
31. Maria D. Lalioti, Ph.D.[1]
32. Derek Christie[3]
33. Sonia N. Samec, Ph.D.[3]
34. Roman Chrast, Ph.D.[1,4]
35. Gaelle Perrin, Ph.D.[4]
36. Colette Rossier, Ph.D.[2]
37. Ariane Paoloni-Giacobino, M.D.[2]
38. Marguerite Neerman-Arbez, Ph.D.[2]
39. Jian Ying Su, M.D.[2]
40. Uppala Radhakrishna, Ph.D.[2]
41. Hamish S. Scott, Ph.D.[2]
42. Silvana Guidi, M.Sc.[4]
43. Laureane Mittaz, Ph.D.[4]
44. Maria Mirotsou Ph.D.[3]
45. Hamid Mehenni, M.D.[2]
46. Despina Kyriakou, M.D.[2]
47. Michel L. Guipponi, Ph.D.[2]
48. Logos Curtis, M.D., Ph.D.[3]
49. Alexandra Schebesta, M.D.[3]
50. Joelle Michaud, Ph.D.[4]
51. Lucia Bartoloni, Ph.D.[2]
52. Marie Wattenhofer, Ph.D.[1,4]
53. Amit L. Maiti, Ph.D.[2]
54. Anouk Imhof, M.D.[2]
55. Samuel Deutsch, Ph.D.[1,2]
56. Olivier Menzel, Ph.D.[4]
57. Fabien Chapeau[4]
58. Robert Lyle, Ph.D.[2]
59. Pascal Chanson[4]
60. Sura A Alwan[4]
61. Alexandre Reymond, Ph.D.[2]
62. Mark Friedli, Ph.D.[1,4]

63. Charlotte Henrichsen, Ph.D.[4]
64. Veronique Falciola [4]
65. Vassilis Doucas, Ph.D.[2]
66. Urmila Choudhury [1,3]
67. Nathalie Lin-Marq, Ph.D.[2]
68. Caroline Tapparel, Ph.D.[2]
69. Manolis T. Dermitzakis, Ph.D.[2]
70. Leonore Rougemont [4]
71. Laurent Cimasoni, M.D.[1]
72. Cedric Howald, Ph.D.[4]
73. Christine Lamon [4]
74. Renaud Desgraz, Ph.D.[3]
75. Sarantis Gagos, Ph.D.[2]
76. Catia Attanasio, Ph.D.[1]
77. Bastien Braillard [4]
78. Carole Burgi [4]
79. Anna Moreno-Bernat [4]
80. Leila Parant, M.Sc.[4]
81. Homa Attar-Cohen, Ph.D.[1,3]
82. Anne-Elisabeth de la Barre, Ph.D.[2]
83. Cristelle Borel, Ph.D.[2]
84. Gipsy Lopez, M.Sc.[4]
85. Irina Surkova, M.D.[2]
86. Paola Prandini, Ph.D.[2]
87. Sergei I. Nikolaev, Ph.D.[2]
88. Lakshman Subrahmanyan [3]
89. Marilena Papaioannou, Ph.D.[3]
90. Daniel Robyr, Ph.D.[2]
91. Bernard Conrad, M.D.[2]
92. Periklis Makrythanasis, M.D.[2]
93. Tetsuji Oishi [3]
94. Natasha Farquet [3]
95. Romain Seppey [3]
96. Christelle Stouder M.Sc.[4]
97. Stefania Gimelli, Ph.D.[2]
98. Andrew J. Sharp, Ph.D.[2]
99. Audrey Letourneau, Ph.D.[1,3]
100. Elizabeth Stathaki, M.Sc.[4]
101. Mohamed Reza Sailani, Ph.D.[1,3]
102. Eugenia Migliavacca, Ph.D.[2]
103. Konstantin Popadin, Ph.D.[2]
104. Mari Nelis, Ph.D.[2]
105. Ximena Bonilla Ph.D.[1,3]
106. Federico Santoni, Ph.D.[2]
107. Celine Brockmann, Ph.D.[2]
108. Marianna Bustamante, M.Sc.[4]
109. George Stamoulis [1,3]
110. Patrick Callier, Ph.D.[2]
111. Mafalda Mucciolo, Ph.D.[3]
112. Sonia Amabile, M.D.[3]
113. Teresa Didonna, M.Sc.[4]
114. Marco Garieri [1,4]
115. Monica Albarca Aguilera, Ph.D.[2]
116. Marco Emilio Poleggi, Ph.D.[2]
117. Eva Hammar, Ph.D.[2]
118. Andreas Massouras, Ph.D.[2]
119. Mohammad Ansar, Ph.D.[2]
120. Vassilis Dionellis, M.Sc.[4]
121. Rouaa ben Chaabene, M.Sc.[4]
122. Maurus Locher M.Sc. [1]
123. Ambra Sartori, M.Sc.[4]
124. Annick Trachsler-Salzmann, Ph.D.[2]
125. Xavier Blanc, M.Sc.[4]
126. Emmanuelle N. Ranza, M.D.[5]
127. Fedor Bezrukov, Ph.D.[2]
128. Czuee Morey, Ph.D.[2]
129. Sayaka Omori M.Sc.[2]
130. Naila Batool [1,3]
131. Tahir Sarwar, Ph.D. [2]
132. Ilya Kolpakov, Ph.D. [2]

Clinical Trainees

1. Albiru Farchadi D.
2. Aliferis K.
3. Al Petracco J.-C.
4. Arzel Andereggen B.
5. Aubry C.
6. Broly M.
7. Brun N.
8. Barker-Schaerer M.
9. Carminho M.T.
10. Cediel L.
11. Chappuis-Bretton B.
12. Conrad B.

13. Chevalier I.
14. Cottet L.
15. Couchepin F.
16. Curtis L.
17. Curtis Bruel J.
18. Dallèves F.
19. D'amato Sizonenko L.
20. Dutoit Marie-H.
21. Egger Jean-F.
22. Ejnès S.
23. ElAyeb Y.
24. Eliez S.
25. Fankhausen L.
26. Filges I.
27. Gersbach M.
28. Giacobino A.
29. Gil R.
30. Goza E.
31. Gwamnesia L.
32. Hamami H.
33. Hariri A.
34. Kern I.
35. Kiraly C.
36. Lacroix L.
37. Lidgren M.
38. Makrythanasis P.
39. Quteineh L.
40. Ranza E.
41. Salvadiray Orag N.
42. Santosh I.
43. Varvagiannis K.
44. Williamson C.
45. Wonkam A.
46. Yerly S.
47. Zufferey R.

Scientific Review Experience

- Member, Special Study Section for Cystic Fibrosis Research, NIH, 1984, 1988, 1989.
- Member, Department of Energy Biology Review, 1990
- Member, Human Genome Study Section, NIH, 1990-1991
- Member, Mammalian Genetics Study Section, NIH 1991-1995
- Member, Program Project site visit committees, NIH, 1985-1993 (including Sickle Cell Centers, Human Genome Research Centers)
- Reviewer for the Human Frontier Research Program, European Union grants, Swiss National Science Foundation, German Human Genome Project program, MRC of the UK, The Netherlands Science Foundation, Israel Science Foundation, Welcome trust.
- Member, Scientific Committee of the Italian Telethon Research Grants, 2000-2004.
- Member, Swiss National Research Council 2004-2013, 2015-2016.
- Member, Swiss National Foundation, Ambizione grants 2014-2017
- Chair, European Research Council (ERC) 2008, 2010, 2012, 2015. LS2 Section Genetics, Genomics, Systems Biology
- Member, Ad-hoc committee to evaluate the Director of the NIH, 2015.

Scientific Advisory Boards

- Member, Scientific Advisory Board (1992-1999); The Cyprus Institute for Genetics and Neurology, Nicosia, Cyprus.
- Member, Scientific Advisory Board (2002-2012); Center for Genomic Regulation, Barcelona, Catalunya, Spain.
- Member, Scientific Advisory Board (2004-2010); Institute for Biomedical Research, Athens Academy, Greece.
- Member, Scientific Advisory Board (2005-2014); The Center for Applied Genomics, University of Toronto, Canada.
- Member, Scientific Advisory Board (2005-present); Fondation "Imagine" Necker Hospital, Paris, France.
- Member, Scientific Advisory Board (2011-2014); Linda Crnic Inst. for Down Syndrome, Univ Denver, Colorado, USA.

Grant support

- New Investigator Research Award 1 R23 HL31503 from National Heart, Lung and Blood Institute, 1983-86
- March of Dimes-National Foundation, Basic Research Grant, 1953, 1985-87
- Principal Investigator, Grant 1R01 HD19591 from National Institutes of Health 1985-88.
- The Johns Hopkins University School of Medicine Institutional Grant 1985-86
- Principal Investigator and since 1994 co-investigator, grant 5R01 HL38165 from the National Institutes of Health, 1987-2000.
- Principal Investigator, Grant 5R01 DK39869 from the National Institutes of Health, 1987-90.
- Project Director, Grant 1PO1 GM41015 from the National Institutes of Health, 1988-93
- Project Director, Grant PO1-HD24605 from the National Institutes on Health, 1989-94.
- Project Director, Grant PO1-HG00373 from the National Institutes on Health, 1988-93.
- Principal Investigator, U.S. Dept of Energy DE-FG02-88ER60-85, 1989-93.
- Co-investigator, Grant 1R01 MH45588 from the National Institutes of Health, 1989-93

- Principal Investigator, Grant 1R13 HG00694 from the National Institutes of Health, 1992-93
- Principal Investigator, Grant 1R01 HG00468 from the National Institutes of Health, 1992-96.
- Principal Investigator, Grant 31-33965.92 from the Swiss National Science Foundation 1992-94.
- Co-Investigator, Grant PL-930015 from the European Community 1994-96.
- Principal Investigator, Grant 31-40500.94 from the Swiss National Science Foundation 1994-1999.
- Principal Investigator, Grant 95-667 from the Swiss League against Cancer 1995-97.
- Co-Investigator, Grant PL-950302 from the European Community 1996-98.
- Co-Investigator, Grant PL-950554 from the European Community 1996-99.
- Co-Investigator, Grant BMH-CT98-3039 from the European Community 1998-2001.
- Principal Investigator, Grant 4038-52845 from the Swiss National Science Foundation 1998-2000.
- Principal Investigator, Grant KFS 724-9-1998 from the Swiss League against Cancer 1999-2001.
- Principal Investigator, Grant KFS 1085-9-2000 from the Swiss League against Cancer 2001-2002.
- Principal Investigator, Grant 31-57149.99 from the Swiss National Science Foundation 1999-2004.
- Principal Investigator, Grant from the Foundation "Child Care" 2000-2005.
- Co-investigator, Grant NCCR 51NF40-11137(Centers of Excellence; Frontiers in Genetics) from the Swiss National Science Foundation 2001-2012.
- Principal Investigator, Grant OCS 1184-9-2001 from the Swiss Federation against Cancer 2002-2004.
- Co-Investigator, Grant QLG1-CT-2002-00816 (T21 targets) from the European Community 2002-2004.
- Principal Investigator, Grant 31-102231 from the Swiss National Science Foundation 2003-2006.
- Co-PInvestigator, Grant HG-03-003 ENCODE, from the National Institutes of Health, 2003-2007.
- Co-Investigator, Grant LSHG-CT-2003-503265 "Biosapiens" from the European Community 2004-2008.
- Principal Investigator, Grant 31-105602 from the Swiss National Science Foundation 2004-2009.
- Co-Investigator, Grant LSHG-CT-2004-512003 "EureXpress" from the European Community 2005-2008.
- Principal Investigator, Integrated Project Grant LSHG-CT-2006-037627 "AnEUploidy" from the European Community FP6 2006-2010.
- Co-PInvestigator, Grant U54 HG004557 ENCODE, from the National Institutes of Health, 2007-2010.
- Principal Investigator, R'Equip Grant 31-113127 from the Swiss National Science Foundation 2006-2007.
- Principal Investigator, R'Equip Grant 31-121418 from the Swiss National Science Foundation 2008-2009.
- Principal Investigator, Grant 31-127375 from the Swiss National Science Foundation 2009-2012.
- Principal Investigator, ERC (European Research Council) Advanced grant 249968 HuCNCs from the European Community FP7 2010-2014.
- Principal Investigator, Gebert Foundation for Rare Disorders grant GRS-047/10, 2011-2014
- Co-Investigator, Grant LSHG-CT-2011-282510 "Blueprint" from the European Community 2011-2016.
- Co-investigator, Grant NCCR (Centers of Excellence; Synapsy) Swiss National Science Foundation 2010-2016.
- Principal Investigator, Bonus of Excellence Grant 31-144082 from the Swiss National Science Foundation 2012-2015.
- Co-Investigator, Grant from SystemsX 2013-2016.
- Principal Investigator, grant from the ProVisu Foundation for Inherited Eye Disorders, 2014-2016.
- Principal Investigator, Grant 31-163180 from the Swiss National Science Foundation 2015-2018.
- Principal Investigator, grant from the ProVisu Foundation for Inherited Eye Disorders, 2019-2021.
- Principal Investigator, grant from the Aclon Foundation 2019-2020.

Honors - Awards

- First among 7500 candidates in The National Exam to enter the Athens Medical School, 1969
- Honor Student, Greek National Scholarship Foundation, (1969-1975)
- The Johns Hopkins Univ. School of Medicine Award for Postdoctoral Investigation, 1982
- Society of Pediatric Research: Young Investigator Award, 1984 (Ped. Res.18: 17A, 1984)
- Hippocrates Orator, London 1988, Hellenic Medical Society.
- Medal of Honor, Tokyo Medical College, 1989, Tokyo, Japan.
- American Academy of Pediatrics: Excellence in Pediatric Research Award 1992.
- Mauro Baschirotto Foundation Grant for Rare Disorders 1998-99.
- Linus Pauling Award and Lecture; Italian Society for Applied Molecular Medicine, 1997.
- Visiting Professor, India Institute of Medical Sciences, Delhi and Indian Institute of Science, Bangalore, India; 1998.
- Maria Lalioti (graduate student); young investigator award, European Society of Human Genetics 1998
- Maria Lalioti (graduate student); young investigator award, American Society of Human Genetics 1998
- M Neerman-Arbez (postdoctoral fellow); young investigator award, European Soc. Human Genetics 1999.
- Hamish S Scott (postdoctoral fellow); young investigator award, American Soc. Human Genetics 1999.

- Pfizer Prize for Research in Neurosciences, 1999 (with my graduate student Maria Lalioti).
- Roman Chrast (graduate student); young investigator award, European Society of Human Genetics 2000.
- Marie Wattenhofer (graduate student); Arditi Prize, Geneva 2001.
- Katia Attanasio (graduate student); Arditi Prize, Geneva 2002.
- Listed as one of the most cited scientists in the literature by the Institute for Scientific Information (http://isihighlycited.com)
- International Jerome Lejeune Prize, 2004, Paris, given by J-F Mattei, French minister of Health.
- 2003- Member (corresponding) of the Athens Academy of Arts and Sciences
- ESHG 2005 Award, from the European Society of Human Genetics (www.eshg.org/eshg_award.htm).
- Society of Scholars 2006; The Johns Hopkins University.
- Member European Molecular Biology Organization 2006 (www.embo.org/communities/members.html)
- Originator of the idea for an International Down Syndrome Day on March 21st. (http://en.wikipedia.org/wiki/World_Down_Syndrome_Day)
- Sam Deutsch (postdoctoral fellow); young investigator award, European Society Human Genetics 2006.
- Medal "Commander of the Order of the Phoenix" from the President of the Greek Democracy, Mr K. Papoulias, 2007.
- Swiss Society for Infectious Diseases Award 2008 (to all Swiss co-authors of Fellay et al Science 19oct07) http://www.sginf.ch/ssi-home/awards/announcement-ssi.html
- Andrew J Sharp (postdoctoral fellow); young investigator award, European Society Human Genetics 2009.
- ERC (European Research Council) Advanced grant 249968 HuCNCs from the European Community FP7 2010-2014.
- 2010- Elected to the AAP (Association of American Physicians; for the advancement of scientific and practical medicine) http://aap-online.org/.
- Audrey Letourneau (graduate student); "Isabel Oberlé" award on the genetics of mental retardation, European Society Human Genetics 2011.
- Reza M Sailani (graduate student); pregraduate award on the genetics of heart defects in Down syndrome; European Society Human Genetics 2012.
- Periklis Makrythanasis (postdoctoral fellow); von Meissner award from the Univ Geneva Medical School 2012.
- Youssef Hibaoui (postdoctoral fellow) Alex F Muller prize of the Faculty of Medicine, Univ Geneva 2015.
- Honorary Doctorate Degree (PhD Honoris Causa), Democritus University of Thrace, Health Sciences School, Alexandroupolis, Greece 2016 (http://www.alexpoli.gr/dimokritio-panepistimio-timise-diethnous-fimis-ellina-epistimona-tou-exoterikou/).
- Honorary Citizen of Mani, Greece 2016 (Mayor P Andreakos) (www.gtp.gr/TDirectoryDetails.asp?ID=13615).
- Prize for Lifetime Outstanding Contributions to Medicine 2016; Univ. of Athens, Faculty of Medicine, Cardiology.
- Prize for Lifetime Outstanding Contributions to Down Syndrome 2017; Hellenic Down Syndrome Society.
- 2017- Honorary Member (elected; highest membership), Swiss Academy of Medical Sciences.
- Emmanuelle Ranza (postgraduate physician); Best MD Thesis award, Medical Faculty, University of Geneva 2018.
- Honorary Doctorate Degree (PhD Honoris Causa), National University of Athens, Greece 2018 (http://en.interel.uoa.gr/department-of-public-relations-protocol-and-cultural-events/events/great-hall.html).
- Muhammad Ansar (postdoctoral fellow) Young Investigator Prize 2019 from the Swiss Society of Medical Genetics.
- "Montserrat Trueta" Outstanding Career Award 2019 from the Catalan Down syndrome Foundation and the Trisomy 21 Research Society.
- **The 2019 "William Allan Award" from the American Society of Human Genetics** (http://www.ashg.org/press/201907-Allan-Award.shtml http://www.ashg.org/awards/index.shtml#collapseallan https://www.unige.ch/apropos/fr/prix-distinctions-nominations/stylianos-antonarakis-laureat-du-william-allan-award-2019/). (https://www.youtube.com/watch?v=Jc2jpoPcQVk&fbclid=IwAR34N3Mf6uWrVeLV2KQtEMY1vY7g96IJX4Kh7W3WgRpnsn3ZnXDYAeZdjfQ).
- **Gregg Semenza (post-doctorate fellow in the lab 1987-1990); Nobel Prize for Medicine 2019** for his work on the impact of Hypoxia in cells. The initial paper for the hypoxia factor was: Semenza GL, Nejfelt MK, Chi SM, Antonarakis SE. Hypoxia-inducible nuclear factors bind to an enhancer element located 3' to the human erythropoietin gene. Proc Natl Acad Sci USA. 1991 Jul 1;88(13):5680-4. PMID: 2062846
- Greek Journalists award for lifetime achievements in Medicine, September 2021, Athens, Greece.
- The Roscoe Prize 2022 (for outstanding human genetics teaching and mentoring), The Jackson Laboratory, Bar Harbor Maine

Editorial Board

- Molecular Biology and Medicine (1986-1990) published by Academic Press, London

- Genomics (1988-2008) published by Academic Press, San Diego
- Human Mutation (1992-) published by Wiley-Liss, New York
- Human Genetics (1994-2007) published by Springer, Berlin
- Journal of Medical Genetics (1995-) published by British Medical Journal, London
- Genome Research (1995-2005) published by Cold Spring Harbor, New York
- Cytogenetics & Cell Genetics (1997-) published by S Karger, Basel
- Journal of Human Genetics (1997-) published by Springer, Berlin
- European Journal of Pediatrics (2006-2013) published by Springer, Berlin
- Journal of Molecular Medicine (2007-) published by Springer, Berlin
- American Journal of Human Genetics (2008-2012), published by Cell Press, Boston
- Pathogenetics (2008-2010); Chief editor with A Ballabio, published by BMC, London
- Annual Reviews of Genomics and Human Genetics (2008-2013) published by Annual Reviews, Palo Alto, CA
- Genome Research (2009-), published by Cold Spring Harbor Laboratory Press, New York
- Senior Editor, eLife (2012-2016), published by the Howard Hughes Institute, Welcome Trust, Max Plank Institutes
- Associate Editor, Molecular Case Studies (2014-), published by Cold Spring Harbor Laboratory Press, New York
- Associate Editor, Nature Genomic Medicine (2015-), published by the Nature Publishing Group

Professional Societies

- American Society of Human Genetics (ASHG)
  Member, Board of Directors (2010-2014)
- Society for Pediatric Research (SPR)
- American Society for Clinical Investigation (ASCI), 1988
- Human Genome Organization (HUGO)
  Elected Member of the Council (2000-2004)
  Senior Editor, Chromosome 21
  Elected Member of the Council (2010-2014)
  President of HUGO, 2013-2017 (President Elect 2012-2013, President Emeritus 2017-)
  Organizer of the HUGO Annual meeting in Geneva 2014, Kuala Lumpur 2015, Houston 2016.
- European Society of Human Genetics (ESHG)
  Member, Scientific Program Committee 1997-2001
  Member, Board of Directors 1998-2004
  President Elect 2000-2001, Vice President 2002-2003
  President, European Society of Medical Genetics, 2001-2002
  Organizer of the Annual meeting in Geneva 1999
- Swiss Society of Medical Genetics (SSMG)
- Societe Academique de Genève
- Swiss Society of Experimental Biology (USGEB)
- European Molecular Biology Organization member (elected 2006) (www.embo.org/communities/members.html)
- Founding Fellow, American College of Medical Genetics (ACMG)
- Association of American Physicians (AAP), 2010
- Trisomy 21 Research Society, 2016
- **Member of the Senat, Swiss Academy of Medical Sciences, 2017**

Corporate Activities

Medigenome, Swiss Institute of Genomic Medicine
Established in 2018
Co-founder, CEO, and Chief Medical Officer since 2018
4 Rue Viollier
1207 Geneva
www.medigenome.ch

Other Appointments

- Elected member, Governing board, International School of Geneva (1998-2002)

Bibliography

Stylianos Emmanuel Antonarakis M.D., D.Sc.

Google scholar H-index 159 (updated Apr 2023); citations 129,000 (Mar 2023)
Boxed papers have more than 50 ISI citations each (N=297; updated Mar 2023)
http://www.ncbi.nlm.nih.gov/pubmed/?term=antonarakis+s
ORCID number https://orcid.org/0000-0001-8907-5823

001-OA **Antonarakis SE**, Boehm CD, Giardina PVJ, Kazazian HH Jr. Non-Random association of the polymorphic restriction sites in the beta-globin gene cluster. Proceedings of the National Academy of Sciences USA 79: 137-141, 1982 [PMID: 6275383] (abstract).

002-R **Antonarakis SE**, Phillips JA, Kazazian HH Jr. Genetic diseases: Diagnosis with restriction endonuclease analysis. Journal of Pediatrics 100: 845-857, 1982.

003-OA Orkin SH, Kazazian HH Jr., **Antonarakis SE**, Goff SC, Boehm CD, Sexton JP, Waber PG, Giardina PVJ. Linkage of beta-thalassemia mutations, and beta-globin gene polymorphisms with DNA polymorphisms in the human beta-globin gene cluster. Nature 296: 627-631, 1982 [PMID: 6280057].

004-BC Boehm CD, Phillips JA, **Antonarakis SE**, Kazazian HH Jr. Prenatal diagnosis by restriction analysis: Methodology and experience. In Scott RB (ed.) "Advances in the Pathophysiology diagnosis and treatment of sickle cell disease". AR Liss, New York, p.17-26, 1982.

005-BC Boehm CD, Phillips JA, **Antonarakis SE**, Kazazian HH Jr. Prenatal diagnosis of hemoglobinopathies by restriction analysis: Methodology and experience. In Willey A, Carter T, Kelly S, Porter I (eds.) "Clinical Genetics: Problems in diagnosis and counseling". Academic Press, New York. pp. 11-20, 1982.

006-OA **Antonarakis SE**, Orkin SH, Kazazian HH Jr, Goff SC, Boehm CD, Waber PG, Sexton JP, Ostrer H, Fairbanks VF, Chakravarti A. Evidence for multiple origins of the beta-E-globin gene in Southeast Asia. Proceedings of the National Academy of Sciences USA 79: 6608-6611, 1982.

007-BC Kazazian HH Jr, Chakravarti A, Orkin SH, **Antonarakis SE**. DNA polymorphisms in the human beta-globin gene cluster. In Nei M. and Kohen RK (eds) "Evolution of genes and proteins". Sinauer Assoc. Sunderland, MA. pp. 137-146, 1983.

008-OA Orkin SH, Kazazian HH Jr, **Antonarakis SE**, Ostrer H, Goff SC, Sexton JP. Abnormal RNA processing due to the exon mutation of the beta-E globin gene. Nature 300: 768-769, 1982 (DOI: 10.1038/300768a0) [PMID: 7177196] (abstract).

009-BC Kazazian HH Jr, **Antonarakis SE**, Cheng TC, Boehm CD, Waber PG. DNA polymorphisms in the beta-globin gene cluster: Use in discovery of mutations and prenatal diagnosis. In Caskey CT and White R. (eds) "Recombinant DNA applications to human diseases". Banbury Report #14, Cold Spring Harbor Lab. publ., pp. 29-39, 1983.

010-BC **Antonarakis SE**, Kazazian HH Jr, Orkin SH, Boehm CD, Waber PG. DNA polymorphisms in the beta-globin gene cluster: Use in clinical, molecular and evolutionary studies. In Bowman, JE (ed) "Distribution and evolution of hemoglobin and globin loci". Elsevier publ. New York, pp. 105-118, 1983.

011-OA Boehm CD, **Antonarakis SE**, Phillips JA, Stetten G, Kazazian HH Jr. Prenatal diagnosis using DNA polymorphisms: Report on 95 pregnancies at risk for sickle cell disease or beta-thalassemia. New England Journal of Medicine 308: 1054-1058, 1983.

012-BC Orkin SH, **Antonarakis SE**, Kazazian HH. The molecular genetics of human globin genes and thalassemias. Proceedings of the 19th Congress of Intern Society of Hematology, 1983.

013-BC Kazazian HH, **Antonarakis SE**, Cheng TC, Boehm CD, Waber PG. DNA polymorphisms in the beta-globin gene

cluster. A strategy for discovering new mutations. In Messer A, Porter IH (eds) "Recombinant DNA and Medical Genetics". Academic Press, NY, pp. 135-145, 1983.

014-OA Prochownik EV, **Antonarakis SE**, Bauer K, Rosenberg RD, Fearon ER, Orkin SH. Molecular heterogeneity of inherited antithrombin III deficiency. New England Journal of Medicine 308: 1549-1552, 1983.

015-BC Kazazian HH, **Antonarakis SE**, Cheng TC, Boehm CD, Waber PG. Use of haplotype analysis in the beta-globin gene cluster to discover beta-thalassemia mutations. In Stamatoyannopoulos G and Nienhuis A (eds) "Globin Gene Expression and Hematopoietic Differentiation". A. Liss, pp. 91-97, 1983.

016-OA Fearon ER, Kazazian HH, Jr., Waber PG, Lee JI, **Antonarakis SE**, Orkin SH, Vanin EF, Henthorn PS, Grosveld FG, Scott AF, Buchanan GR. The entire beta-globin gene cluster is deleted in a form of β-thalassemia. Blood 61: 1273-1278, 1983.

017-BC **Antonarakis SE**, Kazazian HH. Molecular basis of the thalassemia syndromes. In Schmidt, RM and Fairbanks VF, (eds) CRC Handbook in Hematology. CRC Press, Inc., pp. 249-261, 1986.

018-BC Orkin SH, **Antonarakis SE**, Kazazian HH. Polymorphism and molecular pathology of the human beta-globin gene. In Brown EB, (ed) Progress in Hematology, Vol. XIII, 49-73, 1983.

019-OA Kazazian HH, Jr., Orkin SH, Boehm CD, Sexton JP, **Antonarakis SE**. beta-thalassemia due to deletion of the nucleotide which is substituted in sickle cell anemia. American Journal of Human Genetics 35: 1028-1033, 1983.

020-OA Boyer SH, Dover GJ, Serjeant GR, **Antonarakis SE**, Embury SH, Smith KD, Margolet L, Noyes AN, Boyer ML, Bias WE. Production of F cells in sickle cell anemia. Regulation by a polymorphic genetic locus separate from the beta-globin gene cluster. Blood 64: 1053-1058, 1984.

021-OA **Antonarakis SE**, Phillips JA, Mallonee RL, Kazazian HH, Fearon ER, Waber PG, Kronenberg HM, Ullrich A, Meyers DA. beta-globin gene cluster is linked to parathyroid hormone (PTH) gene and lies between the insulin and PTH loci on chromosome 11p in man. Proceedings of the National Academy of Sciences USA 80: 6615-6619, 1983.

022-BC **Antonarakis SE**, Orkin SH, Kazazian HH. Thalassemia model: Genetic heterogeneity in inborn errors of metabolism. In Wapnir RA, (ed) "Congenital Metabolic Diseases". Marcel Dekker, Inc., NY, pp. 51-68, 1985.

023-OA Kazazian HH, Fairbanks VF, Waber PG, Boehm CD, Lee JI, **Antonarakis SE**. Hemoglobin E in Europeans: Further evidence for multiple origins of the beta-E-globin gene. American Journal of Human Genetics 36: 212-217, 1984.

024-OA **Antonarakis SE**, Valle D, Moser HW, Moser A, Qualman SJ, Zinkham WH. Phenotypic variability in siblings with Farber's disease. Journal of Pediatrics 104: 406-409, 1984.

025-OA Fearon ER, **Antonarakis SE**, Meyers DA, Levine MA. c-Ha-Ras-1 oncogene lies between beta globin and insulin loci on human chromosome 11p. American Journal of Human Genetics 36: 329-337, 1984.

026-OA **Antonarakis SE**, Boehm CD, Serjeant GR, Theisen CE, Dover GJ, Kazazian HH. Origin of the beta-S globin gene in Blacks: The contribution of recurrent mutation and/or gene conversion. Proceedings of the National Academy of Sciences USA 81: 853-856, 1984.

027-OA **Antonarakis SE**, Orkin SH, Cheng TC, Scott AF, Sexton JP, Trusko S, Charache S, Kazazian HH. β-thalassemia in American Blacks: Novel mutations in the TATA box and IVS-2 acceptor splice site. Proceedings of the National Academy of Sciences USA 81: 1154-1158, 1984.

028-OA Hutz MH, Michelson AM, **Antonarakis SE**, Orkin SH, Kazazian HH, Jr. Restriction site polymorphism in the phosphoglycerate kinase (PGK) gene on the X chromosome. Human Genetics 66: 217-219, 1984.

029-OA Cheng TC, Orkin SH, **Antonarakis SE**, Potter MJ, Sexton JP, Markham AF, Giardina PJV, Li A, Kazazian HH, Jr. β-thalassemia in Chinese: Use of in vivo RNA analysis and oligonucleotide hybridization in systematic characterization of molecular defects. Proceedings of the National Academy of Sciences USA 81: 2821-2825, 1984.

030-OA Kazazian HH, Orkin SH, **Antonarakis SE**, Sexton JP, Boehm CD, Goff SC, Waber PG. Molecular characterization of seven beta-thalassemia mutations in Asian Indians. EMBO Journal 3: 593-596, 1984.

031-BC **Antonarakis SE,** Kazazian, HH, Jr., Orkin, SH. DNA polymorphisms in the beta and beta-globin gene clusters and molecular etiology of the thalassemia syndromes in man. In "CRC Geographic distribution of hemoglobin variants" Winter, WP, ed. CRC Press, Inc., pp. 29-45, 1986.

032-OA Orkin SH, **Antonarakis SE,** Loucopoulos D. Abnormal processing of beta Knossos gene. Blood 64: 311-313, 1984 [PMID: 6733281] (abstract).

033-OA Chakravarti A, Buetow KH, **Antonarakis SE,** Waber PG, Boehm CD, Kazazian HH. Non-uniform recombination within the human beta-globin gene cluster. American Journal of Human Genetics 36: 1239-1258, 1984.

034-OA Orkin SH, **Antonarakis SE,** Kazazian HH. Base substitution at position -88 in a beta-thalassemia globin gene: Further evidence for the role of distal promoter element ACACCC. Journal of Biological Chemistry 259: 8679-8681, 1984.

035-OA Weinberg RS, **Antonarakis SE,** Kazazian HH, Dover GJ, Orkin SH, Lenes AL, Schofield JM, Alter BP: Fetal hemoglobin synthesis in erythroid cultures in hereditary persistence of fetal hemoglobin and βo-thalassemia. Blood 63: 1278-1284, 1984.

036-OA Boehm CD, Dowling CE, **Antonarakis SE,** Honig GR, Kazazian HH. Evidence supporting an unicentric origin of the beta-C globin gene in Blacks. American Journal of Human Genetics 37: 771-777, 1985.

037-OA Chaganti RSK, Jhanwar SC, **Antonarakis SE,** Hayward WS. Germ line chromosomal localization of genes in chromosome 11p linkage: Parathyroid hormone, beta-globin, c-Ha-Ras-1 and insulin. Somatic Cell Molecular Genetics 11: 197-202, 1985.

038-OA **Antonarakis SE,** Kazazian HH, Orkin SH: DNA polymorphisms and molecular pathology of the human globin gene clusters. Human Genetics 69: 1-14, 1985.

039-OA Noguchi CR, Dover GJ, Rodgers GP, Serjeant GR, **Antonarakis SE,** Anagnou NP, Higgs DR, Weatherall DJ, Schechter AN. beta-thalassemia changes erythrocyte heterogeneity in sickle cell disease. Journal of Clinical Investigation 75: 1632-1637, 1985. [PMID: 2581999] (abstract).

040-OA Orkin SH, Cheng TC, **Antonarakis SE,** Kazazian HH Jr. beta-thalassemia due to a mutation in the cleavage-polyadenylation signal: Evidence against strong transcription termination within 900 bp downstream from the human beta-globin gene. EMBO Journal 4: 453-456, 1985.

041-BC **Antonarakis SE.** Chromosome mapping using DNA probes. A case study (chromosome 11) and future possibilities. In: McKusick, VA, and Roderick, T. (eds) Medical and Experimental Mammalian Genetics, Alan Liss, New York, 1987, pp. 65-77.

042-OA **Antonarakis SE,** Kittur SD, Metaxotou C, Watkins PC, Patel AS. Analysis of DNA haplotypes suggests a genetic predisposition to trisomy 21 associated with DNA sequences on chromosome 21. Proceedings of the National Academy of Sciences USA 82: 3360-3364, 1985.

043-BC **Antonarakis SE,** Kittur SD, Metaxotou C, Bartsocas C, Kitsiou S, Watkins PC, Patel AS, Warren AC, Gusella JF, Groner Y, Chakravarti A, Meyers DA, Kazazian HH. Linkage map on chromosome 21q and the association of a DNA haplotype with a propensity to nondisjunction and trisomy 21. In: Smith GF (ed) Molecular structure of chromosome 21 and Down syndrome. Annals of the New York Academy of Sciences 450: 94-109, 1985.

044-BC Kazazian HH, **Antonarakis SE,** Wong C, Trusko SP, Stetten G, Oliver M, Potter MJ, Gusella JF, Watkins PC. Ring chromosome 21: characterization in DNA sequences of sites of breakage and reunion. In: Smith, GF (ed) Molecular structure of chromosome 21 and Down syndrome. Annals of the New York Academy of Sciences 450: 33-43, 1985.

045-OA Hoppener JWM, dePagter-Holthuizen P, Geurts van Kessell AHM, Jansen M, Kittur SD, **Antonarakis SE,** Lips CJM, Sussenbach JS. Human gene encoding insulin-like growth factor I is located on chromosome 12. Human Genetics 69: 157-160, 1985.

046-OA Kittur SD, Hoppener JWM, **Antonarakis SE,** Daniels JD, Meyers DA, Jansen M, Korneluk RG, Nelkin BD, Kazazian HH, Jr. Linkage map of the short arm of human chromosome 11: Location of the genes for catalase, calcitonin and insulin-like growth factor II. Proceedings of the National Academy of Sciences USA 82: 5064-5067, 1985.

047-OA Kittur SD, **Antonarakis SE**, Tanzi RE, Meyers DA, Chakravarti A, Groner Y, Phillips JA, Watkins PC, Gusella JF, Kazazian HH, Jr. Linkage map of three anonymous human DNA fragments and SOD-1 on chromosome 21. **EMBO Journal** 4: 2257-2260, 1985.

048-OA **Antonarakis SE**, Waber PG, Kittur SD, Patel AS, Kazazian HH, Jr., Mellis MA, Counts RB, Stamatoyannopoulos G, Bowie EJW, Fass DN, Pittman DD, Wozney JM, Toole JJ. Hemophilia A: Molecular defects and carrier detection by DNA analysis. **New England Journal of Medicine** 313: 842-848, 1985 [PMID: 2993888].

049-OA **Antonarakis SE**, Copeland KL, Carpenter RJ, Carta CA, Hoyer LW, Caskey CT, Toole JJ, Kazazian HH, Jr. Prenatal diagnosis of hemophilia A by factor VIII gene analysis. **Lancet** 1: 1407-1409, 1985.

050-OA Moore JW, Hyman S, **Antonarakis SE**, Mules EH, Thomas GH. Isolated familial aniridia associated with a translocation involving chromosomes 11 and 22 [t (11 22) (p13 q12.2)] **Human Genetics** 72: 297-302, 1986.

051-OA Ahn TG, **Antonarakis SE**, Kronenberg HM, Igarashi T, Levine MA. Familial isolated hypoparathyroidism: A molecular genetic analysis of 8 families with 23 affected persons. **Medicine** 65: 73-81, 1986.

052-RFLP Cohen T, Karathanasis SK, Kazazian HH, **Antonarakis SE**. DNA polymorphisms in the ApOAI-CIII-AIV gene cluster: Taq I and Ava I. **Nucleic Acids Research** 14: 1924, 1986.

053-RFLP Oettgen P, **Antonarakis SE**, Karathanasis SK. Pvu II polymorphic site in the ApOAI-CIII-AIV gene cluster. **Nucleic Acids Research** 14: 6571, 1986.

054-RFLP Oettgen P, **Antonarakis SE**, Karathanasis, SK. Bgl II polymorphic site in the ApOAI-CIII-AIV gene cluster. **Nucleic Acids Research** 14: 7138, 1986.

055-OA Janco RL, Phillips JA, Orlando P, Davies KE, Old J, **Antonarakis SE**. Carrier testing strategy in Hemophilia A. **Lancet** 1: 148-149, 1986.

056-OA Youssoufian H, Kazazian HH, Jr., Phillips DG, Aronis S, Tsiftis G, Brown VA, **Antonarakis SE**. Recurrent mutations in hemophilia A: Evidence for CpG dinucleotides as mutation hotspots. **Nature** 324: 380-382, 1986 [PMID: 3097553].

057-OA Wong C, **Antonarakis SE**, Goff SC, Orkin SH, Boehm CD, Kazazian HH. On the origin and spread of β-thalassemia: Recurrent observation of four mutations in different ethnic groups. **Proceedings of the National Academy of Sciences USA** 83: 6529-6532, 1986.

058-OA Atweh GF, Wong C, **Antonarakis SE**, Reed R, Zhu D, Ghosh PK, Maniatis T, Forget BG, Kazazian HH, Jr. A new mutation in IVS-1 of the human beta-globin gene causing beta-thalassemia due to abnormal splicing. **Blood** 70: 147-151, 1987.

059-OA Huang SZ, Wong C, **Antonarakis SE**, Rolien T, Lo WHY, Kazazian HH, Jr. The same TATA box beta thalassemia mutation in Chinese and U.S. Blacks. Another example of independent origins of mutation. **Human Genetics** 74: 162-164, 1986.

060-OA Karathanasis SK, Oettgen P, Hassad IA, **Antonarakis SE**. Structure, evolution and polymorphisms of the human apolipoprotein AIV gene. **Proceedings of the National Academy of Sciences USA** 83: 8457-8461, 1986.

061-OA Meyers DA, Beaty TH, Maestri NE, **Antonarakis SE**, Kazazian HH, Jr. Multipoint mapping studies of the beta-globin, insulin and c-Ras-1 on 11p. **American Journal of Human Genetics** 39: 539-542, 1986.

062-OA Meyers DA, Beaty TH, **Antonarakis SE**, Kazazian HH, Jr., White R. Genetic analysis workshop: The 11p data sets. **American Journal of Epidemiology** 1: 95-99, 1986 (suppl).

063-OA Meyers DA, Beaty TH, Maestri NE, Kittur SD, **Antonarakis SE**, Kazazian HH, Jr. Multipoint mapping studies of six loci on chromosome 11. **Human Heredity** 37: 94-101, 1987.

064-Let Chakravarti A, Buetow KH, **Antonarakis SE**, Waber PG, Boehm CD, Kazazian HH, Jr. Nonuniform recombination within the human beta-globin gene cluster. A reply to B.S. Weir and W.G. Hill. **American Journal of Human Genetics** 38: 779-781, 1986.

065-R **Antonarakis SE.** Hemophilia A. Persistence and gene mutational vulnerability. Hospital Practice 22: 93-102, 1987.

066-OA **Antonarakis SE,** Chakravarti A, Warren AC, Slaugenhaupt SA, Wong C, Halloran SL, Metaxotou C. Reduced recombination rate on chromosomes 21 that have undergone nondisjunction. Cold Spring Harbor Symposia on Quantitative Biology Molecular Biology of Homo Sapiens, 51: 185-190, 1986.

067-OA Kazazian HH, Jr., **Antonarakis SE,** Youssoufian H, Dowling CE, Phillips DG, Wong C, Boehm CD. Comparison of deficiency alleles of the beta-globin and factor VIII:C genes: New lessons from a giant gene. Cold Spring Harbor Symposia on Quantitative Biology, Molecular Biology of Homo Sapiens, 51: 371-379, 1986.

068-OA Youssoufian H, **Antonarakis SE,** Phillips, DG, Aronis S, Tsiftis G, Kazazian HH, Jr. The molecular genetics of hemophilia A: Five different partial deletions of factor VIII:C gene. Proceedings of the National Academy of Sciences USA 84: 3772-3776, 1987.

069-OA Dover GJ, Chang VT, Boyer SH, Serjeant BG, **Antonarakis SE,** Higgs DR. The cellular basis for different fetal hemoglobin levels among sickle cell individuals with two, three and four alpha globin genes. Blood 69: 341-344, 1987.

070-OA Wong C, **Antonarakis SE,** Goff SC, Orkin SH, Forget BG, Nathan DC, Giardina PJV, Kazazian HH, Jr. beta-thalassemia due to two novel nucleotide substitutions in consensus acceptor splice sequences of the beta-globin gene. Blood 73: 914-918, 1989.

071-BC Kazazian HH, Jr. and **Antonarakis SE:** The varieties of mutation. In: Childs B, Holtzman HA, Kazazian HH, Jr., Valle D (eds) "Molecular Genetics in Medicine", Progress in Medical Genetics, Vol. 7. Elsevier: New York, 1987, pp. 43-67.

072-BC **Antonarakis SE.** The molecular genetics of hemophilia A and B in man. In: Harris H and Hirschhorn K (eds) Advances in Human Genetics 17: 27-59, 1988.

073-OA **Antonarakis SE,** Oettgen P, Chakravarti A, Halloran SL, Hudson RR, Feisee L, Karathanasis SK. DNA polymorphism haplotypes of the human apolipoprotein ApOA1-ApoC3-ApOA4 gene cluster. Human Genetics 80: 265-273, 1988.

074-OA Warren AC, Chakravarti A, Wong C, Slaugenhaupt SA, Halloran SL, Watkins PC, Metaxotou C, **Antonarakis SE.** Evidence for reduced recombination on the nondisjoined chromosomes 21 in Down syndrome. Science 237: 652-654, 1987 [PMID: 2955519].

075-BC Warren AC, Chakravarti A, Wong C, Slaugenhaupt SA, Halloran SL, Watkins PC, Metaxotou C, **Antonarakis SE.** Down syndrome: Evidence for reduced recombination on the chromosomes 21 that underwent nondisjunction. In Davies, P. (ed) "Neurochemistry of Aging". Bambury Report. Cold Spring Harbor, N.Y., 1988, pp. 373-375.

076-R **Antonarakis SE,** Youssoufian H, Kazazian, HH, Jr. Molecular genetics of hemophilia A. Molecular Biology in Medicine 4: 81-94, 1987.

077-BC Kazazian HH, Jr., Dowling CE, Waber PG, Huang S-z, Lo, WHY, Li A, Tam JWO, Kang J, **Antonarakis SE.** Molecular characterization of beta-thalassemia major and beta-thalassemia intermedia in China and Southeast Asia. In Stamatoyannopoulos G and Nienhuis A (eds) "Developmental Control of Globin Gene Expression". Alan Liss, Inc., New York, 1987, pp. 401-412.

078-OA Long JC, Chakravarti A, Boehm CD, **Antonarakis SE,** Kazazian HH, Jr. A phylogeny of human beta-globin haplotypes and its implications for recent human evolution. American Journal of Physical Anthropology 81: 113-130, 1990.

079-OA Youssoufian H, **Antonarakis SE,** Bell W, Griffin AM, Kazazian HH, Jr.: Nonsense and missense mutations in hemophilia A: Estimate of the relative mutation rate at CG dinucleotides. American Journal of Human Genetics 42: 718-725, 1988.

080-OA Anagnou NP, **Antonarakis SE,** O'Brien SJ, Modi WS, Nienhuis AW. Chromosomal localization and racial distribution of the polymorphic human dehydrofolate reductase pseudogene (DHFRPs1). American Journal of Human Genetics 42: 345-352, 1988.

081-RFLP Semenza GL, Ladias JA, **Antonarakis SE.** An Xba I polymorphism 3' to the human erythropoietin gene Nucleic Acids Research 15: 6768, 1987.

082-RFLP Anagnou NP, **Antonarakis SE**, O'Brien SJ, Nienhuis AW. A novel form of human polymorphism involving the hDHR-Ps1 pseudogene identifies 3 RFLPs. Nucleic Acids Research 15: 5501, 1987.

083-OA Youssoufian H, Kazazian HH, Jr., Patel A, Aronis S, Tsiftis G, Hoyer LW, **Antonarakis SE**: Mild hemophilia A associated with a cryptic donor splice site mutation in intron 4 of the factor VIII gene. Genomics 2: 32-36, 1988.

084-OA Wong C, Kazazian HH, Jr., Stetten G, van Keuren ML, Earnshaw WC, **Antonarakis SE**. Molecular mechanism in the formation of a ring chromosome 21. Proceedings of the National Academy of Sciences USA 86: 1914-1918, 1989.

085-RFLP Youssoufian H, Phillips DG, Kazazian HH, Jr., **Antonarakis SE**. MspI polymorphism in the 3' flanking region of the human factor VIII gene. Nucleic Acids Research 15: 6312, 1987.

086-RFLP Heinzmann C, Ladias J, **Antonarakis SE**, Kirshgessner T, Schotz M, Lusis AJ. RFLP for the human lipoprotein lipase gene Hind III. Nucl. Acids Res. 15: 6763, 1987.

087-OA Warren AC, Robakis HK, Ramakrishna N, Koo EH, Ross CA, Folstein MF, Price DL, **Antonarakis SE**. beta-amyloid gene is not present in three copies in autopsy-validated Alzheimer's disease. Genomics 1: 307-312, 1987.

088-OA Youssoufian H, Wong C, Aronis S, Platokoukis H, Kazazian HH, Jr., **Antonarakis SE**. Moderately severe hemophilia A resulting from Glu-Gly substitution in exon 7 of the factor VIII gene. American Journal of Human Genetics 42: 867-871, 1988.

089-OA Kazazian HH, Jr., Youssoufian H, Scott AF, Phillips D, **Antonarakis SE**. A novel mechanism of mutation in man: Hemophilia A due to de novo insertion of L1 sequences. Nature 332: 164-166, 1988.

090-OA Ma Y, Ladias JAA, Butler R, Schumaker VN, **Antonarakis SE**, Lusis AJ, Heinzmann C, Kwiterovich PO. Apolipoprotein B gene haplotypes: Association between Ag and DNA polymorphism. Arteriosclerosis 8: 521-524, 1988.

091-OA Youssoufian H, Kasper CK, Phillips DG, Kazazian HH, Jr., **Antonarakis SE**. Restriction endonuclease mapping of six novel deletions of the factor VIII gene in hemophilia A. Human Genetics 80: 143-148, 1988.

092-OA Qualman SJ, Moser HW, Valle D, Moser AE, **Antonarakis SE**, Boitnott JK, Zinkham, WH. Farber disease: Pathologic diagnosis in sibs with phenotypic variability. American Journal of Medical Genetics (suppl) 3: 233-241, 1987.

093-OA **Antonarakis SE**, Kang J, Lam VMS, Tam JWO, Li AMC. Molecular characterization of beta-globin gene mutations in patients with beta thalassemia intermedia in South China. British Journal of Hematology 70: 357-361, 1988.

094-BC Kazazian HH, Jr., Wong C, Youssoufian H, Phillips DG, Scott AF, **Antonarakis SE**. Identification of mutations producing hemophilia A. In: "UCLA Symposia Gene Transfer in Animals". Alan Liss, NY, 87: 293-307, 1988.

095-OA Cutting GR, **Antonarakis SE**, Youssoufian H, Kazazian, HH, Jr. Accuracy and limitations of pulse field gel electrophoresis in sizing partial deletions of the factor VIII gene. Molecular Biology Medicine 5: 173-184, 1988.

096-R **Antonarakis SE**, Kazazian HH, Jr. The molecular basis of hemophilia A in man. Trends in Genetics 4: 233-237, 1988

097-OA Cutting GR, Kazazian HH, Jr., **Antonarakis SE**, Killen PD, Yamada Y, Francomano, CA. Macrorestriction mapping of COL4A1 and COL4A2 collagen genes on human chromosome 13q34. Genomics 3: 256-263, 1988.

098-RFLP Warren AC, Groner Y, **Antonarakis SE**. A DNA polymorphism with KpnI of the human liver-type phosphofructokinase (PFKL) gene. Nucleic Acids Research 16: 9060, 1988.

099-RFLP Heinzmann C, Ladias J, **Antonarakis SE**, Diep A, Schotz M, Lusis AJ. Two polymorphisms for the human hepatic lipase (HL) gene. Nucleic Acids Research 16: 4739, 1988.

100-OA Inaba H, Fujimaki M, Kazazian HH, Jr., **Antonarakis SE**. Mild hemophilia A resulting from Arg to Leu substitution in exon 26 of the factor VIII gene. Human Genetics 81: 335-338, 1989.

101-R **Antonarakis SE**. Diagnosis of genetic disorders at the DNA level. New England Journal of Medicine 320: 153-163, 1989.

102-OA Woods-Samuels P, Wong C, Mathias SL, Scott AF, Kazazian HH, Jr., **Antonarakis SE.** Characterization of a non-deleterious L1 insertion in an intron of the human factor VIII gene and evidence for open reading frame in functional L1 elements. Genomics 4: 290-296, 1989.

103-BC **Antonarakis SE,** Kazazian HH. Hemophilia A in man: Molecular defects in the factor VIII gene. In: Hoffbrand AV, ed. "Recent Advances in Hematology". Churchill Livingston, London, pp. 243-250, 1988.

104-OA Semenza GL, Traystman MD, Gearhart JD, **Antonarakis SE.** Polycythemia in transgenic mice expressing the human erythropoietin gene. Proceedings of the National Academy of Sciences USA 86: 2301-2305, 1989.

105-OA Cutting GR, **Antonarakis SE,** Buetow KH, Kasch LM, Rosenstein BI, Kazazian HH, Jr. Analysis of DNA polymorphism haplotypes linked to the cystic fibrosis locus in North American Blacks and Caucasian families support the existence of multiple mutations of the cystic fibrosis gene. American Journal of Human Genetics 44: 307-318, 1989.

106-OA Warren AC, Slaugenhaupt SA, Lewis JG, Chakravarti A, **Antonarakis SE.** A genetic linkage map of 17 markers on human chromosome 21. Genomics 4: 579-591, 1989.

107-OA Huang S, Waber PG, Dowling CE, Wong C, **Antonarakis SE,** Cai RL, Wang MQ, Lo WH, Kazazian HH., Jr. Beta-thalassemia in China: a systematic molecular characterization of beta-thalassemia mutations. Hemoglobin 12: 621-628, 1988.

108-OA Zhu D, **Antonarakis SE,** Schmeckpeper BJ, Diergaarde PJ, Maumenee IH. Microdeletion in the X chromosome and prenatal diagnosis in a family with Norrie disease. American Journal of Medical Genetics 33: 485-488, 1989.

109-BC Francomano CA, **Antonarakis SE.** Approaches to the diagnosis of renal genetic disorders using DNA analysis. In: Spitzer A and Avner ED, Ed. "Inheritance of kidney and urinary tract diseases" Kluwer Academic Publishers, Boston, pp. 53-63, 1990.

110-OA Stetten G, Tuck-Muller CM, Blakemore KJ, Wong C, Kazazian HH, **Antonarakis SE.** Evidence for involvement of a Robertsonian translocation 13 chromosome in the formation of a ring chromosome 13. Molecular Biology Medicine 7: 479-484, 1990.

111-BC **Antonarakis SE,** Warren AC, McCormick MK, Lewis JG, Hieter PA, Chakravarti A. Molecular Genetics of chromosome 21 and the region responsible for Down Syndrome. In Epstein CJ & Hassold TJ Eds "Molecular genetic & cytogenetic studies of nondisjunction". A Liss, New York, pp. 29-45, 1989.

112-BC Chakravarti A, Majumder PP, Slaugenhaupt S, Deka R, Warren AC, Surti U, Ferrell RE, **Antonarakis SE.** Gene centromere mapping and the study of nondisjunction in autosomal trisomies and ovarian teratomas. In Epstein CJ & Hassold TJ Eds "Molecular genetic & cytogenetic studies of nondisjunction". A Liss, New York pp. 45-81, 1989.

113-BC Roulston D, **Antonarakis SE,** Lewis JG, Cohen MM, Schwartz S. Cytological and molecular studies of nucleolar organizing region variants and recombination in trisomy 21. In Epstein CJ & Hassold TJ Eds "Molecular genetic & cytogenetic studies of nondisjunction". A Liss, New York pp. 81-101, 1989.

114-OA Arai M, Inaba H, Higuchi M, **Antonarakis SE,** Kazazian HH, Jr., Fuzimaki M, Hoyer LW. Direct characterization of factor VIII in plasma: detection of a mutation altering a thrombin cleavage site (Arg 372-> His) Proceedings of the National Academy of Sciences USA 86: 4277-4281, 1989.

115-R **Antonarakis SE** Recombinant DNA technology in the diagnosis of human genetic disorders. Clinical Chemistry 35: B4-6 (suppl.), 1989.

116-OA McCormick MK, Schinzel A, Petersen MB, Stetten G, Driscoll DJ, Cantu ES, Tranebjaerg L. Mikkelsen M, Watkins PC, **Antonarakis SE.** Molecular genetic approach to the characterization of the "Down syndrome" region on chromosome 21. Genomics 5:325-331, 1989.

117-OA Ladias JA, Kwiterovich PO, Smith HH, Karathanasis SK, **Antonarakis SE.** Apolipoprotein A-I Baltimore (Arg10Leu); a new ApoAI variant in a family with hypoAlphalipoproteinemia. Human Genetics 84: 439-445, 1990.

118-OA Ladias JAA, Kwiterovich PO, Smith HH, Miller M, Bachorik PS, Forte T, Lusis AJ, **Antonarakis SE.** Apolipoprotein B100-Hopkins (Arg4019Trp); a new APOB variant in a family with premature atherosclerosis and hyperapobetalipoproteinemia. JAMA 262: 1980-1988, 1989.

119-OA Arai M, Higuchi M, **Antonarakis SE**, Kazazian HH, Jr., Phillips JA, Janco RL, Hoyer LW. Characterization of a thrombin cleavage site mutation (Arg1689Cys) in the factor VIII gene of two unrelated patients with CRM positive Hemophilia A. **Blood** 75: 384-389, 1990.

120-OA Glaser T, Driscoll DJ, **Antonarakis SE**, Valle D, Housman D. A highly polymorphic locus cloned from the breakpoint of a chromosome 11p13 deletion associated with the WAGR syndrome. **Genomics** 5: 880-893, 1989.

121-BC **Antonarakis SE**, Kazazian HH, Jr. The molecular basis of hemophilia A (factor VIII deficiency) in man; progress report from The Johns Hopkins University Hemophilia project. In: Kasper C, Ed. "Recent advances in hemophilia care". A. Liss, New York, pp. 1-11, 1990.

122-OA Williams CA, Frias JL, McCormick MK, **Antonarakis SE**, Cantu ES. Clinical cytogenetic and molecular evaluation of a patient with partial trisomy 21 (21q11-q22) lacking the classical Down Syndrome phenotype. **American Journal of Medical Genetics** (supplement) 7: 110-114, 1990.

123-OA Hofman KJ, **Antonarakis SE**, Missiou-Tsangaraki S, Boehm CD, Valle D. Phenylketonuria in the Greek population: haplotype analysis of the phenylalanine hydroxylase gene and identification of a PKU mutation. **Molecular Biology Medicine** 6: 245-250, 1989.

124-OA Inaba H, Fujimaki M, Kazazian HH, Jr., **Antonarakis SE**. Msp I polymorphic site in intron 22 of the factor VIII gene in Japanese population. **Human Genetics** 84: 214-215, 1990.

125-OA Higuchi M, Wong C, Kochhan L, Olek K, Aronis S, Kasper C, Kazazian HH. Jr., **Antonarakis SE**. Characterization of mutations in the factor VIII gene by direct sequencing of amplified genomic DNA. **Genomics** 6: 65-71, 1990.

126-RFLPPetersen MB, Horisberger MA, Warren AC, **Antonarakis SE**. Two Pst I DNA polymorphisms adjacent to the human gene for the interferon-induced p78 protein (MX1 gene). **Nucleic Acids Research** 17: 7546, 1989.

127-OA McCormick MK, Shero JR, Connelly CJ, **Antonarakis SE**, Hieter PA. Methods for cloning large DNA segments in artificial chromosomes in S Cerevisiae. **Technique** 2: 65-71, 1990.

128-OA Traystman MD, Higuchi M, **Antonarakis SE**, Kazazian HH, Jr. Use of denaturing gradient gel electrophoresis to detect point mutations in the factor VIII gene. **Genomics** 6: 293-301, 1990.

129-OA Zhu D, Alcorn DM, **Antonarakis SE**, Levin LS, Huang PC, Mitchell TN, Warren AC, Maumenee IH. Assignment of the Nance-Horan syndrome to the distal short arm of the X chromosome. **Human Genetics** 86: 54-58, 1990.

130-OA McCormick MK, Shero J, Chung MC, Kan YW, Hieter PA, **Antonarakis SE**. Construction of human chromosome 21 specific yeast artificial chromosomes. **Proceedings of the National Academy of Sciences USA** 86: 9991-9995, 1989.

131-OA Petersen MB, Tranebjaerg L, McCormick MK, Michelsen N, Mikkelsen M, **Antonarakis SE**. Clinical, cytogenetic and molecular genetic characterization of two unrelated patients with partial trisomy 21. **American Journal of Medical Genetics** (supplement) 7: 104-109, 1990.

132-OA Semenza GL, Dureza RC, Traystman MD, Gearhart JD, **Antonarakis SE**. Human Erythropoietin gene expression in transgenic mice: multiple transcription initiation sites and cis-acting regulatory elements. **Molecular Cell Biology** 10: 930-938, 1990.

133- RFLP Petersen MB, Jaworski CJ, Lewis JG, **Antonarakis SE**. Pvu II and Xho I/Eco RV polymorphisms adjacent to the human crystallin (CRYA1) gene on human chromosome 21. **Nucleic Acids Research** 18: 4300, 1990.

134-R **Antonarakis SE**. Trisomy 21; Chromosome 21 and Down Syndrome. **Current Opinion Pediatrics** 1: 445-447, 1989.

135-OA Francomano CA, Cutting GR, McCormick MK, Chu ML, Timpl R, Hong HK, **Antonarakis SE**. The COL6A1 and COL6A2 genes exist as a gene cluster and detect highly informative DNA polymorphisms in the telomeric region of human chromosome 21. **Human Genetics** 87: 162-166, 1991.

136-OA Warren AC, Bowcock AM, Farrer LA, **Antonarakis SE**. A DNA polymorphism of alphoid sequences specific for the centromeric region of chromosome 13. **Genomics** 7: 110-114, 1990.

137-OA Economou EP, Bergen A, Warren AC, **Antonarakis SE.** The poly(A) tail of Alu repetitive elements is polymorphic in the human genome (AluVpA). Proceedings of the National Academy of Sciences USA 87: 2951-2954, 1990.

138-OA Petersen MB, Economou EP, Slaugenhaupt SA, Chakravarti A, **Antonarakis SE.** Linkage analysis of the human HMG14 gene on chromosome 21 using a GT dinucleotide repeat as polymorphic marker. Genomics 7: 136-138, 1990.

139-OA Cutting GR, McGinniss MJ, Kasch LM, Tsipouras P, **Antonarakis SE.** Physical mapping by PFGE localizes the COL3A1 and COL5A2 genes to a 35 kb region on chromosome 2. Genomics 8: 407-410, 1990.

140-OA Cutting GR, Kasch LM, Rosenstein BJ, Zielensky J, Tsui LC, **Antonarakis SE,** Kazazian HH, Jr. A cluster of Cystic Fibrosis mutations in the first nucleotide binding fold domain of the CFTR gene. Nature 346: 366-369, 1990.

141-BC Kazazian HH, Jr., Dowling CE, Boehm CD, Warren TC, Economou EP, Katz J, **Antonarakis SE.** Gene defects in beta-thalassemia and their prenatal diagnosis. Annals of the New York Academy of Sciences 612: 1-14, 1990.

142-OA Lewis JG, Weber JL, Petersen MB, Slaugenhaupt SA, Kwitek A, May PE, Warren AC, Chakravarti A, **Antonarakis SE.** Linkage mapping of DNA marker D21S156 to human chromosome 21 using a polymorphic GT dinucleotide repeat. Genomics 8: 400-402, 1990.

143-OA Cutting GR, Kasch LM, Tsui LC, Rosenstein BJ, Kazazian HH, Jr., **Antonarakis SE.** Two Cystic fibrosis patients with mild pulmonary disease and nonsense mutations in each CFTR gene. New England Journal of Medicine 323: 1685-1689, 1990.

144-R **Antonarakis SE.** The mapping and sequencing of the human genome. Southern Medical Journal 83: 876-878, 1990.

145-OA Jabs EW, Warren AC, Taylor EW, Colyer CR, Meyers DA, **Antonarakis SE.** Alphoid DNA polymorphisms for chromosome 21 can be distinguished from those of chromosome 13 using probes homologous to both. Genomics 9: 141-146, 1991.

146-BC **Antonarakis SE.** Amino acid substitutions in the factor VIII coagulation protein that cause hemophilia A in man. In Albertini A. et al. eds. "Biotechnology of human plasma proteins". S. Karger, Basel, pp 73-77, 1991.

147-OA McCormick MK, **Antonarakis SE,** Hieter PA. Yeast artificial chromosome: Cloning of DNA embedded in an agarose matrix. Genetics Analytical Techniques & Applications 7: 114-118, 1990.

148-OA **Antonarakis SE,** Adelsberger PA, Petersen MB, Binkert F, Schinzel AA. Analysis of DNA polymorphisms suggests that most de novo dup(21q) chromosomes in patients with Down syndrome are isochromosomes and not translocations. American Journal of Human Genetics 47: 968-972, 1990.

149-OA Economou EP, **Antonarakis SE,** Kazazian HH, Jr., Serjeant GR, Dover GJ. The variation in HbF production among normal and sickle cell adults is not related to nucleotide substitutions in the gamma promoter regions. **Blood** 77: 174-177, 1991.

150-OA Heinzmann C, Kirchgessner T, Kwiterovich PO, Ladias JA, Derby C, **Antonarakis SE,** Lusis AJ. DNA polymorphism haplotypes of the human lipoprotein lipase gene: Possible association with high density lipoprotein. Human Genetics 86: 578-584, 1991.

151-OA Traystman M, **Antonarakis SE,** Kazazian HH, Jr. Procedure for using denaturing gradient gel electrophoresis for mutation analysis of polymerase chain reaction amplification products. Technique 2: 206-212, 1990.

152-OA Petersen MB, Slaugenhaupt SA, Lewis JG, Warren AC, Chakravarti A, **Antonarakis SE.** A genetic linkage map of 27 markers on human chromosome 21. Genomics 9: 407-419, 1991.

153-Book McKusick VA with the assistance of Francomano CA, and **Antonarakis SE.** "Mendelian Inheritance in Man. Catalogs of autosomal dominant, autosomal recessive and X-linked phenotypes". 9th edition. The Johns Hopkins University Press, Baltimore and London, 1990.

154-OA Petersen MB, Schinzel A, Binkert F, Tranebjaerg L, Mikkelsen M, Collins FA, Economou EP, **Antonarakis SE.** Use of short sequence polymorphisms after PCR amplification to detect the parental origin of the additional chromosome 21 in

Down syndrome. American Journal of Human Genetics 48: 65-71, 1991.

155-OA Lorda-Sanchez I, Petersen MB, Binkert F, Maechler M, Schmid W, Adelsberger PA, **Antonarakis SE**, Schinzel A. A 48,XXY,+21 Down syndrome patient with additional paternal X and maternal 21. Human Genetics 87: 54-56, 1991.

156-OA Petersen MB, Weber JL, Slaugenhaupt SA, Kwitek AE, McInnis MG, Chakravarti A, **Antonarakis SE**. Linkage mapping of D21S171 to the distal long arm of human chromosome 21 using a polymorphic (AC)n dinucleotide repeat. Human Genetics 87: 401-404, 1991.

157-OA Collins FA, Murphy DL, Reiss A, Sims KB, Lewis JG, Freund L, Zhu D, Maumenee IH, **Antonarakis SE**. Clinical, biochemical and neuropsychiatric evaluation of a patient with a contiguous gene syndrome due to a microdeletion Xp11.3 including the Norrie disease locus and monoamine oxidase (MAOA and MAOB) genes. American Journal of Human Genetics 42: 127-134, 1992.

158-OA **Antonarakis SE**, and the Down Syndrome Collaborative Group (Lewis JG, Adelsberger PA, Petersen MB, Schinzel AA, Binkert F, Schmid W, Pangalos C, Raoul O, Chakravarti A, Hafez M, Cohen MM, Roulston D, Schwartz S, Mikkelsen M, Tranebjaerg L, Greenberg F, HOAr DI, Rudd NL, Warren AC, Metaxotou C, Bartsocas C. Parental origin of the extra chromosome in trisomy 21 using DNA polymorphism analysis. New England Journal of Medicine 324: 872-876, 1991 [PMID 1825697].

159-OA Cutting GR, Lu L, O'Hara B, Kasch LM, Donovan D, Shimada S, **Antonarakis SE**, Guggino WB, Uhl GR, Kazazian HH, Jr. Cloning of the GABA Rho1 cDNA: A novel GABA subunit highly expressed in the retina. Proceedings of the National Academy of Sciences USA 88: 2673-2677, 1991.

160-OA Shero J, McCormick MK, **Antonarakis SE**, Hieter P. Yeast artificial chromosome (YAC) vectors for efficient clone manipulation and mapping. Genomics 10: 505-508, 1991.

161-BC Hieter PA, Connelly C, Shero J, McCormick MK, **Antonarakis SE**, Pavan W, Reeves R. Yeast artificial chromosomes: Promises kept and pending. In: Davies K and Tilghman S, ed. "Genome Analysis". Cold Spring Harbor, N.Y., Vol.1: 83-120, 1990.

162-OA Woods-Samuels P, Kazazian HH, Jr., **Antonarakis SE**. Nonhomologous recombination in the human genome: Deletions in the human factor VIII gene. Genomics 10: 94-101, 1991.

163-R Cutting GR, **Antonarakis SE**. Prenatal diagnosis and carrier detection by DNA analysis. Pediatrics in Review 13: 138-143, 1992.

164-OA Higuchi M, **Antonarakis SE**, Kasch L, Oldenburg J, Economou Petersen E, Olek K, Inaba H, Kazazian HH, Jr.: Towards complete characterization of mild-to-moderate hemophilia A: Detection of the molecular defect in 25 of 29 patients by denaturing gradient gel electrophoresis. Proceedings of the National Academy of Sciences USA 88: 8307-8311, 1991.

165-OA Petersen MB, Adelsberger PA, Schinzel AA, Binkert F, Hinkel GK, **Antonarakis SE**. Down syndrome due to de novo Robertsonian translocation (t14q;21q): DNA polymorphism analysis suggests that the origin of the extra 21q is maternal. American Journal of Human Genetics 49: 529-536, 1991.

166-OA Semenza GL, Nejfelt MK, Chi SM, **Antonarakis SE**. Hypoxia inducible nuclear factors bind 3' to the human erythropoietin gene. Proceedings of the National Academy of Sciences USA 88: 5680-5684, 1991 [PMID: 2062846] (abstract).
This paper is part of the work awarded by the Nobel Prize 2019 to Gregg Semenza

167-OA Zhu D, Economou EP, **Antonarakis SE**, Maumenee IH. Mitochondrial DNA mutation and heteroplasmy in type I Leber hereditary optic atrophy. American Journal of Medical Genetics 42: 173-179, 1992.

168-OA Higuchi M, Kazazian HH, Jr., Kasch L, Warren TC, McGinniss MJ, Phillips JA, III, Kasper C, Janco R, **Antonarakis SE**. Molecular characterization of severe hemophilia A suggests that about half the mutations are not within the coding region and splice junctions of the factor VIII gene. Proceedings of the National Academy of Sciences USA 88: 7405-7409, 1991.

169-OA Epstein CJ, Korenberg Jr., Anneren G, **Antonarakis SE**, Ayme S, Courchesne E, Epstein LB, Fowler A, Groner Y,

Huret JL, Kemper TL, Lott IT, Lubin BH, Magenis E, Opitz JM, Patterson D, Priest JH, Pueschel SM, Rapoport SI, Sinet PM, Tanzi RE, dela Cruz F. Protocols to establish genotype-phenotype correlations in Down syndrome. **American Journal of Human Genetics** 49: 207-235, 1991.

170-OA Economou EP, **Antonarakis SE**, Dowling CC, Ibarra B, DelaMora E, Kazazian HH,Jr. Molecular heterogeneity of beta-thalassemia in Mestizo Indians. **Genomics** 11: 474, 1991.

171-BC **Antonarakis SE**, Kazazian HH,Jr. Prenatal diagnosis and carrier detection of hemophilia A and B by DNA analysis in Brock DJ, Rodeck CH, Ferguson-Smith MA eds. "Prenatal Diagnosis and Screening" Churchill Livingstone publishers, Edinburgh pp 477-482, 1992.

172-OA Reddy KS, Petersen MB, **Antonarakis SE**, Blakemore KJ. The vanishing twin: An explanation for discordance between chorionic villus karyotype and fetal phenotype. **Prenatal Diagnosis** 11: 679-684, 1991.

173-OA McInnis MG, Lutfalla G, Slaugenhaupt SA, Petersen MB, Uze G, Chakravarti A, **Antonarakis SE**. Linkage mapping of highly informative polymorphisms of the human interferon-alpha receptor gene on chromosome 21. **Genomics** 11: 573-576, 1991.

174-OA Semenza GL, Koury ST, Nejfelt MK, Gearhart JD, **Antonarakis SE**. Cell type specific and hypoxic-inducible expression of the human erythropoietin gene in transgenic mice. **Proceedings of the National Academy of Sciences USA** 88: 8725-8729, 1991.

175-OA **Antonarakis SE**, Petersen MB, McInnis MG, Adelsberger PA, Schinzel AA, Binkert F; Pangalos C, Raoul O, Slaugenhaupt SA, Hafez M, Cohen MM, Roulston D, Schwartz S, Mikkelsen M, Tranebjaerg L, Greenberg F, HOAr DI, Rudd NL, Warren AC, Metaxotou C, Bartsocas C, Chakravarti A. The meiotic stage of nondisjunction in trisomy 21; Determination using DNA polymorphisms. **American Journal of Human Genetics** 50: 544-550, 1992.

176-R Tuddenham EGD, Cooper DN, Gitschier J, Higuchi M, Hoyer LW, Yoshioka A, Peake IR, Schwaab R, Olek K, Kazazian HH, Lavergne JM, **Antonarakis SE**. Hemophilia A: Database of nucleotide substitutions, deletions, insertions, and rearrangements of the factor VIII gene. **Nucleic Acids Research** 18: 4821-4833, 1991.

177-OA McGinniss MJ, Kazazian HH Jr, Stetten G, Petersen MB, Boman H, Engel E, Greenberg F, Hertz JM, Johnson A, Laca Z, Mikkelsen M, Patil SR, Schinzel AA, Tranebjaerg L, **Antonarakis SE**. Mechanisms of ring chromosome formation in 11 cases of human ring chromosome 21. **American Journal of Human Genetics** 50: 15-28, 1992.

178-OA Schinzel AA, Adelsberger PA, Binkert F, Basaran S, **Antonarakis SE**. No evidence for a paternal interchromosomal effect from analysis of the origin of nondisjunction in Down Syndrome patients with concomitant familial chromosome rearrangements. **American Journal of Human Genetics** 50: 288-293, 1992.

179-OA Warren AC, **Antonarakis SE**, Chakravarti A. Chromosome 21 genetic linkage data set based on CEPH pedigrees. In MacCluer JN et al eds. Genetic Analysis Workshop 7 **Cytogenet and Cell Genetics** (suppl) 59: 86-87, 1992.

180-RFLP McGinniss MJ, Horisberger MA, Kazazian HH Jr, **Antonarakis SE**. An MspI polymorphism at the MX1 locus in 21q22.3. **Nucleic Acids Research** 19: 5451, 1991.

181-OA Economou EP, Kazazian HH Jr., **Antonarakis SE**. Detection of mutations in the factor VIII gene using single stranded conformational analysis (SSCP). **Genomics** 13: 909-911, 1992.

182-OA Aly AM, Higuchi M, Kasper CK, Kazazian HH Jr., **Antonarakis SE**, Hoyer LW. Hemophilia A due to mutations that create new N-glycosylation sites. **Proceedings of the National Academy of Sciences USA** 89: 4933-4937, 1992.

183-OA Coresh J, Beaty TH, Kwiterovich PO, **Antonarakis SE**. Pedigree and sib-pair linkage analysis suggest the apolipoprotein B gene is not the major gene influencing plasma apolipoprotein B levels. **American Journal of Human Genetics** 50: 1038-1045, 1992.

184-OA Petersen MB, Bartsch O, Adelsberger PA, Mikkelsen M, Schwinger E, **Antonarakis SE**. Uniparental isodisomy due to duplication of chromosome 21 occurring in somatic cells monosomic for chromosome 21. **Genomics** 13: 269-274, 1992.

185-OA Bowden DW, Akots G, Rothschild CR, Falls KF, Sheehy MJ, Hayward C, Mackie A, Baird J, Brock D, **Antonarakis SE**, Fajans SF. Linkage analysis of maturity onset diabetes of the young (MODY): Genetic heterogeneity and non-penetrance.

American Journal of Human Genetics 50: 607-618, 1992.

186-OA Avramopoulos D, Cox T, Forrest GL, Chakravarti A, **Antonarakis SE.** Linkage mapping of the carbonyl reductase (CBR) gene on human chromosome 21 using a DNA polymorphism in the 3' untranslated region. Genomics 13: 447-448, 1992.

187-OA Tanzi RE, Vaula G, Romano AM, Mortilla M, Huang TL, Tupler RG, Wasco W, Hyman BT, Haines JL, Jenkins BJ, Kalaitsidaki M, Warren AC, McInnis MG, **Antonarakis SE,** Karlinsky H, Persy ME, Connor L, Growdon J, Crapper-McLachlan DR, Gusella JF, St. George-Hyslop PH. Assessment of amyloid B protein precursor gene mutation in a large set of familial and sporadic Alzheimer disease cases. American Journal of Human Genetics 51: 273-282, 1992.

188-OA Warren AC, Petersen MB, Van Hul W, McInnis MG, van Broeckhoven C, Cox TK, Chakravarti A, **Antonarakis SE.** D21S215 is a (GT)n polymorphic marker close to the centromeric alphoid sequences on chromosome 21. Genomics 13: 1365-1367, 1992.

189-OA Avramopoulos D, Cox T, Kraus JP, Chakravarti A, **Antonarakis SE.** Linkage mapping of the cystathionine beta synthase (CBS) gene on human chromosome 21 using a DNA polymorphism in the 3' untranslated region. Human Genetics 90: 566-568, 1993.

190-OA Warren AC, McInnis MG, Kalaitsidaki M, Cox TK, Chakravarti A, **Antonarakis SE.** D21S210 is completely linked to the beta-amyloid protein precursor (APP) gene and is identified by a highly polymorphic (GT)n marker. Human Genetics 91: 87-88, 1993.

191-OA Hendricks L, van Duijn CM, Cras P, Cruts M, van Hul W, van Harskamp F, Warren A, McInnis MG, **Antonarakis SE,** Martin JJ, Hofman A, van Broeckhoven C. Presenile dementia and cerebral hemorrhage linked to a mutation of codon 692 of the beta-amyloid precursor protein gene. Nature Genetics 1: 218-221, 1992.

192-OA Avramopoulos D, Cox T, Chakravarti A, **Antonarakis SE.** Linkage mapping of the AML1 gene on human chromosome 21 using a DNA polymorphism in the 3' untranslated region. Genomics 14: 506-507, 1992.

193-OA Petersen MB, Frantzen M, **Antonarakis SE,** Warren AC, van Broeckhoven C, Chakravarati A, Lund C, Olsen B, Poulsen H, Sand A, Cox TK, Mikkelsen M. Comparative study of microsatellite and cytogenetic markers for detecting the origin of the nondisjoined chromosome 21 in Down syndrome. American Journal of Human Genetics 51: 516-525, 1992.

194-OA Pangalos CG, Talbot CC, Lewis JG, Adelsberger PA, Petersen MB, Serre JL, Rethore MO, de Blois MC, Parent P, Schnizel AA, Binkert F, Boue J, Corbin E, Croquette MF, Gilgenkrantz S, deGrouchy J, Bertheas MF, Prieur M, Raoul O, Serville F, Siffroi JP, Thepot F, Lejeune J, **Antonarakis SE.** DNA polymorphism analysis in families with recurrence of free trisomy 21. American Journal of Human Genetics 51: 1015-1027, 1992.

195-OA Siddique T, Hu P, Hentati A, Deng G, Hung WY, McInnis MG, Warren AC, Rimler J, **Antonarakis SE,** Pericak-Vance MA. A molecular approach to Amyotrophic Lateral Sclerosis. International Journal of Neurology 25-26: 60-69, 1992.

196-OA Kamino K, Orr HT, Payami H, Wijsman EM, Alonso ME, Pulst SM, Anderson L, O'Dahl S, Nemens E, White JA, Sadovnick AD, Ball MJ, Kaye J, Warren AC, McInnis MG, **Antonarakis SE,** Korenberg JR, Sharma V, Kukull W, Larson E, Heston LL, Martin GM, Bird TD, Schellenberg GD. Linkage and mutational analysis of familial Alzheimer disease kindreds for the APP gene region. American Journal of Human Genetics 51: 998-1014, 1992.

197-OA Kalaitsidaki M, Cox T, Chakravarti A, **Antonarakis SE.** Cloning and linkage mapping of three polymorphic tetranucleotide (TAAA)n repeats on human chromosome 21. Genomics 14: 1071-1075, 1992.

198-Book McKusick VA with the assistance of Francomano CA and **Antonarakis SE.** "Mendelian Inheritance in Man. Catalogs of autosomal dominant, autosomal recessive and X-linked phenotypes". 10th edition. The Johns Hopkins University Press, Baltimore and London, 1992.

199-OA **Antonarakis SE,** Avramopoulos D, Blouin JL, Talbot CC, Schinzel AA. Mitotic errors in somatic cells cause free trisomy 21 in about 4.5% of cases and are not associated with advanced maternal age. Nature Genetics 3: 146-150, 1993.

200-OA NIH-CEPH collaborative mapping group: A comprehensive genetic linkage map of the human genome; **Antonarakis SE,** Chakravarti A, McInnis MG, Sharma V, Avramopoulos D, Blaschak J, Litt M, Blouin JL. Chromosome 21 map. Science 258: 67-86 and 148-162, 1992.

201-OA Avramopoulos D, Chakravarti A, **Antonarakis SE.** DNA polymorphisms in the 3' untranslated region of 17 genes on human chromosome 21. **Genomics** 15: 98-102, 1993.

202-OA Warren AC, McInnis MG, Blaschak J, Petersen MB, Kalaitsidaki M, Chakravarti A, **Antonarakis SE.** Dinucleotide repeat (GT)n markers on chromosome 21. **Genomics** 14: 818-819, 1992.

203-R **Antonarakis SE.** Third International Workshop on Human Chromosome 21. Meeting Report. **Genomics** 14: 1126-1132, 1992.

204-OA Bowcock AM, Gerken SC, Barnes RI, Shiang R, Jabs EW, Warren AC, **Antonarakis SE,** Retief AE, Vergnaud G, Leppert M, Lalouel JM, White RL, Cavalli-Sforza LL. The CEPH consortium linkage map of human chromosome 13. **Genomics** 16: 486-496, 1993.

205-OA Migeon BR, McGinniss MJ, **Antonarakis SE,** Axelman J, Stasiowski BA, Youssoufian H, Kearns WG, Chung A, Pearson PL, Kazazian HH Jr, Muneer RS. Molecular basis of severe hemophilia A in a female; Order and orientation of Factor VIII gene within Xq28. **Genomics** 16: 20-25, 1993.

206-OA Chen H, Kalaitsidaki M, Warren AC, Avramopoulos D, **Antonarakis SE.** A novel zinc finger cDNA with a polymorphic pentanucleotide repeat (ATTTT)n maps on human chromosome 19. **Genomics** 15: 621-625, 1993.

207-OA Chen H, Phillips HA, Callen DF, Kim RY, Wistow GJ, **Antonarakis SE.** Localization of the human gene for μ-crystallin to chromosome 16p. **Genomics** 14: 1115-1116, 1992.

208-OA Chumakov I, Rigault P, Guillou S, Ougen P, Billaut A, Guasconi G, Gervy P, LeGall I, Soularue P, Grinas L, Bougueleret L, Bellanne-Chantelot C, Lacroix B, Barillot E, Gesnouin P, Pook S, Vaysseix G, Frelat G, Shmitz A, Sambucy JL, Bosch A, Estivill X, Weissenbach J, Vignal A, Riethman H, Cox D, Patterson D, Gardiner K, Hattori M, Sakaki Y, Ichikawa H, Ohki M, Le Paslier D, Heilig R, **Antonarakis SE,** Cohen D. A continuum of overlapping clones spanning the entire human chromosome 21q. **Nature** 359: 380-386, 1992 [PMID 1406950].

209-OA McGinniss MJ, Kazazian HH Jr, Hoyer LW, Bi L, Inaba H, **Antonarakis SE.** Spectrum of mutations in CRM positive and CRM reduced hemophilia A. **Genomics** 15: 392-398, 1993.

210-OA Li SH, McInnis MG, Margolis RL, **Antonarakis SE,** Ross CA. Novel triplet repeat containing genes in human brain: cloning, expression and length polymorphisms. **Genomics** 16: 572-579, 1993.

211-OA McGinniss MJ, Rosenberg C, Stetten G, Schinzel AA, Binkert F, Petersen MB, Kearns WG, Kazazian HH Jr, Pearson PL, **Antonarakis SE.** Unbalanced translocation, t(18:21), detected by fluorescence in situ hybridization (FISH) in a child with 18q- syndrome and a ring chromosome 21. **American Journal of Medical Genetics** 46: 647-651, 1993.

212-OA Van Hul W, Van Camp G, Stuyver L, Delabar J, McInnis MG, Warren AC, **Antonarakis SE,** Van Broeckhoven C. A contiguous physical map of the pericentromeric region of chromosome 21q between D21Z1 and D21S13E. **Genomics** 15: 626-630, 1993.

213-OA Zappata S, Petersen MB, Konig U, Blaschak J, Chakravarti A, Tassone F, Serra A, **Antonarakis SE,** Brahe C. Highly polymorphic repeat marker within the beta amyloid precursor protein gene. **Human Genetics** 93: 85-86, 1994.

214-OA McInnis MG, Chakravarti A, Blaschak J, Petersen MB, Sharma V, Avramopoulos D, Blouin JL, Konig U, Brahe C, Cox T, Warren AC, Talbot CC Jr, Van Broeckhoven C, Litt M, **Antonarakis SE.** A linkage map of human chromosome 21: 43 PCR markers at average interval of 2.5 cM. **Genomics** 16: 562-571, 1993.

215-OA Bartsch O, Petersen MB, Stuhlmann I, Mau G, Frantzen M, Schwinger E, **Antonarakis SE,** Mikkelsen M. "Compensatory" uniparental disomy of chromosome 21 in two cases. **Journal of Medical Genetics** 31: 534-540, 1994.

216-OA **Antonarakis SE,** Blouin JL, Maher J, Avramopoulos D, Thomas G, Talbot CC Jr. Maternal uniparental disomy for human chromosome 14 due to loss of a chromosome 14 from somatic cells with translocation (13;14) trisomy 14. **American Journal of Human Genetics** 52: 1145-1152, 1993.

217-BC Kazazian HH Jr, **Antonarakis SE.** Molecular genetics of the hemoglobin genes. in Singer M & Berg P editors; "Exploring genetic mechanisms". University Science Press, (ISBN 0-935702-70.9) pp 301-336, 1997.

218-R **Antonarakis SE.** Human Chromosome 21: Genome mapping and exploration circa 1993. Trends in Genetics 9: 142-148, 1993.

219-OA Talbot CC, Warren AC, Avramopoulos D, **Antonarakis SE.** Three polymorphic dinucleotide repeats at the D3S1417, D3S1418 and D12S271 loci. Human Molecular Genetics 2: 1325, 1993.

220-R Delabar LM, Creau N, Sinet PM, Ritter O, **Antonarakis SE,** Burmeister M, Chakravarti A, Nizetic D, Ohki M, Patterson D, Petersen MB, Reeves RH, Van Broeckhoven C. Report of the fourth international workshop on human chromosome 21. Genomics 18: 735-744, 1993.

221-OA Petersen MB, **Antonarakis SE,** Hassold TJ, Freeman SB, Sherman SL, Avramopoulos D, Mikkelsen M. Paternal nondisjunction in trisomy 21: excess of male patients. Human Molecular Genetics 2: 1691-1695, 1993.

222-BC Cooper DN, Krawczak M, **Antonarakis SE.** The nature and mechanisms of human gene mutation. In Scriver CR, Beaudet AL, Sly WS, Valle D. editors "The Metabolic and Molecular bases of Inherited Disease" 7th edition. (ISBN 0-07-909826-6) McGraw-Hill Co. pp 259-291, 1995.

223-R Cox DR, **Antonarakis SE,** Shimizu N. Report of the committee on the genetic constitution of chromosome 21. Genome Priority Reports 1: 556-573, 1993.

224-OA Chen H, Lowther W, Avramopoulos D, **Antonarakis SE.** Homologous loci DXYS156X and DXY156Y contain a polymorphic pentanucleotide repeat (TAAAA)n and map to human X and Y chromosomes. Human Mutation 4: 208-211, 1994.

225-BC Kazazian HH Jr, Tuddenham EGD, **Antonarakis SE.** Hemophilia A and parahemophilia: deficiencies of coagulation factors VIII and V. In Scriver CR, Beaudet AL, Sly WS, Valle D. editors "The Metabolic and Molecular bases of Inherited Disease" (ISBN 0-07-909826-6) 7th edition. McGraw-Hill Co. pp 3241-3267, 1995.

226-OL De Marchi N, Jackson L, **Antonarakis SE.** No uniparental disomy for chromosome 3 in Brachmann-de Lange syndrome. American Journal of Medical Genetics 49: 133-135, 1994.

227-OA Blouin JL, Avramopoulos D, Pangalos C, **Antonarakis SE.** Normal phenotype with paternal uniparental isodisomy for chromosome 21. American Journal of Human Genetics 53: 1074-1078, 1993.

228-OA Pulver AE, Karayiorgou M, Wolyniec PS, Lasseter VK, Kasch L, **Antonarakis SE,** Housman D, Kazazian HH, Meyers D, Nestadt G, Ott J, Lamacz M, Liang KY, Hanfelt J, Ullrich G, DeMarchi N, Ramu E, McHugh PR, Adler L, Thomas M, Carpenter WT, Manschreck T, Gordon CT, Kimberland M, Baab R, Puck J, Childs B. Sequencial strategy to identify a susceptibility gene for schizophrenia: report of potential linkage on chromosome 22 q12-q13.1. Part 1. American Journal of Medical Genetics. (Neuropsychiatric Genetics) 54: 36-43, 1994.

229-OA Pulver AE, Karayiorgou M, Lasseter VK, Wolyniec PS, Kasch L, **Antonarakis SE,** Housman D, Kazazian HH, Meyers D, Nestadt G, Ott J, Liang KY, Lamascz M, Thomas M, Childs B, Diehl SR, Wang S, Murphy B, Sun C, O'Neill, Nie L, Sham P, Burke J, Duke BW, Duke F, Kipps BR, Bray J, Hunt W, Shinkwin R, Nuallain MN, Su Y, MacLean CJ, Walsh D, Kendler KS, Gill M, Vallada H, Mant R, Asherson P, Collier D, Parfitt E, Roberts E, Nanko S, Walsh C, Daniels J, Murray R, McGuffin P, Owen M, Laurent C, Dumas JB, d'Amato T, Jay M, Martinez M, Campion D, Mallet J. Follow-up of a report of a potential linkage for schizophrenia on chromosome 22q12-q13.1. Part 2. American Journal of Medical Genetics (Neuropsychiatric Genetics) 54: 44-50, 1994.

230- BC Kaufman RJ, **Antonarakis SE.** Structure, biology and genetics of Factor VIII. in Hoffman R, Benz EJ, Shattil SJ, Furie B, Cohen HJ, and Silberstein LE eds. Hematology; Basic Principles and Practice. 2nd edition. Churchill Livingston (ISBN 0-443-08914-0), New York pp 1633-1648, 1994.

231-OA Pangalos C, Avramopoulos D, Blouin JL, Raoul O, deBlois MC, Prieur M, Schinzel AA, Gika M, Abazis D, **Antonarakis SE.** Understanding the mechanism(s) of mosaic trisomy 21 using DNA polymorphism analysis. American Journal of Human Genetics 54: 473-481, 1994.

232-OA Blouin JL, Binkert F, **Antonarakis SE.** Biparental inheritance of chromosome 21 polymorphic markers indicates that some Robertsonian translocations t(21;21) occur postzygotically. American Journal of Medical Genetics 49: 363-368, 1994.

233-OA Lakich D, Kazazian HH Jr, **Antonarakis SE,** Gitschier J. Inversions disrupting the factor VIII gene as a common

cause of severe hemophilia A. Nature Genetics 5: 236-241, 1993.

234-OA Rabinowitz VS, Maumenee IH, Lundergan MC, Puffenberger E, Zhu D, **Antonarakis SE**, Francomano CA. Molecular genetic analysis in autosomal dominant keratoconus. Cornea 11: 302-308, 1992.

235-OA Nakashima H, Yi M, Ichikawa N, LeBlond GF, **Antonarakis SE**, Ts'o POP. A two-dimentional electrophoresis Southern transfer method for detecting human genome variability using a LINE1 sequence probe. Analytical Biochemistry 227: 319-327, 1995.

236-OA Samec S, Clarkson SG, Blaschak J, Chakravarti A, Morris MA, Scherly D, **Antonarakis SE**. Dinucleotide repeat polymorphism within the human DNA repair gene XPG (ERCC5). Human Molecular Genetics 3: 214, 1994.

237-OA Figlewicz DA, McInnis MG, Goto J, Haines JL, Warren AC, Krizus A, Khodr N, Brown RH, McKenna-Yasek D, **Antonarakis SE**, Rouleau GAS. Identification of flanking markers for the familial Amyotrophic Lateral Sclerosis gene ALS1 on chromosome 21. Journal of Neurological Science (Suppl S) 124: 90-95, 1994.

238-R **Antonarakis SE**, Patterson D, Van Broeckhoven C, Shimizu N. Report of the committee on the genetic constitution of chromosome 21. Human Gene Mapping 1993 eds AJ Cuticchia & PL Pearson, Johns Hopkins Univ Press, Baltimore (ISBN 0-8018-4892-X). pgs 611-640, 1994.

239-OA Hutter P, **Antonarakis SE**, Delozier-Blanchet CD, Morris MA. Exon skipping associated with A to G transition at +4 of the IVS33 splice donor site of the Neurofibromatosis type I (NF1) gene. Human Molecular Genetics 3: 663-665, 1994.

240-BC **Antonarakis SE**, Kazazian HH, Gitschier J, Hutter P, de Moerloose P, Morris MA. Molecular etiology of Factor VIII deficiency in Hemophilia A. in Adelrod LM, Hoyer LW, Lusher JM, Reisner HM, White GC eds. "Inhibitors to Coagulation Factors" Adv. Exper. Med. Biol. Plenum press, New York. 386: 19-34, 1995.

241-OA Margolis RL, Breschel TS, Li SH, Kidwai AS, **Antonarakis SE**, McInnis MG, Ross CA. Identification and characterization of cDNA clones from human brain that contain CCA trinucleotide repeats. Somatic Cell Molular Genetics 21: 279-284, 1995.

242-OA Karayiorgou M, Kasch L, Lasseter VK, Hwang J, Elango R, Bernardini J, Kimberland M, Babb R, Francomano CA, Wolyniec PS, Lamacz M, Nestadt G, Meyers D, Ott J, Childs B, **Antonarakis SE**, Kazazian HH, Housman D, Pulver AE. Report from the Maryland epidemiology schizophrenia linkage study: no evidence for linkage between schizophrenia and a number of candidate and other genomic regions using a complex dominant model. American Journal of Medical Genetics (Neuropsychiatric Genetics) 54: 345-353, 1994.

243-OA Rossiter JP, Young M, Kimberland M, Hutter P, Ketterling RP, Gitschier J, Horst J, Morris MA, Schaid DJ, deMoerloose P, Sommer SS, Kazazian HH & **Antonarakis SE**. Factor VIII gene inversions causing severe hemophilia A originate almost exclusively in male germ cells. Human Molecular Genetics 7: 1035-1039, 1994.

244-OA Celi FS, Cohen MM, **Antonarakis SE**, Wertheimer E, Roth J, Shuldiner AR. Determination of gene dosage by a quantitative adaptation of the polymerase chain reaction (gd-PCR): rapid detection of deletions and duplications of gene sequences. Genomics 21: 304-310, 1994.

245-OL Lasseter VK, Pulver AE, Wolyniec PS, Nestadt G, Meyers D, Karayiorgou M, Housman D, **Antonarakis SE**, Kazazian HH, Kasch L, Babb R, Kimberland M, Childs B. Further analysis of the chromosome 22 linkage in families with schizophrenia. American Journal of Medical Genetics (Neuropsychiatric Genetics) 60: 172-173, 1995.

246-OA Pulver AE, Nestadt G, Goldberg R, Shprintzen RJ, Morrow B, Karayiorgou M, Lamacz M, **Antonarakis SE**, Housman D, Wolyniec PS, Kucherlapati R. Psychotic illness in patients diagnosed with velo-cardio-facial syndrome and their relatives. Journal of Nervous & Mental Disease 182: 476-478, 1994.

247-R **Antonarakis SE**, Kazazian HH Jr, Tuddenham EGD. Molecular etiology of factor VIII deficiency in hemophilia A. Human Mutation 5: 1-22, 1995.

248-OA Berry GT, Mallee JJ, Blouin JL, **Antonarakis SE**. The 21q22.1 STS marker, VN02 (EST00541 cDNA), is part of the 3' sequence of the human Na+/myo-inositol cotransporter (SLC5A3) gene. Cytogenetics & Cell Genetics 73: 77-78, 1996.

249-Let **Antonarakis SE**, McKusick VA. Nomenclature for designating mutations. Human Mutation 4: 166, 1994.

250-Book McKusick VA with the assistance of Francomano CA, **Antonarakis SE** & Pearson PL. "Mendelian Inheritance in Man. A Catalog of Human Genes and Genetic Disorders". 11 th edition. The Johns Hopkins University Press (ISBN 0-8018-4933-0); Baltimore and London, 1994.

251-OA Rogan PK, Close P, Blouin JL, Seip JR, Gannutz L, Ladda RL, **Antonarakis SE.** Duplication and loss of chromosome 21 in two children with Down syndrome and acute leukemia. American Journal of Medical Genetics 59: 174-181, 1995.

252-N&V **Antonarakis SE.** Genomic linkage scanning: systematic or intelligent ? Nature Genetics 8: 211-212, 1994.

253-R Tuddenham EGD, Schwaab R, Seehafer J, Millar DS, Gitschier J, Higuchi M, Bidichandani S, Connor JM, Hoyer LW, Yoshioka A, Peake IR, Olek K, Kazazian HH, Lavergne JM, Giannelli F, **Antonarakis SE,** Cooper DN. Haemophilia A: Database of nucleotide substitutions, deletions, insertions, and rearrangements of the factor VIII gene - second edition. Nucleic Acids Research 22: 3511-3533, 1994.

254-OA Gill M, Vallada H, Collier D, Sham P, Holmans P, Murray R, McGuffin P, Nanko S, Owen M, **Antonarakis SE,** Housman D, Kazazian HH, Nestadt G, Pulver AE, Straub RE, MacLean CJ, Walsh D, Kendler KS, DeLisi L, Polymeropoulos M, Lofthouse R, Gershon E, Golden L, Crow T, Byerley W, Coon H, Freedman R, Laurent C, Bodeau-Pean S, d'Amato T, Jay M, Campion D, Mallet J, Wildenauer D, Lerer B, Albus M, Ackenheil M, Ebstein RP, Hallmayer J, Maier W, Gurling H, Curtis D, Kalsi G, Brynjolfsson J, Sigmundson T, Petursson H, Blackwood D, Muir W, StClair D, He L, Maguire S, Moises H, Hwu HG, Yang L, Wiese C, Li T, Liu X, Kristbjamason H, Levinson D, Mowry BJ, Donnis-Keller H, Hayward NK, Crowe RR, Silverman JM, Nancarrow DJ, Read CM. A combined analysis of D22S278 marker alleles in affected sib-pairs: support for a susceptibility locus for schizophrenia at 22q12. American Journal of Medical Genetics (Neuropsychiatric Genetics) 67: 40-45, 1996.

255-OA Perrin G, Morris MA, **Antonarakis SE,** Boltshauser E, Hutter P. Two novel mutations affecting mRNA splicing of the neurofibromatosis type-1 (NF1) gene. Human Mutation 7: 172-175, 1996.

256-OA Lutfalla G, McInnis MG, **Antonarakis SE,** Uzé G. Structure of the human CRFB4 gene: comparison with its IFNAR neighbour. Journal of Molecular Evolution 41: 338-344, 1995.

257-OA Pulver AE, Lasseter VK, Kasch L, Wolyniec P, Nestadt G, Blouin J-L, Kimberland M, Babb R, Vourlis S, Chen H, Lalioti M, Morris MA, Karayiorgou M, Ott J, Meyers D, **Antonarakis SE,** Housman D, Kazazian HH. Schizophrenia: A genome scan targets chromosomes 3p and 8p as potential sites of susceptibility genes. American Journal of Medical Genetics (Neuropsychiatric Genetics) 60: 252-260, 1995.

258-OA Chrast R, Chen HM, Morris MA, **Antonarakis SE.** Mapping of the human transcription factor GABPA (E4TF1-60) gene to chromosome 21. Genomics 28: 119-122, 1995.

259-OA Chen HM, Morris MA, Rossier C, Blouin JL, **Antonarakis SE.** Cloning of the cDNA for the human ATP synthase OSCP subunit (ATP5O) by exon trapping and mapping to chromosome 21. Genomics 28: 470-476, 1995.

260-OL Chen HM, Chrast R, Rossier C, Gos A, **Antonarakis SE** ¤, Kudoh J, Yamaki A, Shindoh N, Meada H, Minoshima S, Shimizu N ¤. The human homologue of the drosophila SIM (single-minded) gene maps in the Down syndrome critical region. Nature Genetics 10: 9-10, 1995. ¤ labs of these authors equally contributed.

261-OA Bi L, Lawler AM, **Antonarakis SE,** High KA, Gearhart JD, Kazazian HH Jr. Targeted disruption of the mouse factor VIII gene: a model for hemophilia A. Nature Genetics 10: 119-121, 1995.

262-R **Antonarakis SE,** Patterson D, Van Broeckhoven C, Shimizu N, Gardiner K, Delabar J, Korenberg J, Reeves R. Report of the committee on the genetic constitution of chromosome 21. Human Gene Mapping 1994 ed AJ Cuticchia, (ISBN 0-8018-5180-7), Johns Hopkins Univ Press, Baltimore. pp 733-766, 1995.

263-OA Karayiorgou M, Morris MA, Morrow B, Sprintzen R, Goldberg R, Borrow J, Gos A, Nestadt G, Wolyniec PS, Lasseter VK, Eisen H, Childs B, Kazazian HH, Kucherlapati R, **Antonarakis SE,** Pulver AE, Housman DE. Schizophrenia susceptibility associated with interstitial deletions of chromosome 22q11. Proceedings of the National Academy of Sciences USA 92: 7612-7616, 1995 [PMID: 7644464] (abstract).

264-OL Lindsay EA, Morris MA, Gos A, Nestadt G, Wolyniec PS, Lasseter VK, Shprintzen R, **Antonarakis SE,** Baldini A, Pulver AE. Schizophrenia and chromosomal deletions within 22q11.2. American Journal of Human Genetics 56: 1502-1503, 1995.

265-OA Palmer CG, Blouin JL, Bull M, Breitfeld P, Vance GH, Van Meter T, Weaver DD, Heerema NA, Colbern SG, Korenberg J, **Antonarakis SE**, Chen X. Cytogenetic and molecular analysis of a ring(21) in a patient with partial trisomy 21 and megakaryocytic leukemia. American Journal of Medical Genetics 57: 527-536, 1995.

266-R Shimizu N, **Antonarakis SE**, Van Broeckhoven C, Patterson D, Gardiner K, Nizetic D, Creau N, Delabar J-M, Korenberg J, Reeves R, Doering J, Chakravarti A, Minoshima S, Ritter O, Cuttichia J. Report on the fifth international workshop on human chromosome 21 mapping 1994. Cytogenetics & Cell Genetics 70: 147-164, 1995.

267-OA Talbot CC Jr, Avramopoulos D, Gerken S, Chakravarti A, Armour JA, Matsunami N, White R, **Antonarakis SE**. A tetranucleotide repeat polymorphism (D21S1245) on human chromosome 21 demonstrates hypermutability in germline and somatic cells. Human Molecular Genetics 4: 1193-1199, 1995.

268-R **Antonarakis SE**. Molecular genetics of coagulation factor VIII gene and hemophilia A. Thrombosis & Haemostasis 74: 322-328, 1995.

269-OA **Antonarakis SE**, Rossiter JP, Young M, Horst J, de Moerloose P, Sommer SS, Ketterling RP, Kazazian HHJr, Négrier C, Vinciguerra C, Gitschier J, Goossens M, Girodon E, Ghanem N, Plassa F, Lavergne JM, Vidaud M, Costa JM, Laurian Y, Lin SW, Lin SR, Shen MC, Lillicrap D, Taylor SAM, Windsor S, Valleix SV, Nafa K, Sultan Y, Delpech M, Vnencak-Jones CL, Phillips JA III, Ljung RCR, Koumbarelis E, Gialeraki A, Mandalaki T, Jenkins PV, Collins PW, Pasi KJ, Goodeve A, Peake I, Preston FE, Schwartz M, Scheibel E, Ingerslev J, Cooper DN, Millar DS, Kakkar VV, Giannelli F, Naylor JA, Tizzano EF, Baiget M, Domenech M, Altisent C, Tusell J, Beneyto M, Lorenzo JI, Gaucher C, Mazurier C, Peerlinck K, Matthijs G, Cassiman JJ, Vermylen J, Mori PG, Acquila M, Caprino D, Inaba H. Factor VIII gene inversions in severe hemophilia A: Results of an international consortium study. Blood 86: 2206-2212, 1995.

270-OA Valera S, Talabot F, Evans RJ, Gos A, **Antonarakis SE**, Morris MA, Buell GN. Characterization and chromosomal localization of a human P2X receptor from urinary bladder. Receptors & Channels 3: 283-289, 1995.

271-OA Lucente D, Chen HM, Rutter M, Samec SN, Shea D, Chrast R, Rossier C, Buckler A, **Antonarakis SE**, McCormick MK. Localization of 102 exons to a 2.7 Mb region involved in Down syndrome. Human Molecular Genetics 4: 1305-1312, 1995.

272-OA Blouin J-L, Christie DH, Gos A, Lynn A, Morris MA, Ledbetter DH, Chakravarti A, **Antonarakis SE**. A new dinucleotide repeat polymorphism at the telomere of chromosome 21q reveals a significant difference between male and female rates of recombination. American Journal of Human Genetics 57: 388-394, 1995.

273-OA Chen HM, **Antonarakis SE**. Localization of a human homolog of the mouse Tiam-1 gene to chromosome 21q22.1. Genomics 30: 123-127, 1995.

274-OA Chen H, Chrast R, Rossier C, Morris MA, Lalioti M, **Antonarakis SE**. Cloning of 559 potential exons of genes on human chromosome 21 by exon trapping. Genome Research 6: 747-760, 1996.

275-BC **Antonarakis SE**. Mutations in human diseases; Nature and consequences. in Rimoin DL, Connor JM, Pyeritz RE eds. "Emery and Rimoin's Principles and Practice of Medical Genetics" 3rd ed Churchill Livingstone (ISBN 0-443-04851-7), pp 53-66, 1996.

276-OL **Antonarakis SE**, Blouin JL, Pulver AE, Wolyniec P, Lasseter VK, Nestadt G, Kasch L, Babb R, Kazazian HH Jr, Dombroski B, Kimberland M, Ott J, Housman D, MacLean C. Follow-up of potential vulnerability locus for schizophrenia on chromosome 6p24-22. Nature Genetics 11: 235-236, 1995.

277-OA Young M, Chen HM, Lalioti MD, **Antonarakis SE**. The human lanosterol synthase gene maps to chromosome 21q22.3. Human Genetics 97: 620-624, 1996.

278-OA Chen HM, Rossier C, Lalioti M, Lynn A, Chakravarti A, Perrin G, **Antonarakis SE**. Cloning of a human homolog of the drosophila white gene and mapping to chromosome 21q22.3. American Journal of Human Genetics 59: 66-75, 1996.

279-OA Blouin JL, Duriaux-Sail G, Chen HM, Gos A, Morris MA, Rossier C, **Antonarakis SE**. Mapping of the gene for the p60 subunit of the human chromatin assembly factor (CAF1A) to the Down syndrome critical region of chromosome 21. Genomics 33: 309-312, 1996.

280-CR Beaudet AL, **Antonarakis SE**, Beutler E, Cotton RGH, Desnick RJ, Kazazian HH, McAlpine PJ, McKusick VA,

Motulski AG, Scriver CR, Shows TB, Tsui LC, Valle D (Ad hoc committee on mutation nomenclature). Update on nomenclature for mutations. Human Mutation 8: 197-202, 1996.

281-OA Lalioti MD, Gos A, Green MR, Rossier C, Morris MA, **Antonarakis SE.** The gene for human U2 snRNP auxiliary factor small 35-kDa subunit (U2AF1) maps to the progressive myoclonus epilepsy (EPM1) critical region on chromosome 21. Genomics 33: 298-300, 1996.

282-R Giannacopoulos P, Hof PR, Savioz A, Guimon J, **Antonarakis SE,** Bouras C. Early-onset dementias: Clinical, neuropathologic and genetic characteristics. ACTA Neuropathologica 91: 451-465, 1996.

283-OA Chen HM, Gos A, Morris MA, **Antonarakis SE.** Localization of a human homolog of the mouse pericentrin gene to chromosome 21qter. Genomics 35: 620-624, 1996.

284-OA Radhakrishna U, Blouin JL, Solanki JV, Dhoriani GM, **Antonarakis SE.** An autosomal dominant triphalangeal thumb, polysyndactyly syndrome with variable expression in a large Indian family maps to 7q36. American Journal of Medical Genetics 66: 209-215, 1996 [PMID: 8958333].

285-OA Blouin JL, Duriaux-Sail G, **Antonarakis SE.** Mapping of the human holocarboxylase synthetase gene (HCS) to the Down syndrome critical region of chromosome 21q22. Annales de Génétique 39: 185-188, 1996.

286-R **Antonarakis SE,** Patterson D, Van Broeckhoven C, Shimizu N, Gardiner K, Delabar J, Korenberg J, Reeves R. Report of the committee on the genetic constitution of chromosome 21. Human Gene Mapping 1995 eds. Cuticchia AJ, Chipperfield M, Foster PA, Johns Hopkins Univ Press (ISBN 0-8018-5343-5). pp 1060-1099, 1996.

287-OA Sertie AL, Quimby M, Moreira ES, Murray J, Zatz M, **Antonarakis SE,** Passos-Bueno MR. A gene which causes severe ocular alterations and occipital encephalocele (Knobloch syndrome) is mapped to 21q22.3. Human Molecular Genetics 5: 843-848, 1996.

288-OA Lalioti MD, Chen HM, Rossier C, Shafaatian R, Reid JD, **Antonarakis SE.** Cloning of the cDNA for human PWP2 that encodes a protein with WD repeats and maps in 21q22.3. Genomics 35: 321-327, 1996.

289-OA Wacey AI, Kemball-Cook G, Kazazian HH, **Antonarakis SE,** Schwaab R, Lindley P, Tuddenham EGD. The haemophilia A mutation search test and resource site; home page of the factor VIII mutation database. Nucleic Acids Research 24: 100-102, 1996.

290-BC **Antonarakis SE.** Down syndrome. In "Principles of Molecular Medicine" Jameson JL, Jabs EW eds. Humana Press Inc, Totowa, NJ. (ISBN 0-89603-529-8). pp 1069-1078, 1998

291-OA Young M, Inaba H, Hoyer LW, Higuchi M, Kazazian HH Jr, **Antonarakis SE.** Partial correction of a severe molecular defect in hemophilia A due to errors during expression of the factor VIII gene. American Journal of Human Genetics 60: 565-574, 1997.

292-OA Nadal M, Mila M, Prichard M, Mur A, Pujals J, Blouin JL, **Antonarakis SE,** Ballesta F, Estivill X. YAC and cosmid FISH mapping of an unbalanced chromosome translocation causing partial trisomy 21 and Down syndrome. Human Genetics 98: 460-466, 1996.

293-OA Hutter P, Couturier A, Scott RJ, Alday P, Delozier-Blanchet C, Cachat F, **Antonarakis SE,** Joris F, Gaudin M, D'Amato L, Buerstedde JM. Complex genetic predisposition to cancer in an extended HNPCC family with an ancestral hMLH1 mutation. Journal of Medical Genetics 33: 636-640, 1996.

294-OA Eliez S, Morris MA, Dahoun-Hadorn S, Delozier-Blanchet C, Gos A, Sizonenco P, **Antonarakis SE.** Familial translocation t(Y;15)(q12;p11) and de novo deletion of the Prader-Willi syndrome (PWS) critical region on 15q11-q13. American Journal of Medical Genetics 70: 222-228, 1997.

295-OA Karayiorgou M, Gogos JA, Galke BL, Jeffery JA, Nestadt G, Lasseter VK, Wolyniec PS, **Antonarakis SE,** Kazazian HH, Housman DE, Driscoll DA, Pulver AE. Molecular characterization of the 22q11 psychosis susceptibility locus: the role of COMT. Biological Psychiatry 43: 425-431, 1998.

296-OA Chen HM, Bouras C, **Antonarakis SE.** Cloning of the cDNA for a human homologue of the rat PEP-19 gene and mapping to chromosome 21q22.2. Human Genetics 98: 672-677, 1996.

297-OA Chen HM, Rossier C, **Antonarakis SE.** Cloning of a human homolog of the Drosophila Enhancer of zeste gene (EZH2) which maps to chromosome 21q22.2. **Genomics** 38: 30-37, 1996.

298-BC **Antonarakis SE,** Scott HS. The human genome project and its impact in Medicine. In Birnstiel ML ed. "Gene Technology" **European Review** 4: 415-426, 1996.

299-OA Chen HM, **Antonarakis SE.** Localization of a human homolog of the Drosophila mnb and rat Dyrk genes to chromosome 21q22.2. **Human Genetics** 99: 262-265, 1997.

300-N&V **Antonarakis SE.** Mapping by sequence homology. **European Journal of Human Genetics** 4: 247-249, 1996.

301-OA Schizophrenia collaborative linkage group for chromosomes 3, 6, and 8 (Blackwood D, Muir W, St Clair D, Morris S, Moises HW, Yang L, Kristbjarnarson H, Helgason T, Collier DA, Holmans P, Daniels J, Rees M, Asherson P, Roberts Q, Cardno A, Arranz MJ, Ball D, Kunugi H, Murray RM, Powell JF, McGuffin P, Owen MJ, Pulver AE, **Antonarakis SE,** Babb R, Blouin JL, DeMarchi N, Dombrowski B, Housman D, Karayiorgou M, Kasch L, Kazazian HH, Lasseter VK, Loetscher E, Luebbert H, Nestadt G, Ton C, Cox-Matise T, Wolyniec PS, Laurent C, deChaldee M, Thibaut F, Jay M, Samolyk D, Petit M, Campion D, Mallet J, Straub RE, MacLean CJ, Easter SM, O'Neill FA, Walsh D, Kendler KS, Wildernauer DB, Schwab SG, Albus M, Halimeyer J, Hnig S, Borrman M, Lichtermann D, Ebstein RP, Ackenheil M, Lerer B, Risch N, Maier W, Gejman PV, Cao Q, Gerson E, Badner J, Goldin L, Zhang J, Riley BP, Rajagopalan S, Moduri-Carter M, Jenkins T, Williamson R, Garner C, Joslyn G, Carey A, LeDuc C, Cardon L, Lichter J, Harris T, Loftus J, Shields G, Comasi M, Vita A, Smith A, Dann J, Crow TJ, DeLisi LE, Gurling H, Kalsi G, Brynjolfsson J, Curtis D, Sigmundsson T, Butler R, Read T, Murphy P, Chen ACH, Petursson H, Byerley W, Coon H, Levinson DF, Nancarrow DJ, Kryglyak L, Crowe RR, Andreasen N, Silverman JM, Mohs RC, Siever LJ, Endicott J, Sharpe L, Walters MK, Lennon DP, Hayward NK, Sandkuijl LA, Mowry BJ, Aschauer HN, Meszaros K, Lenzinger E, Fuchs K, Holmans P). Support for schizophrenia linkage findings on chromosomes 6 and 8: a multicenter study. **American Journal of Medical Genetics** (Neuropsychiatric Genetics) 67: 580-594, 1996.

302-OA Buell GN, Talabot F, Gos A, Lorenz J, Lai E, Morris MA, **Antonarakis SE.** Gene structure and chromosomal localization of the human P2X7 receptor gene. **Receptors & Channels** 5: 347-354, 1998.

303-OA Bittencourt MC, Morris MA, Chabod J, Gos A, Lamy B, Fellmann F, **Antonarakis SE,** Plouvier E, Herve P, Tiberghien P. Fortuitous detection of uniparental disomy of chromosome 6. **Journal of Medical Genetics** 34: 77-78, 1997.

304-OA Radhakrishna U, Blouin JL, Mehenni H, Mehta TY, Sheth FJ, Sheth JJ, Solanki JV, **Antonarakis SE.** The gene for autosomal dominant hidrotic ectodermal dysplasia (Clouston syndrome) in a large Indian family maps to the pericentromeric region of chromosome 13q11-q12.1. **American Journal of Medical Genetics** 71: 80-86, 1997.

305-Let **Antonarakis SE.** Premeiotic trisomy 21 in oocytes and Down syndrome: a reply to Zheng and Byers hypothesis. **American Journal of Human Genetics** 59: 1395-1397, 1996.

306-OA Scott HS, Chen H, Rossier C, Lalioti MD, **Antonarakis SE.** Isolation of a human gene (HES1) with homology to an E.Coli and a zebrafish protein that maps to chromosome 21q22.3. **Human Genetics** 99: 616-623, 1997.

307-OA Lalioti MD, Mirotsou M, Buresi C, Peitsch MC, Rossier C, Quazzani R, Baldy-Moulinier M, Bottani A, Malafosse A, **Antonarakis SE.** Identification of mutations in Cystatin B, the gene responsible for the Unverricht-Lundborg type of progressive myoclonus epilepsy (EPM1). **American Journal of Human Genetics** 60: 342-351, 1997.

308-OA Fokstuen S, Bottani A, Medeiros PFV, **Antonarakis SE,** Stoll C, Schinzel A. Laryngeal atresia type III (glottic web) with 22q11.2 microdeletion: report of three patients. **American Journal of Medical Genetics** 70: 130-133, 1997.

309-OA Pulver AE, Wolyniec PS, Housman D, Kazazian HH, **Antonarakis SE,** Nestadt G, Lasseter VK, McGrath JA, Dombroski B, Karayiorgou M, Ton C, Blouin JL, Kempf L. The Johns Hopkins University Collaborative Schizophrenia study: An epidemiologic-genetic apprOAch to test the heterogeneity hypothesis and identify schizophrenia susceptibility genes. **Cold Spring Harbor Symposia on Quantitative Biology** LXI: 797-814, 1996.

310-OA Mittaz L, Scott HS, Rossier C, Seeburg PH, Higuchi M, **Antonarakis SE.** Cloning of a human RNA editing deaminase (ADARB1 or RED1) of glutamate receptors which maps to chromosome 21q22.3. **Genomics** 41: 210-217, 1997.

311-OA Chen H, Rossier C, Nakamura Y, Lynn A, Chakravarti A, **Antonarakis SE.** Cloning of a novel homeobox-containing gene PKNOX1 and mapping to human chromosome 21q22.3. **Genomics** 41: 193-200, 1997.

312-OA Savioz A, Blouin JL, Guidi S, **Antonarakis SE,** Bouras C. A method of extraction of genomic DNA from human brain fixed and stored in formalin for many years. **ACTA Neuropathology** 93: 408-413, 1997.

313-OA Radhakrishna U, Blouin JL, Mehenni H, Patel UC, Patel MM, Solanki JV, **Antonarakis SE.** Mapping one form of autosomal dominant postaxial polydactyly type-A (PAP-A) to chromosome 7p15-q11.23 by linkage analysis. **American Journal of Human Genetics** 60: 597-604, 1997.

314-OA Chen H, **Antonarakis SE.** The SH3D1A gene maps to human chromosome 21q22.1-q22.2. **Cytogenetics & Cell Genetics** 78: 213-215, 1997.

315-OA Bartsch O, Hinkel GK, Petersen MB, Konig U, Bugge M, Mikkelsen M, Avramopoulos D, Morris M, **Antonarakis SE.** A large family with subtelomeric translocation t(18;21)(q23;q22.13) and molecular breakpoint in the Down syndrome critical region. **Human Genetics** 100: 669-675, 1997.

316-OA Chrast R, Scott HS, Chen H, Kudoh J, Rossier C, Minoshima S, Wang Y, Shimizu N, **Antonarakis SE.** Cloning of two human homologs of the Drosophila neurodevelopmental regulator single-minded (sim) gene, SIM1 on chromosome 6q and SIM2 on 21q within the Down syndrome chromosomal region. **Genome Research** 7: 615-624, 1997.

317-OA Paoloni-Giacobino A, Chen H, **Antonarakis SE.** Cloning of a human Neural Cell Adhesion molecule (NCAM-2) gene that maps on chromosome 21q21, and is potentially involved in Down syndrome. **Genomics** 43: 43-51, 1997.

318-OA Mittaz L, **Antonarakis SE,** Higuchi M, Scott HS. Localization of a novel human RNA editing deaminase (hRED2 or ADARB2) to chromosome 10p15. **Human Genetics** 100: 398-400, 1997.

319-OA Villard J, Reith W, Barras E, Gos A, Morris MA, **Antonarakis SE,** Van der Elsen PJ, Mach B. Analysis of mutations and chromosomal localization of the gene encoding RFX5, a novel transcription factor affected in major histocompatibility complex class II deficiency. **Human Mutation** 10: 430-435, 1997.

320-N&V Scriver C, Cotton R, **Antonarakis SE,** McKusick V. Mutation database initiative to go forward under HUGO. **Genetics Digest** 4: 12-15, 1997.

321-OA Lalioti MD, Scott HS, Buresi C, Rossier C, Bottani A, Morris MA, Malafosse A, **Antonarakis SE.** Dodecamer repeat expansion in cystatin B gene in progressive myoclonus epilepsy. **Nature** 386: 847-851, 1997.

322-OA Neerman-Arbez M, **Antonarakis SE,** Blouin JL, Zeinali S, Akhtari M, Afshar Y, Tuddenham EGD. The locus for combined factor V- factor VIII (F5F8D) maps to 18q21 between D18S849 and D18S1103. **American Journal of Human Genetics** 61: 143-150, 1997.

323-OA Scott HS, **Antonarakis SE,** Lalioti MD, Rossier C, Silver PA, Henry MF. Identification and characterization of two putative human arginine methyltransferases (HRMT1L1 and HRMT1L2) that map to chromosome 21q22.3 and 19q respectively. **Genomics** 48: 330-340, 1998.

324-OA Paoloni-Giacobino A, Chen H, Peitsch MC, Rossier C, **Antonarakis SE.** Cloning of the TMPRSS2 gene which encodes a novel serine protease with transmembrane, LDLRA, and SRCR domains, and maps to 21q22.3. **Genomics** 44: 309-320, 1997.

325-BC Scott HS, **Antonarakis SE.** The "genomics" of chromosome 21 and Down syndrome. In Schumacher G, Sauer U, eds. "Genetics of Cardiopathies" (ISBN 3-8047-1635-0) Wissenschaftliche Verlag, Stuttgart. Pp 164-179, 1999.

326-OL Lalioti MD, Scott HS, **Antonarakis SE.** What is expanded in progressive myoclonus epilepsy (EPM1) ? **Nature Genetics** 17: 17, 1997.

327-OA Radhakrishna U, Wild A, Grzeschik KH, **Antonarakis SE.** Mutation in GLI3 in postaxial polydactyly type A. **Nature Genetics** 17: 269-271, 1997 (doi: 10.1038/ng1197-269) PMID: 935478.

328-OA Scott HS, Kyriakou DS, Peterson P, Heino M, Tahtinen M, Krohn K, Chen H, Rossier C, Lalioti MD, **Antonarakis SE.** Cloning of a novel gene, C21ORF2, in the APECED critical region of human chromosome 21q22.3, and its mutation analysis. **Genomics** 47: 64-70, 1998.

329-OA Ives JH, Dagna-Bricarelli F, Basso G, **Antonarakis SE,** Jee R, Cotter F, Nizetic D. Increased levels of the

chromosome 21 encoded tumour invasion and metastasis (TIAM1) mRNA in bone marrow of Down syndrome children during the acute phase of AML (M7). **Genes Chromosomes & Cancer** 23: 61-66, 1998.

330-OA Karayiorgou M, Gogos JA, Galke BL, Jeffery JA, Nestadt G, Wolyniec PS, **Antonarakis SE**, Kazazian HH, Housman DE, Driscoll DA, Pulver AE. Genotype and phenotype analysis at the 22q11 schizophrenia susceptibility locus. **Cold Spring Harbor Symposia on Quantitative Biology** LXI: 835-843, 1996.

331-OA Mehenni H, Blouin JL, Radhakrishna U, Baradwaj S, Baradwaj K, Dixit VB, Richards KF, Fenoll BA, Silva Leal A, Raval RC, **Antonarakis SE.** Peutz-Jeghers syndrome: confirmation of linkage to chromosome 19q13.3 and identification of a potential second locus on 19q13.4. **American Journal of Human Genetics** 61: 1327-1334, 1997.

332-OA Nagamine K, Peterson P, Scott HS, Kudoh J, Minoshima S, Heino M, Krohn KJE§, Lalioti MD, Mullis PE, **Antonarakis SE§**, Kawasaki K, Asakawa S, Ito F, Shimizu N§. Positional cloning of the APECED gene. §labs equally contributed. **Nature Genetics** 17: 393-398, 1997 (doi: 10.1038/ng1297-393) PMID: 9398839.

333-OA Krohn K, Ovod V, Vilja P, Heino M, Scott H, Kyriakou DS, **Antonarakis SE**, Jacobs HT, Isola J, Peterson P. Immunochemical characterization of a novel mitochondrially located protein encoded by a nuclear gene within the DFNB8/10 critical region on 21q22.3. **Biochemical Biophysical Research Communications** 238: 806-810, 1997.

334-OA Dahmane N, Ait Ghezala G, Gosset P, Chamoun Z, Dufresne-Zacharia MC, Lopes C, Rabatel N, Gassanova-Maugenre S, Chettouh Z, Abramowski V, Fayet E, Yaspo ML, Korn B, Blouin JL, Lehrach H, Poutska A, **Antonarakis SE**, Sinet PM, Creau N, Delabar JM. Transcriptional map of the 2.5 Mb CBR-ERG region of chromosome 21 involved in Down syndrome. **Genomics** 48: 12-23, 1998.

335-CR White JA, McAlpine PJ, **Antonarakis SE**, Cann H, Eppig J, Frazer K, Frezal J, Lancet D, Nahmias J, Pearson P, Peters J, Scott A, Scott H, Spurr N, Talbot C, Povey S. Guidelines for human gene nomenclature (1997). **Genomics** 45: 468-471, 1997.

336-R Neerman-Arbez M, **Antonarakis SE.** Vers le développement de thérapies géniques pour l' hémophilie. **Médecine. et Hygiene** 55: 1543-1548, 1997.

337-CR **Antonarakis SE** and the mutation nomenclature working group. Recommendations for a nomenclature system for human gene mutations. **Human Mutation** 11: 1-3, 1998.

338-BC Cooper DN, Krawczak M, **Antonarakis SE.** The nature and mechanisms of human gene mutation. In Vogelstein B, Kinzler KW editors "The Genetic basis of human cancer" (ISBN 0-07-067596-1) McGraw-Hill Co. pp. 65-94, 1997.

339-OA Vargas FR, Roessler E, Gaudenz K, Belloni E, Whitehead S, Kirke PN, Mills J, Hooper G, Stevenson RE, Cordeiro I, Correia P, Felix T, Gereige R, Cunningham ML, Canun S, **Antonarakis SE**, Strachan T, Tsui LC, Scherer SW, Muenke M. Analysis of the human Sonic Hedgehog coding and promoter regions in sacral agenesis, triphalangeal thumb, and mirror polydactyly. **Human Genetics** 102: 387-392, 1998.

340-R **Antonarakis SE.** Molecular genetics of coagulation factor VIII gene and Haemophilia A. **Haemophilia** 4: 1-11 (Suppl 2), 1998.

341-OA Blouin LJ, Duriaux-Sail G, Guipponi M, Rossier C, Papasavvas MP, **Antonarakis SE.** Isolation of the human BACH1 transcription regulator gene which maps to chromosome 21q22.1. **Human Genetics** 102: 282-288, 1998.

342-OA Chen H, Rossier C, Morris MA, Scott HS, Gos A, Bairoch A, **Antonarakis SE.** A testis-expressed gene (TPTE), encodes a putative transmembrane tyrosine phosphatase and maps to the pericentromeric region of human chromosome 21, and to chromosomes 13, 15, 22, and Y. **Human Genetics** 105: 399-409, 1999.

343-OA Lapenta V, Sossi V, Gosset P, Vitali T, Rabatel N, Tassone F, Vayssettes C, Blouin JL, Scott HS, **Antonarakis SE**, Creau N, Brahe C. Construction of a 2.5 Mb integrated physical, genetic and gene map of distal 21q22.3. **Genomics** 49: 1-13, 1998.

344-R **Antonarakis SE.** Expansion of a 12mer (CCCCGCCCCGCG) repeat in progressive myoclonus epilepsy. In "Genetic instabilities and hereditary neurological disorders" Wells RD & Warren ST eds. Academic Press. (ISBN 0-12-742935-2), pp 779-790, 1998.

345-OA Lalioti MD, Scott HS, Genton P, Grid D, Ouazzani R, M'Rabet A, Ibrahim S, Gouider R, Dravet C, Chkili T, Bottani A,

Burezi C, Malafosse A, **Antonarakis SE.** A PCR amplification method reveals instability of the dodecamer repeat in Progressive myoclonus epilepsy (EPM1) and no correlation between the size of the repeat and age of onset. American Journal of Human Genetics 62: 842-847, 1998.

346-OA Knoepfler PS, Calvo KR, Chen H, **Antonarakis SE,** Kamps MP. Meis1 and Pknox1 bind DNA cooperatively with Pbx1 utilizing an interaction surface disrupted in oncoprotein E2a-Pbx1. Proceedings of the National Academy of Sciences USA 94: 14553-14558, 1997.

347-OA Scott HS, Heino M, Peterson P, Mittaz L, Lalioti MD, Betterle C, Cohen A, Romeo G, Seri M, Lerone M, Collin P, Salo M, Metcalfe R, Weetman A, Papasavvas MP, Rossier C, Nagamine K, Kudoh J, Shimizu N, Krohn KJE, **Antonarakis SE.** Common mutations in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED) patients of different origins. Molecular Endocrinology 12: 1112-1119, 1998.

348-BC Kaufman RJ, **Antonarakis SE.** Structure, biology and genetics of Factor VIII. in Hoffman R, Benz EJ, Shattil SJ, Furie B, Cohen HJ, Silberstein LE and McGlave P eds. "Hematology; Basic Principles and Practice". 3rd edition. Churchill Livingston (ISBN 0-443-07954-4), New York pp 1850-1869, 1999.

349-R **Antonarakis SE.** 10 years of Genomics, chromosome 21 and Down syndrome. Genomics 51: 1-16, 1998.

350-OA Curtis L, Blouin JL, Radhakrishna U, Gehrig C, Lasseter VK, Wolyniec P, Nestadt G, Dombroski B, Kazazian HH, Pulver AE, Housman D, Bertrand D, **Antonarakis SE.** No evidence for linkage between schizophrenia and markers at chromosome 15q13-14. American Journal of Medical Genetics (Neuropsychiatric Genetics) 88: 109-112, 1999.

351-OA Rosatelli MC*, Meloni AI, Meloni An, Devoto M, Cao A, Scott HS*, Peterson P, Heino M, Krohn KJE, Nagamine K, Kudoh J, Shimizu N, **Antonarakis SE.** A common mutation in Sardinian autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED) patients. (*equally contributed). Human Genetics 103: 428-434, 1998.

352-OA Peterson P, Nagamine K, Scott HS, Heino M, Kudoh J, Shimizu N, **Antonarakis SE,** Krohn KJE. APECED: a monogenic autoimmune disease providing new clues to self-tolerance. Immunology Today 19: 384-386, 1998.

353-OA Blouin JL*, Dombroski B*, Nath SK*, Lasseter VK, Wolyniec PS, Nestadt G, Thornquist M, Ullrich G, McGrath J, Kasch L, Lamacz M, Gehrig C, Radhakrishna U, Snyder SE, Balk KG, Neufeld K, DeMarchi N, Papadimitriou GN, Dikeos DG, Stefanis CN, Chakravarti A, Childs B, Housman DE, Kazazian HH, **Antonarakis SE,** Pulver AE. Multigenic inheritance of schizophrenia: A genome scan identifies susceptibility loci on chromosomes 13q32 and 8p21. (*equally contributed). Nature Genetics 20: 70-73, 1998.

354-OA Stratakis CA, Kirschner LS, Taymans SE, Tomlinson IPM, Marsh DJ, Torpy DJ, Giatzakis C, Eccles DM, Theaker J, Houlston RS, Blouin JL, **Antonarakis SE,** Basson CT, Eng C, Carney JA. Carney complex, Peutz-Jeghers syndrome, Cowden disease and Bannayan-Zonana syndrome share cutaneous and endocrine manifestations but not genetic loci. Journal of Clinical Endocrinology Metabolism 83: 2972-2976, 1998.

355-OA Theophilus BDM, Enayat MS, Higuchi M, Kazazian HH Jr, **Antonarakis SE,** Hill FGH. Independent occurrence of the novel Arg2163His mutation in the factor VIII gene in three unrelated families with Hemophilia A with different phenotypes. Human Mutation 11: 334, 1998.

356-OA Schizophrenia collaborative linkage group for chromosome 22, Vallada H, Curtis D, Sham P, Kinugi H, Zhao J, Murray R, McGuffin P, Nanko S, Owen M, Gill M, Collier D, **Antonarakis SE,** Housman D, Kazazian HH, Nestadt G, Pulver AE, Straub RE, MacLean CJ, Walsh D, Kendler KS, DeLisi L, Polymeropoulos M, Coon H, Byerley W, Lofthouse R, Gershon E, Goldin L, Freedman R, Laurent C, Bodeau-Pean S, D'Amato T, Jay M, Campion D, Mallet J, Wildenauer D, Lerer B, Albus M, Ackenheil M, Ebstein RP, Hallmayer J, Maier W, Gurling H, Kalsi G, Brynjolfsson J, Sigmundson T, Petursson H, Blackwood D, Muir W, StClair D, He L, Maguire S, Moises H, Hwu HG, Yang L, Wiese C, Kristbjamason H, Levinson D, Mowry BJ, Donnis-Keller H, Hayward NK, Crowe RR, Silverman JM, Nancarrow DJ, Read CM. A transmission disequilibrium and linkage analysis of D22S278 marker alleles in 574 families: further support for a susceptibility locus for schizophrenia at 22q12. Schizophrenia Research 32: 115-121, 1998.

357-N&V **Antonarakis SE,** Van Aelst L. Mind the GAP, Rho, Rab, and GDI. Nature Genetics 19: 106-108, 1998.

358-CR Jones P, **Antonarakis SE,** Battistella L, Chuansumrit A, Dowdeswell B, Evatt B, Ginter EK, Lee C, Peake I, Preston E, Rickard K, Srivastava A, Kasper C, Robillard L, Napalkov NP, Tsechkovski MS, Wasunna A, Boulyjenkov V, Emmanuel J, Richstein K. Control of Haemophilia: Haemophilia care in developing countries. Report of a joint W.H.O./World Federation of

Haemophilia meeting. WHO/HGN/WFH/WG/98.3 pp 1-29, 1998.

359-OA Heino M, Scott HS, Chen Q, Peterson P, Maenpaa U, Papasavvas MP, Mittaz L, Barras C, Rossier C, Chroussos GP, Stratakis CA, Nagamine K, Kudoh J, Shimizu N, McLaren N, **Antonarakis SE**, Krohn KJE. Mutation analysis of Northern American APS-1 patients. **Human Mutation** 13: 69-74, 1999.

360-OA **Antonarakis SE**, Blouin JL, Lasseter VK, Gehrig C, Radhakrishna U, Nestadt G, Housman D, Kazazian HH, Kalman K, Gutman G, Fantino E, Chandy KG, Gargus JJ, Pulver AE. Lack of linkage or association between schizophrenia and the polymorphic trinucleotide repeat within the KCNN3 gene on chromosome 1q21. **American Journal of Medical Genetics** (Neuropsychiatric Genetics) 83: 348-351, 1999.

361-OA Guipponi ML, Scott HS, Chen H, Schebesta A, Rossier C, **Antonarakis SE**. Two isoforms of a human intersectin protein are produced by brain specific alternative splicing in a stop codon. **Genomics** 53: 369-376, 1998.

362-OA Mehenni H, Gehrig C, Nezu JI, Oku A, Shimanand M, Rossier C, Blouin JL, Guex N, Radhakrishna U, Scott HS, **Antonarakis SE**. Loss of LKB1 activity in Peutz-Jeghers syndrome and evidence for allelic and locus heterogeneity. **American Journal of Human Genetics** 63: 1641-1650, 1998.

363-BC Kaufman RJ, **Antonarakis SE**, Fay PJ. Factor VIII and Hemophilia A. in Colman RW, Hirsh J, Marder VJ, Clowes AW, George JN eds. "Hemostasis and Thrombosis: Basic principles and clinical practice" 4th edition, chapter 8, Lippincott, Williams & Wilkins (ISBN 0-7817-1455-9), Philadelphia, PA. pp 135-156, 2000.

364-OA Guipponi M, Scott HS, Kudoh J, Kawasaki K, Shibuya K, Shintani A, Asakawa S, Chen H, Lalioti MD, Rossier C, Minoshima S, Shimizu N, **Antonarakis SE**. Identification and characterization of a novel cyclic nucleotide phosphodiesterase gene (PDE9A) that maps to 21q22.3; alternative splicing of mRNA transcripts, genomic structure and sequence. **Human Genetics** 103: 386-392, 1998.

365-OA Radhakrishna U, Bornholdt D, Scott HS, Patel UC, Rossier C, Engel H, Bottani A, Singh D, Blouin JL, Solanki JV, Grzeschik KH, **Antonarakis SE**. The phenotypic spectrum of GLI3 morphopathies includes autosomal dominant preaxial polydactyly type-IV (PPD-IV) and postaxial polydactyly type-A/B (PAP-A/B); no phenotype prediction from the position of GLI3 mutations. **American Journal of Human Genetics** 65: 645-655, 1999.

366-OA Neerman-Arbez M, Johnson KM, Morris MA, McVey JH, Peyvandi F, Nichols WC, Ginsburg D, Rossier C, **Antonarakis SE**, Tuddenham EGD. Molecular analysis of the ERGIC-53 gene in 35 families with combined factor V-factor VIII deficiency (F5F8D). **Blood** 93: 2253-2260, 1999.

367-Book McKusick VA with the assistance of **Antonarakis SE**, Francomano CA, Hurko O, Scott AF, Smith M, Valle D. "Mendelian Inheritance in Man. A Catalog of Human Genes and Genetic Disorders". 12th edition. The Johns Hopkins University Press (ISBN 0-8018-5742-2); Baltimore and London, 1998.

368-OA Neerman-Arbez M, Honsberger A, **Antonarakis SE**, Morris MA. Deletions of the fibrinogen alpha-chain gene (FGA) cause congenital afibrinogenemia. **Journal of Clinical Investigation** 103: 215-218, 1999.

369-OA Heino M, Peterson P, Kudoh J, Nagamine K, Lagerstedt A, Ovod V, Ranki A, Rantala I, Nieminen M, Tuukkanen J, Scott HS, **Antonarakis SE**, Shimizu N, Krohn KJE. Autoimmune regulator (AIRE) is expressed both in epithelial and non-epithelial cells in thymus medulla and in peripheral lymphoid tissues. **Biochemical Biophysical Research Communications** 257: 821-825, 1999.

370-OA Wang SY, Cruts M, Del-Favero J, Zhang Y, Bosch, Chen HM, Bennett L, Patterson D, Potier MC, Nizetic D, Estivill X, **Antonarakis SE**, Kessling A, Van Broeckhoven C. An STS-based contig map of human chromosome 21p ranging from the centromere to the rDNA gene cluster. **Genome Research** 9: 1059-1073, 1999.

371-OA Guipponi ML, Scott HS, Hattori M, Ishii K, Sakaki Y, **Antonarakis SE**. Genomic structure, sequence and refined mapping of the human intersectin (ITSN) gene which encompasses 250 kb on chromosome 21q22.1-q22.2. **Cytogenetics & Cell Genetics** 83: 218-220, 1999.

372-BC **Antonarakis SE**, Krawczak M, Cooper DN. The nature and mechanisms of human gene mutation (chapter 13). In Scriver CR, Beaudet AL, Sly WS, Valle D, Childs B, Kinzler KW, Vogelstein B, editors "The Metabolic and Molecular bases of Inherited Disease" 8th edition. (ISBN 0-07-913035-6) McGraw-Hill Co. pp 343-378, 2001.

373-BC Kazazian HH Jr, Tuddenham EGD, **Antonarakis SE.** Hemophilia A: Deficiency of coagulation factor VIII. In Scriver CR, Beaudet AL, Sly WS, Valle D, Childs B, Kinzler KW, Vogelstein B. editors "The Metabolic and Molecular bases of Inherited Disease" 8th edition. (ISBN 0-07-913035-6) McGraw-Hill Co. pp 4367-4392, 2001.

374-OA Chrast R, Scott HS, **Antonarakis SE.** Optimized protocols for linearization and purification of BAC DNA for the development of transgenic mice. Transgenic Research 8: 147-150, 1999.

375-OA Bartoloni L, Blouin JL, Sainsbury AJ, Gos A, Morris MA, Affara NA, DeLozier-Blanchet C.D, **Antonarakis SE.** Assignment of the human dynein heavy chain gene DNEL1 to human chromosome 17p12 by in situ hybridization and radiation hybrid mapping. Cytogenetics & Cell Genetics 84: 188-189, 1999.

376-R Paoloni-Giacobino A, **Antonarakis SE.** Vieillissement et longévité: hypothèses génétiques. Médécine et Hygiène 57: 1718-1725, 1999.

377-OA Mittaz L, Rossier C, Heino M, Gos A, Morris MA, Peterson P, Krohn KJE, Kudoh J, Shimizu N, **Antonarakis SE,** Scott HS. Isolation and characterization of the mouse Aire gene. Biochemical Biophysical Research Communications 255: 483-490, 1999.

378-R Scott HS, **Antonarakis SE,** Mittaz L, Lalioti MD, Younus F, Mohyuddin A, Mehdi SQ, Gal A. Refined genetic mapping of the autosomal recessive non-syndromic deafness locus DFNB8 on human chromosome 21q22.3. Advances in Otorhinolaryngology 56: 158-163, 2000.

379-OA Kibar Z, Dubé MP, Powell J, McCuaïg C, Hayflick SJ, Zonana J, Hovnanian A, Radhakrishna U, **Antonarakis SE,** Benohanian A, Sheeran A, Stephan ML, Gosselin R, Kelsell D, Christianson AL, Fraser FC, Der Kaloustian VM, Rouleau GY. Clouston hidrotic ectodermal dysplasia (HED): genetic homogeneity, presence of a founder effect in the French Canadian population and fine genetic mapping. European Journal of Human Genetics 8: 372-380, 2000.

380-OA Lalioti MD, Scott HS, **Antonarakis SE.** Altered spacing of promoter elements due to the dodecamer repeat expansion contributes to reduced expression of the cystatin B gene in EPM1. Human Molecular Genetics 8: 1791-1798, 1999.

381-OA Roessler E, Mittaz L, Du A, Scott HS, Chang J, Rossier C, Guipponi M, Matsuda SPT, Muenke M, **Antonarakis SE.** Structure of the human Lanosterol synthetase gene and its analysis as a candidate for holoprocencephaly. (HPE1). Human Genetics 105: 489-495, 1999.

382-OA Neerman-Arbez M, **Antonarakis SE,** Honsberger A, Morris MA. The 11 kb deletion responsible for congenital afibrinogenemia is mediated by a short direct repeat in the fibrinogen gene cluster. European Journal of Human Genetics 7: 897-902, 1999.

383-OA Blouin J-L, Meeks M, Radhakrishna U, Sainsbury AJ, Gehrig C, Duriaux-Saïl G, Dombi V, O'Rawe A, Walne A, Chung E, Afzelius BA, Armengot M, Jorissen M, Schidlow DV, Van Maldergem L, Walt H, Gardiner RM, Probst D, Guerne P-A, Delozier-Blanchet CD, **Antonarakis SE.** Primary Ciliary Dyskinesia: A genome-wide linkage analysis reveals extensive locus heterogeneity. European Journal of Human Genetics 8: 109-118, 2000.

384-OA Lynn A, Kashuk C, Petersen MB, Bailey JA, Cox DR, **Antonarakis SE,** Chakravarti A. Patterns of meiotic recombination on the long arm of human chromosome 21. Genome Research 10: 1319-1332, 2000.

385-R Blouin JL, **Antonarakis SE.** Le syndrome de Kartagener et le syndrome des cils immobiles. Médécine et Hygiène 57: 1881-1884, 1999.

386-OA Neerman-Arbez M, De Moerloose P, Bridel C, Honsberger A, Schonborner A, Rossier C, Peerlink K, Claessens S, Di Michele D, D'Oiron R, Dreyfus M, Laubriat-Blanchin M, Dieval J, **Antonarakis SE,** Morris MA. Mutations in the fibrinogen A gene (FGA) account for the majority of congenital afibrinogenemia. Blood 96: 149-152, 2000.

387-CR Groner Y, **Antonarakis SE,** Levanon D. Summary report on the EMBO workshop on "Molecular Biology of Chromosome 21 and Down syndrome. Cytogenetics & Cell Genetics 86: 1-20, 1999.

388-CR den Dunnen JT, **Antonarakis SE.** Mutation nomenclature extensions and suggestions to describe complex mutations: a discussion paper. Human Mutation 15: 7-12, 2000.

389-OA Radhakrishna U, Senol S, Herken H, Gucuyener K, Gehrig C, Blouin JL, Akarsu NA, **Antonarakis SE.** An apparently dominant bipolar affective disorder (BPAD) locus on chromosome 20p11.2-q11.2 in a large Turkish pedigree. European Journal of Human Genetics 9: 39-44, 2001.

390-OA Aapola U, Shibuya K, Scott HS, Ollila J, Vihinen M, Heino M, Shintani A, Kawasaki K, Minoshima S, Krohn K, **Antonarakis SE**, Shimizu N, Kudoh J, Peterson P. Isolation and initial characterization of a novel zinc-finger gene, DNMT3L on 21q22.3, related to cytosine-5-methyltransferase-3 gene family. Genomics 65: 293-298, 2000.

391-OA Guipponi M, Brunschwig K, Chamoun Z, Scott HS, Shibuya K, Kudoh J, Delezoide AL, El-Samadi S, Chettouh Z, Rossier C, Shimizu N, Mueller F, Delabar JM, **Antonarakis SE.** C21orf5, a novel human chromosome 21 gene that is potentially involved in Down syndrome has a C.Elegans ortholog (pad-1) required for embryonic patterning. Genomics 68: 30-40, 2000.

392-OA Heino M, Peterson P, Sillanpaa N, Guerin S, Naquet P, Anderson G, Scott HS, Kudoh J, Shimizu N, **Antonarakis SE**, Jenkinson EJ, Krohn KJE. RNA and protein expression of the murine autoimmune regulator gene (Aire) in normal, RelB deficient and NOD mouse. European Journal of Immunology 30: 1884-1893, 2000.

393-OA Eliez S, **Antonarakis SE,** Morris MA, Dahoun SP, Reiss AL. Parental origin of the deletion 22q11.2 and brain development in Velocardiofacial syndrome; a preliminary study. Archives of General Psychiatry 58: 64-68, 2001.

394-OA Pitkanen J, Doucas V, Sternsdorf T, Nakajima T, Aratani S, Jensen K, Will H, Vahamurto P, Ollila J, Vihinen M, Scott HS, **Antonarakis SE**, Kudoh J, Shimizu N, Krohn KJE, Peterson P. The autoimmune regulator protein (AIRE) has transcriptional transactivating properties and interacts with common co-activator CREB-binding protein (CBP). Journal of Biological Chemistry 275: 16802-16809, 2000.

395-OA Chrast R, Scott HS, Madani R, Huber L, Wolfer DP, Prinz M, Aguzzi A, Lipp HP, **Antonarakis SE.** Mice trisomic for a BAC with the single minded 2 gene (Sim2) show some of the phenotypes of the partial trisomy 16 mouse models of Down syndrome. Human Molecular Genetics 9: 1853-1864, 2000 (DOI: 10.1093/hmg/9.12.1853) PMID: 10915774.

396-OA Eliez S, Palacio-Espasa F, Spira A, Lacroix M, Pont C, Luthi F, Schorderet D, Robert-Tissot C, **Antonarakis SE**, Cramer B. Young children presenting a 22q11.2 deletion; psychological and language phenotypes. European Journal Child & Adolescent Psychiatry 9: 109-114, 2000.

397-OA Michaud J, Kudoh J, Berry A, Bonne-Tamir B, Lalioti MD, Rossier C, Shibuya K, Kawasaki K, Asakawa S, Minoshima S, Shimizu N, **Antonarakis SE**, Scott HS. Isolation and characterization of a human chromosome 21q22.3 Gene (WDR4) and its mouse homologue that code for a WD-Repeat protein. Genomics 68: 71-79, 2000.

398-OA Maiti AK, Bartoloni L, Mitchison, H, Meeks M, Chung E, Spiden S, Gehrig C, Rossier C, DeLozier-Blanchet CD, Blouin JL, Gardiner RM, **Antonarakis SE.** No deleterious mutations in the FOXJ1 (alias HFH4) gene in patients with Primary Ciliary Dyskinesia (PCD). Cytogenetics & Cell Genetics 90: 119-122, 2000.

399-OA Berry A, Scott HS, Kudoh J, Talior I, Korostichevsky M, Wattenhofer M, Guipponi ML, Barras C, Rossier C, Shibuya K, Wang J, Kawasaki K, Asakawa S, Minoshima S, Shimizu N, **Antonarakis SE**, Bonné-Tamir B. Refined localization of autosomal recessive non-syndromic deafness DFNB10 locus using 34 novel microsatellite markers, genomic structure and exclusion of six known genes in the region. Genomics 68: 22-29, 2000.

400-N&V **Antonarakis SE**, McKusick VA. OMIM passes the 1,000-disease-gene mark. Nature Genetics 25: 11, 2000.

401-OA Levinson DF, Holmans P, Straub RE, Owen MJ, Wildenauer DB, Gejman PV, Pulver AE, Laurent C, Kendler KS, Walsh D, Norton N, Williams NM, Schwab SG, Lerer B, Mowry BJ, Sanders AR, **Antonarakis SE**, Blouin JL, DeLeuze JF, Mallet J. Multicenter linkage study of schizophrenia candidate regions on chromosomes 5q, 6q, 10p and 13q: Schizophrenia Linkage Collaborative Group III. American Journal of Human Genetics 67: 652-663, 2000.

402-OA The chromosome 21 mapping and sequencing consortium: Hattori M, Fujiyama A, Taylor TD, Watanabe H, Yada T, Park HS, Toyoda A, Ishii K, Totoki Y, Choi DK, Soeda E, Ohki M, Takagi T, Groner Y, Sakaki Y; Taudien S, Blechschmidt K, Polley A, Menzel U, Delabar J, Kumpf K, Lehmann R, Patterson D, Reichwald K, Rump A, Schillhabel M, Schudy A, Zimmermann W, Rosenthal A; Kudoh J, Shibuya K, Kawasaki K, Asakawa S, Shintani A, Sasaki T, Nagamine K, Mitsuyama S, **Antonarakis SE**, Minoshima S, Shimizu N; Nordsiek G, Hornischer K, Brandt P, Scharfe M, Schön O, Desario A, Reichelt J, Kauer G, Blöcker H; Ramser J, Beck A, Klages S, Hennig S, Riesselmann L, Dagand E, Wehrmeyer S, Borzym K, Gardiner K, Nizetic D, Francis F, Lehrach H, Reinhardt R, Yaspo ML. The DNA sequence of human chromosome 21. Nature 405: 311-319, 2000.

403-OA Guipponi M, Yaspo ML, Riesselman L, Chen H, De Sario A, Roizès G, **Antonarakis SE.** Genomic structure of a copy of the human TPTE gene which encompasses 85 kb on the short arm of chromosome 21. **Human Genetics** 107: 127-131, 2000.

404-R **Antonarakis SE,** Krawczak M, Cooper DN. Disease-causing mutations in the human genome. **European Journal of Pediatrics** 159: S173-S178, 2000.

405-OA Deutsch S, Iseli C, Bucher P, **Antonarakis SE,** Scott HS. A cSNP map and database for human chromosome 21. **Genome Research** 11: 300-307, 2001.

406-OA Marneros A, Mehenni H, Krieg T, **Antonarakis SE,** Olsen BR. Gene for the human transmembrane-type protein tyrosine phosphatase H (PTPRH): genomic structure, fine mapping and its exclusion as a candidate for Peutz-Jeghers syndrome. **Cytogenetics & Cell Genetics** 92: 213-216, 2001.

407-OA Chrast R, Scott HS, Papasavvas MP, Rossier C, Antonarakis ES, Barras C, Davisson MT, Schmidt C, Estivill X, Dierssen M, Pritchard M, **Antonarakis SE.** The mouse brain transcriptome by Serial Analysis of Gene Expression (SAGE): Differences in gene expression between P30 brains of the partial trisomy 16 mouse model of Down syndrome (Ts65Dn) and normals and between males and females. **Genome Research** 10: 2006-2021, 2000.

408-OA Bartoloni L, Wattenhofer M, Kudoh J, Berry A, Shibuya K, Kawasaki K, Wang J, Asakawa S, Talior I, Bonne-Tamir B, Rossier C, Michaud J, McCabe ERB, Minoshima S, Shimizu N, Scott HS, **Antonarakis SE.** Cloning and characterization of a putative human glycerol 3-phosphate permease gene (SLC37A1 or G3PP) on 21q22.3: mutation analysis in 2 candidate phenotypes, DFNB10 and a glycerol kinase deficiency. **Genomics** 70: 190-200, 2000.

409-OA Wattenhofer M, Shibuya K, Kudoh J, Lyle R, Michaud J, Rossier C, Kawasaki K, Asakawa S, Minoshima S, Berry A, Bonne-Tamir B, Shimizu N, **Antonarakis SE,** Scott HS. Isolation and characterization of the UBASH3A gene on 21q22.3 encoding a potential nuclear protein with a novel combination of protein domains: mutation analysis in DFNB10. **Human Genetics** 108: 140-147, 2001.

410-OA Pulver AE, Mulle J, Nestadt G, Blouin JL, Dombroski B, Liang KY, Housman DE, Kazazian HH, **Antonarakis SE,** Lasseter VK, Wolyniec PS, Thornquist MH, McGrath JA. Genetic heterogeneity in schizophrenia: stratification of genome scan data using co-segregating related phenotypes. **Molecular Psychiatry** 5: 650-653, 2000.

411-OA Lamartine J, Munhoz Essenfelder G, Kibar Z, Lanneluc I, Callouet E, Laoudj D, Lemaître G, Hand C, Hayflick SJ, Zonana J, **Antonarakis SE,** Radhakrishna U, Kelsell DP, Christianson AL, Pitaval A, Der Kaloustian V, Fraser C, Blanchet-Bardon C, Rouleau GA, Waksman G. Mutations in the human connexin-30 gene (GJB6) cause hidrotic ectodermal dysplasia (Clouston syndrome). **Nature Genetics** 26: 142-144, 2000.

412-OA Scott HS, Kudoh J, Wattenhofer M, Shibuya K, Berry A, Chrast R, Guipponi M, Wang J, Kawasaki K, Asakawa S, Minoshima S, Younus F, Mehdi SQ, Radhakrishna U, Papasavvas MP, Gehrig C, Rossier C, Korostishevsky M, Gal A, Shimizu N, Bonne-Tamir B, **Antonarakis SE.** Insertion of beta-satellite repeats identifies a transmembrane protease causing both congenital and childhood onset autosomal recessive deafness (DFNB8/10). **Nature Genetics** 27: 59–63, 2001.

413-OA Bartoloni L, Blouin JL, Maiti AK, Sainsbury A, Rossier C, Gehrig C, She JX, Marron MP, Lander ES, Meeks M, Chung E, Armengot M, Jorissen M, Scott HS, Delozier-Blanchet CD, Gardiner RM, **Antonarakis SE.** Axonemal beta heavy chain dynein DNAH9: cDNA sequence, genomic structure and investigation of its role in Primary Ciliary Dyskinesia. **Genomics** 72: 21-33, 2001.

414-BC **Antonarakis SE,** Krawczak M, Cooper DN. Mutations in human diseases; Nature and consequences. Chapter 5 in Rimoin DL, Connor JM, Pyeritz RE, Korf BR, eds. "Emery and Rimoin's Principles and Practice of Medical Genetics" 4th ed Churchill Livingstone. (ISBN 0-443-06434-2), London pp 83-104, 2002.

415-OA Paoloni-Giacobino A, Rossier C, Papasavvas MP, **Antonarakis SE.** Frequency of replication/ transcription errors in (A)/(T) runs of human genes. **Human Genetics** 109: 40-47, 2001.

416-OA Aapola U, Lyle R, Krohn K, **Antonarakis SE,** Peterson P. Isolation and initial characterization of the mouse Dnmt3l gene. **Cytogenetics & Cell Genetics** 92: 122-126, 2001.

417-OA Neerman-Arbez M, deMoerloose P, Honsberger A, Parlier G, Arnuti B, Biron C, Borg JY, Eber S, Meili E, Peter-Salonen K, Ripoll L, Vervel C, d'Oiron R, Staeger P, **Antonarakis SE,** Morris MA. Molecular analysis of the fibrinogen gene cluster in 16 patients with congenital afibrinogenemia: novel truncating mutations in the FGA and FGG genes. **Human**

Genetics 108: 237-240, 2001.

418-OA Attanasio C, deMoerloose P, **Antonarakis SE**, Morris MA, Neerman-Arbez M. Activation of multiple cryptic donor splice sites by the common congenital afibrinogenemia mutation, FGA IVS4+1 G>T. Blood 97: 1879-1881, 2001.

419-OA Ben-Yosef T, Wattenhofer M, Riazuddin S, Ahmed ZM, Scott HS, Kudoh J, **Antonarakis SE**, Naz S, Ahmed Z, Shimizu N, Bonne-Tamir B, Riazuddin S, Pandya A, Nance WE, Wilcox ER, Friedman TB, Morell RJ. Novel mutations of TMPRSS3 in four DFNB8/B10 families segregating congenital autosomal recessive deafness. Journal of Medical Genetics 38: 396-400, 2001.

420-N&V **Antonarakis SE.** BACking up the promises. Nature Genetics 27: 230-232, 2001.

421-R **Antonarakis SE.** Chromosome 21: from sequence to applications. Current Opinion in Genetics & Development 11: 241-246, 2001.

422-R Fokstuen S, Mach F, **Antonarakis SE**, Sztajzel, J. Le conseil génétique en cardiologie: l'exemple de la dysplasie arythmogène du ventricule droit. Médecine et Hygiene 61: 414-418, 2003.

423-R Heino M, Scott HS, Kudoh J, Shimizu N, **Antonarakis SE**, Peterson P, Krohn K. APECED mutations in the Autoimmune Regulator (AIRE) gene. Human Mutation 18: 205-211, 2001.

424-OA Michaud J, Feng W, Osato M, Cottles G, Yanagida M, Asou N, Shigesada K, Ito Y, Benson KF, Raskind WH, Rossier C, **Antonarakis SE**, Israels S, McNichols A, Weiss H, Horwitz M, Scott HS. In vitro analyses of known and novel RUNX1 mutations in dominant familial platelet disorder with predisposition to acute myelogenous leukemia (FPD/AML): implications for mechanisms of pathogenesis. Blood 99: 1364-1372, 2002.

425-OA Masmoudi S, **Antonarakis SE**, Schwede T, Ghorbel AM, Gratri1 M, Pappasavas MP, Drira M, Elgaied-Boulila1 A, Wattenhofer M, Rossier C, Scott HS, Ayadi H, Guipponi ML. Novel missense mutations of TMPRSS3 in two consanguineous Tunisian families with non-syndromic autosomal recessive deafness. Human Mutation 18: 101-108, 2001.

426-CR Pichert G, and Stahel RA for the Swiss Institute for Applied Cancer Research Network for Cancer Predisposition Testing and Counseling. Organizing cancer genetics programs (**Antonarakis SE** among 23 members): the Swiss model. Journal of Clinical Oncology 18 (Suppl): 65-69, 2000.

427-R **Antonarakis SE**, Lyle R, Chrast R, Scott HS. Differential Gene Expression studies to explore the molecular pathophysiology of Down syndrome. Brain Research Reviews 36: 265-274, 2001.

428-OA Glaser B, Mumme DL, Blasey C, Morris MA, Dahoun SP, **Antonarakis SE**, Reiss AL, Eliez S. Language Skills in Children with Velo-cardio-facial Syndrome (Deletion 22q11.2). Journal of Pediatrics 140: 753-758, 2002.

429-OA Soravia C, Delozier-Blanchet C, Blouin JL, Brundler MA, Egger JF, Queneau PE, Roth AD, Marti MC, **Antonarakis SE**, Morel P, Hutter P. Prise en charge multidisciplinaire du cancer colorectal héréditaire. Swiss Surg. 7: 99-104, 2001.

430-Book Engel E, **Antonarakis SE.** Genomic Imprinting and Uniparental Disomy in Medicine; clinical and molecular aspects. J. Wiley Publ. New York. (ISBN 0-471-35126-1) Pgs 304, 2001.

431-R **Antonarakis SE.** The search for allelic variants that cause monogenic disorders or predispose to common, complex, polygenic phenotypes. Dialogues in Clinical Neurosciences 3: 7-16, 2001.

432-OA Guipponi M, Tapparel C, Jousson O, Scamuffa N, Mas C, Rossier C, Hutter P, Meda P, Lyle R, Reymond A, **Antonarakis SE.** The murine orthologue of the Golgi localized TPTE protein provides clues to the evolutionary history of the human TPTE gene family. Human Genetics 109: 569-575, 2001.

433-CR den Dunnen JT, **Antonarakis SE.** Nomenclature for the description of human sequence variations. Human Genetics 109: 121-124, 2001.

434-OA Reymond A, Friedli M, Neergaard-Henrichsen C, Chapot F, Deutsch S, Ucla C, Lyle R, Guipponi M, **Antonarakis SE.** From PREDs and ORFs to cDNA isolation, revisiting the Human Chromosome 21 transcription map. Genomics 78: 46-54, 2001.

435-OA Spitz F, Montavon T, Monso-Hinard C, Morris MA, Ventruto ML, **Antonarakis SE**, Ventruto V, Duboule D. A t(2;8) balanced translocation with breakpoints near the HOXD complex causes mesomelic dysplasia and vertebral defects. **Genomics** 79: 493-498, 2002.

436-OA Wattenhofer M, Di Iorio MV, Rabionet R, Dougherty L, Pampanos A, Schwede T, Montserrat-Sentis B, Arbones ML, Iliades T, Pasquadibisceglie A, D'Amelio M, Alwan S, Rossier C, Dahl HHM, Petersen MB, Estivill X, Gasparini P, Scott HS, **Antonarakis SE.** Mutations in the TMPRSS3 gene are a rare cause of childhood non-syndromic deafness in Caucasian patients. **Journal of Molecular Medicine** 80: 124-131, 2002 (doi: 10.1007/s00109-001-0310-6) [PMID: 11907649] (abstract)

437-OA Hussain NK, Jenna S, Glogauer M, Quinn CC, Wasiak S, Guipponi M, **Antonarakis SE**, Kay BK, Stossel TP, Lamarche-Vane N, McPherson PS. The endocytic protein intersectin-l regulates actin assembly via CDC42 and N-WASP. **Nature Cell Biology** 3: 927-932, 2001.

438-R **Antonarakis SE**, Lyle R, Deutsch S, Reymond A. Chromosome 21: a small land of fascinating disorders with unknown pathophysiology. **International Journal of Developmental Biology** 46: 89-96, 2002.

439-OA Reymond A, Camargo AA, Deutsch S, Stevenson BJ, Parmigiani RB, Ucla C, Bettoni F, Rossier C, Lyle R, Guipponi M, de Souza S, Iseli C, Jongeneel CV, Bucher P, Simpson AJG, **Antonarakis SE.** Characterization of 19 novel transcripts from human chromosome 21. **Genomics** 79: 824-832, 2002.

440-OA Bartoloni L, Blouin JL, Pan Y, Gehrig C, Maiti AK, Scamuffa N, Rossier C, Jorissen M, Armengot M, Meeks M, Mitchison HM, Chung EMK, Delozier-Blanchet CD, Craigen WJ, **Antonarakis SE.** Mutations in the DNAH11 (axonemal heavy chain dynein type 11) gene cause one form of situs inversus totalis and most likely Primary Ciliary Dyskinesia. **Proceedings of the National Academy of Sciences USA** 99: 10282-10286, 2002.

441-OA Chen H, Wang N, Huo Y, Sklar P, MacKinnon DF, Potash JB, McMahon FJ, **Antonarakis SE**, DePaulo JRJr, Ross CA, McInnis MG. Trapping and sequence analysis of 1138 putative exons from human chromosome 18. **Molecular Psychiatry** 8: 619 – 623, 2003.

442-OA Paoloni-Giacobino A, Rey-Berthod C, Couturier A, **Antonarakis SE**, Hutter P. Different rates of frameshift alterations in 4 coding repeat sequences in HNPCC tumors. **Human Genetics** 111: 284-289, 2002.

443-OA Gold G, Blouin JL, Herrmann FR, Michon A, Mulligan R, Duriaux-Saïl G, Bouras C, Giannakopoulos P, **Antonarakis SE.** Specific BACE1 genotypes provide additional risk for late onset Alzheimer disease in APOE4 carriers. **American Journal of Medical Genetics** (Neuropsychiatric Genetics) 119B: 44-47, 2003.

444-OA Guipponi M, Vuagniaux G, Wattenhofer M, Shibuya K, Vazquez M, Dougherty L, Scamuffa N, Guida E, Okui M, Rossier C, Hancock M, Buchet K, Reymond A, Hummler E, Marzella PL, Kudoh J, Shimizu N, Scott HS, **Antonarakis SE***, Rossier BC*. The transmembrane serine protease (TMPRSS3) mutated in deafness DFNB8/10 activates the epithelial sodium channel (EnaC) in vitro. *labs equally contributed. **Human Molecular Genetics** 11: 2829-2836, 2002 (DOI: 10.1093/hmg/11.23.2829) [PMID: 12393794] (abstract).

445-OA Reymond A, Marigo V, Yaylaoglu MB, Leoni A, Ucla C, Scamuffa N, Caccioppoli C, Dermitzakis ET, Lyle R, Banfi S, Eichele G*, **Antonarakis SE***, Ballabio A*. Human chromosome 21 gene expression atlas in the mouse. *labs equally contributed. **Nature** 420: 582–586, 2002 (DOI: 10.1038/nature01178) [PMID: 12466854] (abstract).

446-OA Fokstuen S, **Antonarakis SE**, Blouin JL. FOXL2 mutations in blepharophimosis-ptosis-epicanthus inversus syndrome (BPES); challenges for genetic counseling in female patients. **American Journal of Medical Genetics** 117: 143-146, 2003.

447-OA Lewis CM, Levinson DF, Wise LH, DeLisi LE, Straub RE, Hovatta I, Williams NM, Schwab SG, Pulver AE, Faraone SV, Brzustowicz LM, Kaufman CA, Garver DL, Gurling HMD, Lindholm E, Coon H, Moises HWM, Byerley W, Shaw SH, Mesen A, Sherrington R, O'Neill FA, Walsh D, Kendler KS, Ekelund J, Paunio T, Lönnqvist J, Peltonen L, O'Donovan MC, Owen MJ, Wildenauer DB, Nestadt G, Blouin JL, **Antonarakis SE**, Mowry BJ, Silverman JM, Crowe RR, Cloninger CR, Tsuang MT, Malaspina D, Harkavy Friedman JM, Svrakic DM, Bassett AS, Holcomb J, Kalsi G, McQuillin A, Curtis D, Brynjolfson J, Sigmundsson T, Petursson H, Jazin E, Zoëga T, Helgason T. Genome Scan Meta-Analysis of Schizophrenia and Bipolar Disorder II: Schizophrenia. **American Journal of Human Genetics** 73: 34–48, 2003.

448-BC **Antonarakis SE**, Scott HS. TMPRSS3, a transmembrane protease mutated in congenital and childhood onset autosomal recessive deafness (DFNB10/DFNB8). In "Genetic hearing Loss" Willems P ed., Marcel Decker Publ, (ISBN 0-8247-

4309-1) pp 403-418, 2003.

449-BC **Antonarakis SE**, Krawczak M,. Cooper DN. The nature and mechanisms of human gene mutation. Chapter 2 in Vogelstein B, Kinzler KW editors "The Genetic basis of human cancer" 2nd edition, (ISBN 0-07-137050-1) McGraw-Hill Co. pp. 7-41, 2002.

450-OA Dermitzakis ET, Reymond A, Lyle R, Scamuffa N, Ucla C, Deutsch S, Stevenson BJ, Flegel V, Bucher P, Jongeneel CV, **Antonarakis SE.** Large number of potentially functional but non-genic conserved sequences on human chromosome 21. **Nature** 420: 578-582, 2002.

451-OA Buchet-Poyau K, Mehenni H, Radhakrishna U, **Antonarakis SE.** Search for the second Peutz-Jeghers syndrome locus: exclusion of the STK13, PRKCG, KLK10, and PSCD2 genes on chromosome 19 and the STK11IP gene on chromosome 2. **Cytogenetics & Genome Research** 97: 171-178, 2002.

452-OA Deutsch S, Rideau A, Bochaton-Piallat ML, Merla G, Geinoz A, Gabianni G, Schwede T, Matthes T, **Antonarakis SE,** Beris P. Asp1424Asn MYH9 mutation results in an unstable protein responsible for the phenotypes in May-Hegglin anomaly/Fechtner syndrome. **Blood** 102: 529-534, 2003.

453-OA Guigo R*, Dermitzakis ET*, Agarwal P, Ponting CP, Parra G, Reymond A, Abril JF, Keibler E, Lyle R, Ucla C, **Antonarakis SE,** Brent MR. Comparison of mouse and human genomes followed by experimental verification yields an estimated 1,019 additional genes. *equally contributed. **Proceedings of the National Academy of Sciences USA** 100: 1140-1145, 2003.

454-OA The Mouse Genome Sequencing Consortium; (223 authors including 4 from Medical Genetics in Geneva). Waterston R, Lindblad-Toh K, Birney E, Rogers J, Abril JF, Agarwal P, Agarwala R, Ainscough R, Alexandersson M, An P, **Antonarakis SE,** Attwood J, Baertsch R, Bailey J, Barlow K, Beck S, Berry E, Birren B, Bloom T, Bork P, Botcherby M, Bray N, Brent MR, Brown DB, Bult C, Burton J, Butler J, Campbell RD, Carninci P, Cawley S, Chiaromonte F, Chinwalla A, Church D, Clamp M, Clee C, Collins FS, Cook L, Copley RR, Coulson A, Couronne O, Cuff J, Curwen V, Cutts T, Daly M, David R, Davies J, Delehaunty K, Deri J, Dermitzakis ET, Dewey C, Dickens NJ, Diekhans M, Dodge S, Dubchak I, Dunn DM, Eddy SR, Elnitski L, Emes RD, Eswara P, Eyras E, Felsenfeld A, Fewell G, Flicek P, Foley K, Frankel WN, Fulton L, Fulton R, Furey TS, Gage D, Gibbs RA, Glusman G, Gnerre S, Goldman N, Goodstadt L, Graffham D, Graves T, Green ED, Gregory S, Guigó R, Guyer M, Hardison RC, Haussler D, Hayashizaki Y, Hillier L, Hinrichs A, Hlavina W, Holzer T, Hsu F, Hua A, Hubbard T, Hunt A, Jackson I, Jaffe DB, Johnson LS, Jones M, Jones TA, Joy A, Kamal M, Karlsson EK, Karolchik D, Kasprzyk A, Kawai J, Keibler E, Kells C, Kent WJ, Kirby A, Kolbe D, Korf I, Kucherlapati RS, Kulbokas EJ III, Kulp D, Landers T, Leger JP, Leonard S, Letunic I, Levine R, Li J, Li M, Lloyd C, Lucas S, Ma B, Maglott DR, Maier J, Mardis ER, Matthews L, Evan Mauceli E, Mayer JH, McCarthy M, McCombie R, McLaren S, McLay K, McPherson J, Meldrim J, Meredith B, Mesirov JP, Miller W, Miner T, Mongin E, Montgomery KT, Morgan M, Mott R, Mullikin JC, Muzny DM, Nash W, Nelson J, Nhan M, Nicol R, Ning Z, Nusbaum C, O'Connor MJ, Okazaki Y, Oliver K, Overton-Larty E, Pachter L, Parra G, Pepin K, Peterson J, Pezvner P, Plumb R, Pohl C, Poliakov A, Ponce T, Ponting C, Potter S, Quail M, Reymond A, Roe BA, Roskin KM, Rubin E, Rust AG, Santos R, Sapojnikov V, Schultz B, Schultz J, Schwartz MS, Schwartz S, Scott C, Seaman S, Searle S, Sharpe T, Sheridan A, Shownkeen R, Sims S, Singer JB, Slater G, Smit A, Smith DR, Spencer B, Stabenau A, Stange-Thomann N, Sugnet C, Suyama M, Tesler G, Thompson J, Torrents D, Trevaskis E, Tromp J, Ucla C, Ureta-Vidal A, Vinson JP, von Niederhausern AC, Wade CM, Wall M, Weber RJ, Weiss RB, Wendl M, West T, Wetterstrand K, Wheeler R, Whelan S, Wierzbowski J, Willey T, Williams S, Wilson R, Winter E, Worley KC, Wyman D, Yang S, Yang SP, Zdobnov E, Zody MC, Lander ES. Initial sequencing and comparative analysis of the mouse genome. **Nature** 420: 520–562, 2002.

455-OA Segurado R, Detera-Wadleigh SD, Levinson DF, Lewis CM, Gill M, Nurnberger JI, Craddock N, DePaulo JR, Baron M, Gershon ES, Ekholm J, Cichon S, Turecki G, Claes S, Kelsoe JR, Schofield PR, Badenhop RF, Morissette J, Coon H, Blackwood D, Curtis D, McInnes LA, Foroud T, Edenberg H, Reich T, Rice J, GOAte A, McInnis M, McMahon FJ, Badner JA, Goldin LR, Bennett P, Willour V, Zandi P, Liu J, Gilliam C, Juo SH, Berrettini WH, Yoshikawa T, Peltonen L, Lönnqvist J, Nöthen MM, Schumacher J, Windemuth C, Rietschel M, Propping P, Alda M, Grof P, Rouleau GA, Del-Favero J, VanBroeckhoven C, Mendlewicz J, Adolfsson R, Spence MA, Luebbert H, Adams LJ, Donald JA, Mitchell PB, Barden N, Shink E, Byerley W, Muir W, Visscher P, Macgregor S, Gurling H, Kalsi G, McQuillan A, Escamilla MA, Reus VI, Leon P, Freimer NB, Ewald H, Kruse TA, Mors O, Radhakrishna U, Blouin JL, **Antonarakis SE,** Akarsu N. Genome Scan Meta-Analysis of Schizophrenia and Bipolar Disorder Part III: Bipolar Disorder. **American Journal of Human Genetics** 73: 49–62, 2003.

456-OA Menzel O, Bekkeheien RCJ, Reymond A, Fukai N, Boye E, Kosztolanyi G, Aftimos S, Deutsch S, Gehrig C, Scott HS, Olsen BR, **Antonarakis SE,** Guipponi M. Knobloch syndrome: novel mutations in COL18A1, evidence for genetic heterogeneity, and a functionally impaired polymorphism in endostatin. **Human Mutation** 23: 77-84, 2004.

457-OA Hoda JC, Gu WL, Friedli M, Phillips HA, Bertrand S, **Antonarakis SE**, Goudie D, Roberts R, Scheffer IE, Marini C, Patel J, Berkovic SF, Mulley JC, Steinlein OK, Bertrand D. Human nocturnal frontal lobe epilepsy: pharmocogenomic profiles of new nAChR mutations outside the ion channel pore. **Molecular Pharmacology** 74: 379-391, 2008 [Epub May 2, 2008] [PMID: 18456869] [IF 4.47] (abstract).

458-BC **Antonarakis SE**, Cooper DN. Mutations in Human Genetic Disease: Nature and Consequences. In Cooper DN et al, Encyclopedia of the Human Genome (ISBN 0-333-80386-8). Nature Publishing Group, London. Vol4, pp 227-253, 2003.

459-BC **Antonarakis SE.** CpG dinucleotides and human disorders. In Cooper DN et al, Encyclopedia of the Human Genome (ISBN 0-333-80386-8). Nature Publishing Group, London. Vol1, pp 950-956, 2003.

460-BC den Dunnen JT, **Antonarakis SE.** Mutation nomenclature. In Current Protocols in Human Genetics; John Wiley Press, Madison, Supplement 37; 7.13.1-9, 2003 [PMID: 18428344].

461-OA Menzel O, Vellai T, Takacs-Vellai K, Reymond A, Mueller F, **Antonarakis SE**, Guipponi M. Disruption and overexpression of K10G9.3 the Caenorhabditis elegans ortholog of C21orf80, results in embryonic patterning defects and affects morphogenesis, respectively. **Genomics** 84: 320-330, 2004.

462-BC Deutsch S, **Antonarakis SE**, Reymond A. Disorders of Chromosome 21. in "Encyclopedic Reference of Genomics and Proteomics in Molecular Medicine", Ganten D & Ruckpaul K eds, Springer Verlag, Berlin. Pgs. 280-287, 2006.

463-R Lalioti MD, **Antonarakis SE**, Scott HS. The epilepsy, the protease inhibitor and the dodecamer: Progressive Myoclonus Epilepsy, Cystatin B and a 12-mer Repeat Expansion. **Cytogenetics & Genome Research** 100: 213-223, 2003.

464-OA Ferrari SL*, Deutsch S*, Choudhury U, Chevalley T, Bonjour JP, Dermitzakis ET, Rizzoli R, **Antonarakis SE.** Polymorphisms in the Low-Density Lipoprotein Receptor-Related Protein 5 (LRP5) Gene Are Associated with Variation in Vertebral Bone Mass, Vertebral Bone Size, and Stature in Whites. *authors equally contributed. **American Journal of Human Genetics** 74: 866-875, 2004 (Epub Apr 7, 2004).

465-R Bartoloni L, **Antonarakis SE.** The human sugar-phosphate/phosphate exchanger family SLC37. Europ. J. Physiol (Pflügers Archiv): Special Issue, "The ABC of Solute Carriers: Physiological, Pathological and Therapeutic Implications of Human Membrane Transport Proteins"; ISSN: 0031-6768; Hediger M ed. 447: 780-783, 2004 (Epub Jun 17, 2003).

466-OA Pruitt WM, Karnoub AE, Rakauskas AC, Guipponi M, **Antonarakis SE**, Kurakin A, Kay BK, Sondek J, Siderovski DP, Der CJ. Role of the pleckstrin homology domain in Intersectin-L Dbl homology domain activation of Cdc42 and signaling. **Biochimical & Biophysical Acta.** (BBA-MCR) 1640: 61-68, 2003.

467-OA Mehenni H, Lin-Marq N, Buchet-Poyau K, Reymond A, Collart MA, Picard D, **Antonarakis SE.** LKB1 interacts with and phosphorylates PTEN, a functional link between two proteins involved in cancer predisposing syndromes. **Human Molecular Genetics** 14: 2209-2219, 2005, [Epub Jun 29, 2005] [IF 8.597].

468-OA Friedli M, Guipponi ML, Bertrand S, Bertrand D, Neerman-Arbez M, Scott HS, **Antonarakis SE**, Reymond A. Identification of a novel member of the CLIC family, CLIC6, mapping to 21q22.12. **Gene** 320: 31-40, 2003.

469-OA Dermitzakis ET, Reymond A, Scamuffa N, Ucla C, Kirkness E, Rossier C, **Antonarakis SE.** Evolutionary Discrimination of Mammalian Conserved Non-Genic Sequences (CNGs). **Science** 302: 1033-1035, 2003 (Epub Oct 2, 2003).

470-OA Tapparel C, Reymond A, Girardet C, Guillou L, Lyle R, Lamon C, Hutter P, **Antonarakis SE.** The TPTE gene family: cellular expression, subcellular localization and alternative splicing. **Gene** 323: 189-199, 2003 [PMID: 14659893] (abstract).

471-BC **Antonarakis SE,** Mauron A. Living with DNA. In "50 years of DNA"-Commemorative issue book of Business Weekly and The Wellcome Trust, London UK, pgs 46-47, 2003.

472-OA de Mollerat XJ, Gurrieri F, Morgan CT, Sangiorgi E, Everman DB, Gaspari P, Amiel J, Bamshad MJ, Lyle R, Blouin JL, Allanson JE, Le Marec B, Wilson M, Braverman NE, Radhakrishna U, Delozier-Blanchet C, Abbott A, Elghouzzi V, **Antonarakis SE**, Stevenson RE, Munnich A, Neri G, Schwartz CE. A Genomic Rearrangement Resulting in a Tandem Duplication is Associated with Split Hand-Split Foot Malformation 3 (SHFM3) at 10q24. **Human Molecular Genetics** 12: 1959-1971, 2003.

473-OA Weinhäusel A, Morris MA, **Antonarakis SE**, Haas OA. DNA deamination enables direct PCR amplification of the cystatin B gene-associated dodecamer repeat expansion in myoclonus epilepsy type Unverricht-Lundborg (EPM1). **Human**

Mutation 22: 404-408, 2003.

474-OA Lyle R, Gehrig C, Neergaard-Henrichsen C, Deutsch S, **Antonarakis SE.** Gene expression from the aneuploid chromosome in a segmental trisomy mouse model of Down syndrome. Genome Research 14: 1268-1274, 2004.

475-OA Dayer AG, Bottani A, Bouchardy I, Fluss J, **Antonarakis SE,** Haenggeli CA, Morris MA. MECP2 mutant allele in a boy with Rett syndrome and his unaffected heterozygous mother. Brain Development 29: 47-50, 2007 [Epub Jul 14, 2006] [IF 1.38].

476-OA **Antonarakis SE,** Reymond A, Lyle R, Deutsch S, Dermitzakis MT. Chromosome 21 and Down syndrome; the post sequence era. Cold Spring Harbor Symposia on Quantitative Biology LXVIII: 425-430, 2004.

477-OA Dermitzakis ET, Kirkness E, Schwarz S, Birney E, Reymond A, **Antonarakis SE.** Comparison of human chromosome 21 Conserved Nongenic sequences (CNGs) with the mouse and dog genomes shows that their selective constraint is independent of their genic environment. Genome Research 14: 852-859, 2004 (Epub Apr 12, 2004) [IF 13.59].

478-Book Cooper DN editor-in-chief, and Thomas NS, **Antonarakis SE,** Binz PA, Clarke A, Dear P, Gardiner K, Gottesman I section editors. Nature Encyclopedia of the Human Genome (ISBN 0-333-80386-8). Volumes 1-5. Nature Publishing company, London. 2003.

479-BC Kaufman RJ, **Antonarakis SE.** Structure, biology and genetics of Factor VIII. Chapter 114 in Hoffman R, Benz EJ, Shattil SJ, Furie B, Cohen HJ, Silberstein LE and McGlave P eds. "Hematology; Basic Principles and Practice". 4th edition (ISBN 0-443-06628-0). Churchill Livingston, New York. pp 2011-2030, 2005.

480-OA Lin-Marq N, Borel C, **Antonarakis SE.** Peutz-Jeghers LKB1 mutants fail to activate GSK-3beta preventing LKB1 to inhibit Wnt signaling. Molecular Genetics & Genomics 273: 184–196, 2005; [Epub Feb 25, 2005)].

481-OA Mas C, Taske N, Deutsch S, Guipponi M, Thomas P, Covanis T, Friis M, Kjeldsen MJ, Pizzolato GP, Villemure JG, Buresi C, Rees M, Malafosse A, Gardiner M, **Antonarakis SE,** Meda P. Association of the Connexin36 Gene (CX36) with Juvenile Myoclonic Epilepsy Journal of Medical Genetics 41: E93, 2004.

482-OA Sheth FJ, Radhakrishna U, Morris MA, Blouin JL, Sheth JJ, Multani A, **Antonarakis SE.** Cytogenetic, molecular and FISH analysis of an isodicentric chromosome 21 idic(21)(q22.3) in a mildly affected patient with Down syndrome. International Journal of Human Genetics 7: 215-218, 2007.

483-OA Landau R, Cahana A, Smiley RM, **Antonarakis SE,** Blouin JL. Genetic variability of the m-opioid receptor gene in an obstetric population. Anesthesiology 100: 1030-1033, 2004.

484-OA Dauvilliers Y, Blouin JL, Neidhart E, Carlander B, Eliaou JF, **Antonarakis SE,** Billiard M, Tafti M. A narcolepsy susceptibility locus maps to a 5Mb region of chromosome 21. Annals of Neurology 56: 382-388, 2004.

485-OA Neergaard-Henrichsen C, Delorme R, Boucherie M, Marelli D, Baud P, Bellivier F, Courtet P, Nadia Chabane N, Henry C, Leboyer M, Malafosse A, **Antonarakis SE,** Dahoun S. No Association between DUP25 and Anxiety Disorders. American Journal of Medical Genetics (Neuropsychiatric Genetics) 128B: 80-83, 2004.

486-OA Webster KE, O'Bryan MK, Fletcher S, Crewther PE, Aapola U, Craig J, Harrison DK, Aung H, Phutikanit N, Lyle R, Meachem SJ, **Antonarakis SE,** De Kretser DM, Hedger MP, Peterson P, Carroll BJ, Scott HS. Meiotic and epigenetic modification defects in Dnmt3L knockout mouse spermatogenesis. Proceedings of the National Academy of Sciences USA 102: 4068-4073, 2005 [Epub Mar 7, 2005].

487-R Dermitzakis ET, Reymond A, **Antonarakis SE.** Conserved Non-Genic sequences – an unexpected feature of mammalian genomes. Nature Reviews Genetics 6: 151-157, 2005.

488-OA Merla G, Howald C, **Antonarakis SE,** Reymond A. The subcellular localization of the ChoRE binding protein, encoded by the Williams-Beuren syndrome critical region gene 14, is regulated by 14-3-3. Human Molecular Genetics 13: 1505-1514, 2004. [Epub May 26, 2004] [PMID: 15163635] (abstract).

489-OA Deutsch S, Choudhury U, Merla G, Howald C, Sylvan A, **Antonarakis SE.** Detection of aneuploidies by paralogous sequence quantification. Journal of Medical Genetics 41: 908-915, 2004.

490-R **Antonarakis SE**, Lyle R, Dermitzakis ET, Reymond A, Deutsch S. Chromosome 21 and Down syndrome; from genomics to pathophysiology. **Nature Reviews Genetics** 5: 725-738, 2004.

491-OA Bonafé L, Dermitzakis ET, Unger S, Greenberg C, Xavier B, Zankl A, **Antonarakis SE**, Superti-Furga A, Reymond A. The RMRP gene and Cartilage-Hair Hypoplasia: mutation review, haplotyping, and comparative genomics. **PLoS Genetics** 1: 1 444-454, 2005, e47 [Epub Oct 21, 2005] [PMID: 16244706] [IF 7.67] (abstract).

492-OA Valiente M, Gomar B, Torres J, Tapparel C, **Antonarakis SE**, Pulido R. PTEN binds multiple PDZ domains through distinct regions at its C-terminal tail: a role of PTEN/PDZ-domain binding in PTEN stability and function. **Journal of Biological Chemistry (JBC)** 280: 28936-28944, 2005 [Epub Jun 10, 2005] [IF 6.35].

493-OA The International Chicken Genome Sequencing Consortium; (173 authors including 3 from Medical Genetics in Geneva). Hillier LW, Miller W, Birney E, Warren W, Hardison RC, Ponting C, Bork P, Burt DW, Groenen MAM, Delany ME, Dodgson JB, Chinwalla AT, Cliften PF, Clifton SW, Delehaunty KD, Fronick C, Fulton RS, Graves TA, Kremitzki C, Layman D, Magrini V, McPherson JD, Miner TL, Minx P, Nash WE, Nhan MN, Nelson JO, Oddy LG, Pohl CS, Randall-Maher J, Smith SM, Wallis JW, Yang SP, Romanov MN, Rondelli CM, Paton B, Smith J, Morrice D, Daniels L, Tempest HG, Robertson L, Masabanda JS, Griffin DK, Vignal A, Fillon V, Jacobbson L, Kerje S, Andersson L, Crooijmans RPM, Aerts J, van der Poel JJ, Ellegren H, Caldwell RB, Hubbard SJ, Grafham DV, Kierzek AM, McLaren SR, Overton IM, Arakawa H, Beattie KJ, Bezzubov Y, Boardman PE, Bonfield LK, Croning MDR, Davies RM, Francis MD, Humphray SJ, Scott CE, Taylor RG, Tickle C, Brown WRA, Rogers J, Buerstedde JM, Wilson SA, Stubbs L, Ovcharenko I, Gordon L, Lucas S, Miller MM, Inoko H, Shiina T, Kaufman J, Salomonsen J, Skjoedt K, Wong GKS, Wang J, Liu B, Wang J, Yu J, Yang H, Nefedov M, Koriabine M, deJong PJ, Goodstadt L, Webber C, Dickens NJ, Letunic I, Suyama M, Torrents D, Mering C, Zdobnov E, Makova K, Nekrutenko A, Elnitski L, Eswara P, King DC, Yang S, Tyekucheva S, Radakrishnan A, Harris RS, Chiaromonte F, Taylor J, He J, Rijnkels M, Griffiths-Jones S, Ureta-Vidal A, Hoffman MM, Severin J, Searle SMJ, Law AS, Speed D, Waddington D, Cheng Z, Tuzun E, Eichler E, Bao Z, Flicek P, Shteynberg DD, Brent MR, Bye JM, Huckle EJ, Chatterji S, Dewey C, Pachter L, Kouranov A, Mourelatos Z, Hatzigeorgiou AG, Paterson AH, Ivarie R, Brandstrom M, Axelsson E, Backstrom N, Berlin S, Webster MT, Pourquie O, Reymond A, Ucla C, **Antonarakis SE**, Long M, Emerson JJ, Betran E, Dupanloup I, Kaessmann H, Hinrichs AS, Bejerano G, Furey TS, Harte RA, Raney B, Siepel A, Kent WJ, Haussler D, Eyras E, Castelo R, Abril JF, Castellano S, Camara F, Guigo R, Bourque G, Tesler G, Pevzner PA, Smit A, Fulton LA, Mardis ER, Wilson RK. Sequencing and comparative analysis of the chicken genome provide unique perspectives on vertebrate evolution.
Nature 432: 695-716, 2004 [IF 26.68].

494-OA Eyras E, Reymond A, Castelo R, Bye JM, Camara F, Flicek P, Huckle EJ, Parra G, Shteynberg DD, Wyss C, Rogers J, **Antonarakis SE**, Birney E, Guigo R, Brent MR. Gene finding in the chicken genome. **BMC Bioinformatics** 6: 131, 2005 (doi:10.1186/1471-2105-6-131) [IF 5.42].

495-OA Wattenhofer M, Reymond A, Falciola V, Charollais A, Caille D, Borel C, Lyle R, Estivill X, Petersen MB, Meda P, **Antonarakis SE.** Different mechanisms preclude mutant CLDN14 proteins from forming tight junctions in vitro. **Human Mutation** 25: 543–549, 2005.

496-OA The ENCODE Project Consortium. Feingold EA, Good PJ, Guyer M, Kamholz S, Liefer L, Wetterstrand K, Collins FS, Gingeras TR, Kampa D, Sekinger EA, Cheng J, Hirsch H, Ghosh S, Zhu Z, Patel S, Piccolboni A, Yang A, Tammana H, Bekiranov S, Kapranov P, Harrison R, Church G, Struhl K, Ren B, Kim TH, Barrera LO, Qu C, Van Calcar S, Luna R, Glass CK, Rosenfeld MG, Guigo R, **Antonarakis SE**, Birney E, Brent M, Patcher L, Reymond A, Dermitzakis E, Dewey C, Keefe D, Denoeud F, Lagarde J, Ashurst J, Hubbard T, Wesselink JJ, Castelo R, Eyras E, Myers RM, Sidow A, Batzoglou S, Trinklein ND, Hartman SJ, Aldred SF, Anton E, Schroeder DI, Marticke SS, Nguyen L, Schmutz J, Grimwood J, Dickson M, Cooper GM, Stone EA, Asimenos G, Brudno M, Dutta A, Karnani N, Taylor CM, Kim HK, Robins G, Stamatoyannopoulos G, Stamatoyannopoulos JA, Dorschner M, Sabo P, Hawrylycz M, Humbert R, Wallace J, Yu M, Navas P, Olson M, McArthur M, Noble WS, Dunham I, Koch CM, Andrews RM, Clelland GK, Wilcox S, Fowler JM, James KD, P. Groth P, Dovey OM, Ellis PD, Wraight VL, Mungall AJ, Dhami P, Fiegler H, Langford CF, Carter NP, Vetrie D, Snyder M, Euskirchen G, Urban A, Nagalakshmi U, Rinn J, Popescu G, Bertone P, Rozowsky J, Emanuelsson O, Royce T, Chung S, Gerstein M, Lian Z, Lian L, Nakayama Y, Weissman S, Stolc V, Tongprasit W, Sethi H, Jones S, Marra M, Shin H, Schein J, Clamp M, Lindblad-Toh K, Chang J, Jaffe DB, Kamal M, Lander ES, Mikkelson T, Vinson J, Zody MC, de Jong PJ, Osoegawa K, Nefedov M, Zhu B, Baxevanis AD, Wolfsberg TG, Crawford GE, Green ED, Bouffard GG, Margulies EH, Portnoy ME, Hansen NF, Thomas PJ, McDowell JC, Maskeri B, Young AC, Idol JR, Blakesley RW, Schuler G, Miller W, Hardison R, Elnitski L, Shah P, Salzberg SL, Pertea M, Majoros WH, Haussler D, Thomas D, Rosenbloom K, Clawson H, Siepel A, Kent WJ, Weng Z, Jin S, Halees A, Burden H, Karaoz U, Fu Y, Yu Y, Ding C, Cantor CR, Kingston RE, Dennis J, Green RD, Singer MA, Richmond TA, Norton JE, Farnham PJ, Oberley MJ, Inman DR, McCormick MR, Kim H, Middle CL, Pirrung MC, Fu XD, Kwon YS, Ye Z, Dekker J, Tabuchi TM, Gheldof N, Dostie J, Harvey SC. The ENCODE (ENCyclopedia Of DNA Elements) Project. **Science** 306: 636-640, 2004 [PMID: 15499007].

497-OA Dauphinot L, Lyle R, Rivals I, Tran Dang M, Moldrich R, Golfier G, Ettwiller L, Toyama K, Rossier J, Personnaz L, **Antonarakis SE**, Epstein CJ, Sinet PM, Potier MC. Cerebellum transcriptome during postnatal development of the Ts1Cje mice, a partial model for Down syndrome. **Human Molecular Genetics** 14: 373-384, 2005 [Epub Dec 8, 2004] [IF 8.60].

498-OA Merla G, Howald C, Lyle R, Wyss C, Zabot MT, **Antonarakis SE**, Reymond A. Large genomic deletions influence expression levels of the non-hemizygous flanking genes. **American Journal of Human Genetics** 79: 332-341, 2006 [IF 12.65].

499-OA Ferrari SL, Deutsch S, Baudoin C, Cohen-Solal M, Ostertag A, **Antonarakis SE**, Rizzoli R, de Vernejoul MC. LRP5 gene polymorphisms and idiopathic osteoporosis in men. **Bone** 37: 770-775, 2005. [Epub Sep 14, 2005][IF 3.57].

500-OA Glaser B, Debbane M, Hinard C, Morris MA, Dahoun SP, **Antonarakis SE**, Eliez S. No evidence for an effect of COMT Val158Met genotype on executive function in patients with 22q11 Deletion Syndrome (22q11.2DS). **American Journal of Psychiatry** 163: 537-539, 2006.

501-OA Wattenhofer M, Sahin-Calapoglu N, Andreasen D, Kalay E, Caylan R, Braillard B, Fowler-Jaeger N, Reymond A, Rossier BC, Karaguzel A, **Antonarakis SE.** A novel TMPRSS3 missense mutation in a DFNB8/10 family prevents proteolytic activation of the protein. **Human Genetics** 117: 528-35, 2005. [Epub Jul 14, 2005].

502-OA Castelo R, Reymond A, Wyss C, Camara F, Parra G, **Antonarakis SE**, Guigo R, Eyras E. Comparative gene finding in chicken indicates that we are closing in on the human gene set of multi-exonic widely-expressed human genes. **Nucleic Acids Research** 33: 1935-1939, 2005 [Epub Apr 4, 2005][IF 7.26].

503-OA Parra G, Reymond A, Dabbouseh N, Dermitzakis ET, Castelo R, Thomson TM, **Antonarakis SE**, Guigo R. Tandem chimerism as a means to increase protein diversity in the human genome. **Genome Research** 16: 37-44, 2006 [Epub Dec 12, 2005] [IF 13.59].

504-R Ferrari SL, Deutsch S, **Antonarakis SE.** Pathogenic mutations and polymorphisms in the lipoprotein receptor-Related protein 5 reveal a new biological pathway for the control of bone mass. **Current Opinion Lipidology** 16: 207-214, 2005 [IF 6.23].

505-R **Antonarakis SE**, Epstein CJ. The challenge of Down syndrome. **Trends Molecular Medicine.** 12: 473-479, 2006 [Epub Aug 24, 2006] [IF 5.50].

506-CR Gardiner K, Davisson MT, Pritchard M, Patterson D, Groner Y, Crnic LS, **Antonarakis SE**, Mobley W. Report on the 'Expert Workshop on the Biology of Chromosome 21: towards gene-phenotype correlations in Down syndrome', held June 11–14, 2004, Washington D.C. **Cytogenetics & Genome Research** 108: 269–277, 2005.

507-BC Kaufman RJ, **Antonarakis SE**, Fay PJ. Factor VIII and Hemophilia A. in Colman RW, Hirsh J, Marder VJ, Clowes AW, George JN eds. "Hemostasis and Thrombosis: Basic principles and clinical practice" 5th edition, chapter 8, Lippincott, Williams & Wilkins, Philadelphia, PA. 2007.

508-OA Howald C, Merla G, Digilio MC, Amenta S, Lyle R, Deutsch S, Choudhury U, Bottani A, **Antonarakis SE**, Fryssira H, Dallapiccola B, Reymond A. Two high-throughput technologies to detect segmental aneuploidies identify new Williams-Beuren Syndrome patients with atypical deletions. **Journal of Medical Genetics** 43: 266-273, 2006 [Epub Jul 1, 2005][IF 6.37].

509-OA Gothelf D, Eliez S, Thompson T, Hinard C, Penniman L, Feinstein C, Kwon H, Jin S, Jo B, **Antonarakis SE**, Morris MA, Reiss AL. COMT genotype predicts longitudinal cognitive decline and psychosis in 22q11.2 deletion syndrome. **Nature Neuroscience** 8: 1500-1502, 2005. [Epub Oct 23, 2005] [IF 16.98].

510-OA Deutsch S, Lyle R, Dermitzakis ET, Attar H, Subrahmanyan L, Gehrig C, Parand L, Gagnebin M, Rougemont J, Jongeneel CV, **Antonarakis SE.** Gene Expression variation and expression quantitative trait (eQTL) mapping of human chromosome 21 genes. **Human Molecular Genetics** 14: 3741-3749, 2005.

511-BC Dermitzakis ET, **Antonarakis SE.** The biological significance of Conserved Non-Genic DNA. In Adams M ed. "Comparative Genomics" book chapter. 2007.

512-OA Leupin O, Attanasio A, Marguerat S, Tapernoux M, **Antonarakis SE**, Conrad B. Transcriptional activation via bidirectional RNA polymerase II elongation over a silent promoter. **EMBO Reports** 6: 956-960, 2005. [Epub Aug 12, 2005] [IF 7.55].

513-OA Loeuillet C*, Weale M*, Deutsch S*, Soranzo N, Wyniger J, Lettre G, Dupré Y, Thuillard D, Beckmann J, **Antonarakis SE**, Goldstein DB, Telenti A. Promoter polymorphisms and allelic imbalance in ABCB1 expression. *equally contributed **Pharmacogenetics & Genomics** 17: 951-959, 2007 [PMID: 18075465] [IF 5.39].

514-R **Antonarakis SE**, Beckmann JS. Mendelian disorders deserve more attention. **Nature Reviews Genetics** 7: 277-282, 2006 [Epub Mar 14, 2006].

514a-BC **Antonarakis SE**, Beckmann JS. Mendelian disorders deserve more attention. In Fifty years of Human Genetics; Festschrift for George Fraser. O Mayo and C Leach eds. Wakefield press, South Australia. (ISBN 9781862547537) pgs 188-194, 2007.

515-OA Stranger BE, Forrest MS, Clark AG, Deutsch S, Lyle R, Hunt S, Kahl B, **Antonarakis SE**, Tavaré S, Deloukas P, Dermitzakis ET. Genome-wide associations of gene expression variation in humans. **PLOS Genetics** 1:e78, 2006 [Epub Dec 16, 2005] [IF 7.67].

516-OA Drake JA, Bird C, Nemesh J, Thomas D, Newton-Cheh C, Reymond A, Excoffier L, Attar H, **Antonarakis SE**, Dermitzakis ET, Hirschhorn JN. Conserved non-coding sequences are selectively constrained and not mutation cold spots. **Nature Genetics** 38: 223-227, 2006 [Epub Dec 25, 2005] [IF 25.55].

517-OA Landau R, Morales MA, **Antonarakis SE**, Blouin JL, Smiley RM. Arg16 homozygosity of the beta2-adrenoceptor improves outcome from beta2-agonist tocolysis for preterm labor. **Clinical Pharmacology & Therapy** 78: 656-663, 2005. [IF 6.82].

518-BC Lalioti MD, **Antonarakis SE**, Scott HS. Dodecamer repeat expansion in Progressive Myoclonus Epilepsy 1 (EPM1). In Fry M, Usdin K "Human Nucleotide Expansion Disorders" in Vol 19 of Nucleic Acids and Molecular Biology series ed HJ Gross Springer-Verlag, Berlin, Heidelberg; (ISBN 10-3-540-33335-5) pgs 121-141, 2006.

519-OA Everman DB, Morgan CT, Lyle R, Laughridge ME, Bamshad MJ, Clarkson KB, Colby R, Gurrieri F, Innes M, Roberson J, Schrander-Stumpel C, van Bokhoven H, **Antonarakis SE**, Schwartz CE. Frequency of genomic rearrangements involving the SHFM3 locus at chromosome 10q24 in syndromic and nonsyndromic split-hand/foot malformation. **American Journal of Medical Genetics** 140: 1375-1383, 2006 [Epub Jun 7, 2006].

520-OA Lyle R, Radhakrishna U, Blouin JL, Gagos S, Everman D, Gehrig C, Delozier-Blanchet C, Solanki JV, Patel UC, Nath SK, Gurrieri F, Neri G, Schwartz C, **Antonarakis SE.** Split hand-split foot malformation 3 (SHFM3) at 10q24, development of rapid diagnostic methods and gene expression from the region. **American Journal of Medical Genetics** 140:1384-1395, 2006 [Epub May 11, 2006].

521-OA Canzonetta C, Mulligan C, Deutsch S, Ruf S, O'Doherty A, Lyle R, Borel C, Lin-Marq N, Delom F, Groet J, Schnappauf F, De Vita S, Averill JV, Priestley J, Martin JE, Shipley J, Denyer G, Epstein CJ, Fillat C, Estivill X, Tybulewicz V, Fisher EM, **Antonarakis SE**, Nizetic D. DYRK1A-dosage imbalance perturbs NRSF/REST levels, deregulating pluripotency and embryonic stem cell fate in Down syndrome. **American Journal of Human Genetics** 83: 388-400, 2008 [Epub Sep 3, 2008] [PMID: 18771760] (abstract)

522-N&V BioSapiens Network Excellence: Thornton J, Tramontano A, Roma U, Valencia A, Brunak R, **Antonarakis SE**, Bork P, Casadio R, Danchin A, Durbin R, Frishman D, Guigo R, von Heijne G, van Helden J, Lengauer T, Linial M, Mott R, Orengo C, Jones D, Patthy L, Rychlewski L, Schachter V, Schomburg D, Ukkonen E, Veuthey AL, Vingron M, Vriend G, Nyberg K. BioSapiens: a European network for integrated genome annotation. **European Journal of Human Genetics** 13: 994-997, 2005.

523-OA Harrow J, Denoeud F, Frankish A, Reymond A, Chen CK, Chrast J, Lagarde J, Gilbert JGR, Storey R, Swarbreck D, Rossier C, Ucla C, Hubbard T, **Antonarakis SE**, Guigo R. GENCODE: Producing a reference annotation for ENCODE. **Genome Biology** Suppl 1:S4 pgs 1-9 (doi: 10.1186/gb-2006-7-s1-s4) [Epub Aug 7, 2006] [IF 9.71].

524-BC **Antonarakis SE.** Genome and Postgenome; present and future; Subtitle: Human Genome Variability and Disorders. Vatican book on the Human Genome. ed. Cardinal J Lozano Barragan 2008.

525-OA Gothelf D, Maeda F, C Hinard C, Hallmayer JF, **Antonarakis SE**, Morris MA, Reiss AL. Effect of Hemizygous COMT Genotypes on Cortical Activation Associated with Response Inhibition in 22q11.2 Deletion Syndrome. **Human Brain Mapping** 28: 533-542, 2007 [Epub Apr 10, 2007] [PMID: 17427209] [IF 4.31].

526-OA Guigó R, Flicek P, Abril JF, Reymond A, Lagarde J, Denoeud F, **Antonarakis SE**, Ashburner M, Bajic VB, Birney E, Castelo R, Eyras E, Gingeras T, Good P, Harrow J, Hubbard T, Lewis S, Ucla C, Reese MG. EGASP: The human ENCODE genome annotation assessment project. **Genome Biology** Suppl 1:S2 pgs 1-31 [Epub Aug 7, 2006] [IF 9.71].

527-BC **Antonarakis SE**, Cooper DN. Mutations in human genetic disease; Nature and consequences. Chapter 7 in Rimoin DL, Connor JM, Pyeritz RE, Korf BR, eds. "Emery and Rimoin's Principles and Practice of Medical Genetics" 5th ed (ISBN 0-443-06890-9) Churchill Livingstone. Pgs. 101-128, 2006.

528-OA Hornef N, Olbrich H, Horvath J, Zariwala MA, Fliegauf M, Tomas-Loges N, Wildhaber J, Noone PG, Kennedy M, **Antonarakis SE**, Blouin JL, Bartoloni L, Nüßlein T, Ahrens P, Griese M, Kuhl H, Sudbrak R, Knowles MR, Reinhardt R, Omran H. DNAH5 mutations are a common cause of Primary Ciliary Dyskinesia. **American Journal of Respiratory Critical Care Medicine** 174: 120-126, 2006; [Epub Apr 20, 2006] [IF 8.12].

529-OA Radhakrishna U, Ratnamala U, Gaines M, Beiraghi S, Hutchings D, Husain SA, Gambhir PS, Sheth JJ, Sheth FJ, Chetan GK, Naveed M, Solanki JV, Patel UC, Memon R, Antonarakis GS, **Antonarakis SE**, Nath SK. Genome-wide scan for non-syndromic cleft lip and palate in multigenerational Indian families reveals significant evidence for linkage at 13q33.1-34. **American Journal of Human Genetics** 79: 580-585, 2006 [Epub Jul 21, 2006] [IF 12.64].

530-OA Naveed M, Al-Ali MT, Murthy SK, Al-Hajali S, Al-Khaja N, Deutsch S, Bottani A, **Antonarakis SE**, Nath SK, Radhakrishna U, Ectrodactyly with Aplasia of Long Bones (OMIM; 119100) in a Large Inbred Arab Family with an Apparent Autosomal Dominant Inheritance and Reduced Penetrance: Clinical and Genetic Analysis. **American Journal of Medical Genetics** 140: 1440-1446, 2006 [Epub May 10, 2006].

531-OA The ENCODE Project Consortium;
Analysis Coordination: Birney E, Stamatoyannopoulos JA, Dutta A, Guigo R, Gingeras TR, Margulies EH, Weng Z, Snyder M, Dermitzakis ET
Chromatin and Replication: Stamatoyannopoulos JA, Thurman R, Kuehn MS, Taylor CM, Neph S, Koch CM, Asthana S, Malhotra A, Adzhubey I, Greenbaum JA, Andrews RM, Flicek P, Boyle PJ, Cao H, Carter NP, Clelland GK, Davis S, Day N, Dhami P, Dillon SC, Dorschner MO, Giresi PG, Goldy J, Hawrylycz M, Haydock A, Humbert R, James KD, Johnson BE, Johnson SM, Karnani N, Lee K, Lefebvre GC, Navas PA, Neri F, Parker SCJ, Sabo PJ, Sandstrom R, Shafer A, Vetrie D, Weaver M, Wilcox S, Yu M, Collins FS, Dekker J, Lieb JD, Tullius TD, Crawford GE, Sunayev S, Noble WS, Dunham I, Dutta A,
Genes and Transcripts: Guigó R, Denoeud F, Reymond A, Kapranov P, Rozowsky J, Zheng D, Castelo R, Frankish A, Harrow J, Ghosh S, Sandelin A, Hofacker IL, Baertsch R, Keefe D, Flicek P, Dike S, Cheng J, Hirsch H, Sekinger EA, Moqtaderi Z, Yang A, Zhu Z, Lagarde J, Abril JF, Shahab A, Flamm C, Fried C, Hackermüller J, Hertel J, Lindemeyer M, Missal K, Tanzer A, Washietl S, Korbel J, Emanuelsson O, Pedersen JS, Holroyd N, Taylor R, Swarbreck D, Matthews N, Dickson MC, Thomas DJ, Weirauch MT, Gilbert J, Drenkow J, Bell I, Zhao XD, Srinivasan KG, Sung WK, Ooi HS, Chiu KP, Foissac S, Alioto T, Choo SW, Choo CY, Ucla C, Manzano C, Wyss C, Cheung E, Clarke T, Brown JB, Ganesh M, Patel S, Tammana H, Chrast J, Henrichsen CN, Kai C, Kawai J, Nagalakshmi U, Wu J, Lian Z, Lian J, Newburger P, Zhang X, Bickel P, Mattick JS, Carninci P, Hayashizaki Y, Weissman SM, Dermitzakis ET, Margulies EH, Hubbard T, Myers RM, Rogers J, Stadler PF, Lowe TM, Wei CL, Ruan Y, Snyder M, Birney E, Struhl K, Gerstein M, **Antonarakis SE**, Gingeras TR;
Management Group: Liefer L, Collins FS, Good PJ, Guyer MS, Wetterstrand KA, Feingold EA;
Multi-species Sequence Analysis: Margulies EH, Cooper GM, Asimenos G, Thomas DJ, Dewey CN, Siepel A, Birney E, Keefe D, Hou M, Taylor J, Nikolaev S, Montoya-Burgos JI, Löytynoja A, Whelan S, Pardi F, Massingham T, Brown JB, Bickel P, Holmes I, Mullikin JC, Ureta-Vidal A, Paten B, Schuler G, Church D, Rosenbloom KR, Kent WJ, NISC Comparative Sequencing Program, Baylor College of Medicine Human Genome Sequencing Center, Washington University Genome Sequencing Center, Broad Institute, UCSC Genome Browser Team, **Antonarakis SE**, Batzoglou S, Goldman N, Hardison RC, Haussler D, Miller W, Pachter L, Green ED, Sidow A;
Transcriptional Regulatory Elements: Weng Z, Trinklein ND, Fu Y, Zhang Z, Karaöz U, Barrera L, Stuart R, Zheng D, Ghosh S, Flicek P, King DC, Taylor J, Ameur A, Enroth S, Bieda MC, Koch CM, Hirsch HA, Wei CL, Cheng J, Kim J, Bhinge AA, Giresi P, Jiang N, Liu J, Yao F, Sung WK, Chiu KP, Vega VB, Lee CWH, Ng HH, Ng P, Shahab A, Sekinger EA, Yang A, Moqtaderi Z, Zhu Z, Xu X, Squazzo S, Oberley MJ, Inman D, Singer MA, Richmond TA, Munn KJ, Iglesias AR, Wallerman O, Komorowski J, Clelland GK, Wilcox S, Dillon SC, Andrews RM, Fowler JC, Couttet P, James KD, Lefebvre GC, Bruce AW, Dovey OM, Ellis PD, Dhami P, Langford CF, Carter NP, Vetrie D, Kapranov P, Nix DA, Bell I, Patel S, Rozowsky J, Euskirchen G, Hartman S, Lian J, Wu J, Kraus P, Van Calcar S, Heintzman N, Kim TH, Wang K, Qu C, Hon G, Force SA, Cooper SJ, Halees A, Lin JM, Shulha HP, Zhang X, Xu M, Haidar JNS, Yu Y, Birney E, Weissman S, Ruan Y, Lieb JD, Iyer VR, Green RD, Gingeras T, Wadelius C, Dunham I, Struhl K, Hardison RC, Gerstein M, Farnham PJ, Myers RM, Ren B, Snyder M;
Variation: Dermitzakis ET, Armengol L, Bird CP, Clark TG, Cooper GM, Bakker PIW, Kern A, Lopez-Bigas N, Martin JD, Stranger BE, Thomas DJ, Woodroffe A, Batzoglou S, Davydov E, Eyras E, Hallgrimsdottir I, Hardison RC, Huppert J, Sidow A, Taylor J, Trumbower H, Zody M, Guigó R, Mullikin JC, Abecasis G, Estivill X, Birney E. Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. **Nature** 447: 799-816, 2007. [PMID: 17571346] [IF 26.68].

532-OA Naveed M, Nath SK, Gaines M, Al-Ali MT, Al-Khaja N, Hutchings D, Golla J, Deutsch S, Bottani A, **Antonarakis SE**, Ratnamala U, Radhakrishna U. Genome-wide linkage scan for split-hand/foot malformation with long bone deficiency in a large Arab family identifies two novel susceptibility loci on chromosomes 1q42.2-q43 and 6q14.1. **American Journal of Human Genetics** 80: 105-111, 2007. [Epub Nov 29, 2006][IF 12.65].

533-OA Nikolaev S, Montoya-Burgos JI, Margulies EH, NISC Comparative Sequencing Program, Jongeneel V, Rougemont J, Nyffeler B, **Antonarakis SE.** Early History of Mammals is Elucidated with the ENCODE Multiple Species Sequencing Data. **PLoS Genetics** 3: e2, 2007 [PMID: 17206863][Epub Jan 5, 2007][IF 7.67].

534-OA Zheng D, Frankish A, Baertsch R, Kapranov P, Reymond A, Lu Y, Choo SW, Denoeud F, **Antonarakis SE**, Snyder M, Ruan Y, Wei CL, Gingeras TR, Guigo R, Harrow J, Gerstein MB. Consensus Annotation of Pseudogenes in the ENCODE Regions and Analysis of their Transcription and Evolution. **Genome Research** 17: 839-851, 2007 [PMID: 17568002] [IF 13.59].

535-OA Margulies EH, Cooper GM, Asimenos G, Thomas DJ, Dewey CN, Siepel A, Birney E, Keefe D, Schwartz AS, Hou M, Taylor J, Nikolaev S, Montoya-Burgos JI, Löytynoja A, Whelan S, Pardi F, Massingham T, Brown JB, Bickel P, Holmes I, Mullikin JC, Ureta-Vidal A, Paten B, Schuler G, Church D, Rosenbloom KR, Kent WJ, NISC Comparative Sequencing Program, Baylor College of Medicine Human Genome Sequencing Center, Washington University Genome Sequencing Center, Broad Institute, UCSC Genome Browser Team, **Antonarakis SE**, Batzoglou S, Goldman N, Hardison R, Haussler D, Miller W, Pachter L, Green ED, Sidow A. Analyses of deep mammalian sequence alignments and constraint predictions for 1% of the human genome. **Genome Research** 17: 760-774, 2007 [PMID: 17567995] [IF 13.59].

536-OA Washietl S, Pedersen JS, Korbel JO, Fried C, Gruber A, Hackermuller J, Hertel J, Lindemeyer M, Missal K, Tanzer A, Ucla C, **Antonarakis SE**, Reymond A, Denoeud F, Lagarde J, Drenkow J, Kapranov P, Gingeras TR, Snyder M, Gerstein MB, Hofacker IL, Stadler PF. Structured RNAs in the ENCODE Selected Regions of the Human Genome. **Genome Research** 17: 852-864, 2007 [PMID: 17568003] [IF 13.59].

537-OA Denoeud F, Kapranov P, Ucla C, Castelo R, Drenkow J, Lagarde J, Manzano C, Chrast J, Dike S, Wyss C, Henrichsen CN, Frankish A, Harrow J, Holroyd N, Dickson MC, Taylor R, Hance Z, Myers RM, Rogers J, Hubbard T, Guigó R, Gingeras TR, **Antonarakis SE**, Reymond A. Prominent use of distal 5' transcription start sites and discovery of a large number of additional exons in ENCODE regions. **Genome Research** 17: 746-759, 2007 [PMID: 17567994] [IF 13.59].

538-R Conrad B, **Antonarakis SE.** Gene Duplication: A Drive for Phenotypic Diversity and Cause of Human Disease. **Annual Reviews Genomics & Human Genetics** 8: 17-35, 2007 [Epub Mar 26, 2007] [IF 10.09].

539-OA Gagos S, Chiourea M, Kouri G, Deutsch S, Papaioannou G, Apostolou E, Mareli D, Jefford CE, Irminger-Finger I, Shay JW, **Antonarakis SE.** Pericentromeric instability and spontaneous emergence of human neo-acrocentric and minute chromosomes in the alternative pathway of telomere lengthening. **Cancer Research** 68: 8146-8155, 2008 [PMID: 18829574] [IF 7.67].

540-OA Loeuillet C, Deutsch S, Taffé P, Rotger M, Beckmann JS*, **Antonarakis SE***, Telenti A*. In vitro whole-genome analysis identifies a susceptibility locus for HIV-1. * labs equally contributed. **PLoS Biology** 6: e32, 2008 6:319-327, 2008 [PMID: 18288889] [IF 14.10].

541-OA De Pontual L, Pelet A, Clement-Ziza M, Trochet A, **Antonarakis SE**, Attie-Bitach T, Beales P, Blouin JL, Dastot F, Dollfus H, Goossens M, Katsanis N, Touraine R, Feingold J, Munnich A, Lyonnet S, Amiel J. Epistatic interactions with a common hypomorphic Ret allele in syndromic Hirschsprung disease. **Human Mutation** 28: 790-796, 2007 [Epub Mar 30, 2007] [PMID: 17397038] [IF 7.92].

542-OA Schwabe GC, Hoffmann K, Birker D, de Santi MM, Ollbrich H, Failly M, Loges NT, Fliegauf M, Liebers U, Collura M, Gaedicke G, Mundlos S, Wahn U, Blouin JL, Omran H, Niggemann B, **Antonarakis SE**, Bartoloni L. Normal axoneme ultrastructure and perturbed ciliary beating in Primary Ciliary Dyskinesia and Kartagener Syndrome due to C-terminal mutations in the dynein axonemal heavy chain DNAH11. **Human Mutation** 29: 289-298, 2008 [Epub Nov 16, 2007] [PMID: 18022865] [IF 7.92].

543-BC **Antonarakis SE.** STK11 (LKB1) and Peutz-Jeghers syndrome. In Epstein CJ, Erickson RP, Wynshaw-Boris A, eds. Inborn Errors of Development; chapter 58. 2nd edition, Oxford University Press. (ISBN 13-978-0-19-53061-0) pp 589-593, 2008.

544-OL Bottani A, Chelly J, de Brouwer APM, Pardo B, Barker M, Capra V, Bartoloni L, **Antonarakis SE**, Conrad B.

Sequence Variation in Ultraconserved and highly conserved elements does not cause X-Linked Mental Retardation **American Journal of Medical Genetics** 143: 888-890, 2007.

545-OA Lyle R, Prandini P, Osoegawa K, Ten Hallers B, Humphray S, Zhu B, Eyras E, Castelo R, Bird CP, Gagos S, Scott C, Cox A, Deutsch S, Ucla C, Cruts M, Dahoun S, She X, Bena F, Wang SY, Van Broeckhoven C, Eichler EE, Guigo R, Rogers J, de Jong PJ, Reymond A, **Antonarakis SE.** Islands of euchromatin-like sequence and expressed polymorphic sequences within the short arm of human chromosome 21. **Genome Research** 17: 1690-1696, 2007 [Epub Sep 25, 2007] [PMID: 17895424] [IF 13.59].

546-OA Sethupathy P*, Borel C*, Gagnebin M, Deutsch S, Elton TS, Grant GR, Hatzigeorgiou AG, **Antonarakis SE.** Human miR-155 on chromosome 21 differentially interacts with its polymorphic target in the AGTR1 3'UTR – a mechanism for functional SNPs related to phenotypes. (*equally contributed). **American Journal of Human Genetics** 81: 405-413, 2007 [Epub Jul 12, 2007] [PMID: 17668390] [IF 12.64].

547-OA Attanasio C, Reymond A, Humbert R, Lyle R, Kuehn MS, Neph S, Sabo PJ, Goldy J, Weaver M, Lee K, Haydock A, Dermitzakis ET, Dorschner MO, **Antonarakis SE***, Stamatoyannopoulos JA*. Assaying the regulatory potential of mammalian conserved non-coding sequences in human cells. (*labs equally contributed). **Genome Biology** 9: R168, 2008 [Epub Dec 2, 2008] [PMID: 19055709] [IF 6.58].

548-BC **Antonarakis SE**, Cooper DN. Human gene mutation; mechanisms and consequences. in Speicher M, Antonarakis SE, Motulsky A eds. "Vogel & Motulsky's Human Genetics" 4th ed Springer Verlag (ISBN 978-3-540-37653-8) pp 319-363, 2010.

549-OA Voronov SV, Frere SG, Giovedi S, Pollina EA, Borel C, Zhang H, Schmidt C, Akeson EC, Wenk MR, Cimasoni L, Arancio O, Davisson MT, **Antonarakis SE**, Gardiner K, De Camilli P, Di Paolo G. Synaptojanin 1-linked phosphoinositide dyshomeostasis and cognitive deficits in mouse models of Down's syndrome. **Proceedings of the National Academy of Sciences USA** 105: 9415-9420, 2008 [PMID: 18591654] [Epub Jun 30, 2008] [IF 9.64].

550-R Beckmann JS, Estivill X, **Antonarakis SE.** Copy number variants and genetic traits: closer to the resolution of the enigma of phenotypic to genotypic variability. **Nature Reviews Genetics** 8: 639-646, 2007 [PMID: 17637735] [IF 22.94].

551-OA Tress ML, Martelli PL, Frankish A, Reeves GA, Wesselink JJ, Yeats C, Olason PI, Albrecht M, Hegyi H, Giorgetti A, Raimondo D, Lagarde J, Laskowski RA, Lopez G, Sadowski MI, Watson JD, Fariselli P, Rossi I, Nagy A, Kai W, Storling Z, Orsini M, Assenov Y, Blankenburg H, Huthmacher C, Ramirez F, Schlicker A, Denoued F, Jones P, Kerrien S, Orchard S, **Antonarakis SE**, Reymond A, Birney E, Brunak S, Casadio R, Guigo R, Harrow J, Hermjakob H, Jones DT, Lengauer T, Orengo CA, Patthy L, Thornton JM, Tramontano A, Valencia A. The implications of alternative splicing in the ENCODE protein complement. **Proceedings of the National Academy of Sciences USA** 104: 5495-5500, 2007 [Epub Mar 19, 2007] [PMID: 17372197] [IF 10.23].

552-OA Prandini P, Deutsch S, Lyle R, Gagnebin M, Delucinge Vivier C, Delorenzi M, Gehrig C, Descombes P, Sherman S, Dagna Bricarelli F, Baldo C, Novelli A, Dallapiccola B, **Antonarakis SE.** Natural gene expression variation in Down syndrome modulates the outcome of gene dosage imbalance. **American Journal of Human Genetics** 81: 252-263, 2007 [Epub Jun 20, 2007] [PMID: 17668376] [IF 12.64].

553-OA Beiraghi S, Nath SK, Gaines M, Mandhyan DD, Hutchings D, Ratnamala U, McElreavey K, Bartoloni L, Antonarakis GS, **Antonarakis SE**, Radhakrishna U. Autosomal dominant non-syndromic cleft lip and palate: Significant evidence for linkage at 18q21.1. **American Journal of Human Genetics** 81: 180-188, 2007 [Epub May 18, 2007] [PMID: 17564975] [IF 12.65].

554-R Guipponi M, **Antonarakis SE**, Scott HS. TMPRSS3, a type II transmembrane serine protease mutated in non-syndromic autosomal recessive deafness. **Frontiers in Biosciences** 13: 1557-1567, 2008. [PMID: 17981648] [IF 2.77].

555-OA Nikolaev S, Montoya-Burgos JI, Popadin K, Parand L, Margulies EH, NISC Comparative Sequencing Program, **Antonarakis SE.** Life-History Traits Driving the Evolutionary Rates in Coding and Non-Coding Elements in Mammals. **Proceedings of the National Academy of Sciences USA** 104: 20443-20448, 2007 [Epub Dec 11, 2007] [PMID: 18077382] [IF 9.64].

556-OA Fellay J, Shianna KV, Ge D, Colombo S, Ledergerber B, Weale M, Zhang K, Gumbs C, Castagna A, Cossarizza A, Cozzi-Lepri A, De Luca A, Easterbrook P, Francioli P, Mallal S, Martinez-Picado J, Miro JM, Obel N, Smith JP, Wyniger J, Descombes P, **Antonarakis SE**, Letvin NL, McMichael AJ, Haynes BF, Telenti A, Goldstein DB. A Whole-Genome Association Study of Major Determinants for Host Control of HIV-1. **Science** 317: 944-947, 2007 [Epub Jul 19, 2007] [PMID: 17641165] [IF

30.02].

557-BC Patrinos GP, **Antonarakis SE.** Human Hemoglobin. in Speicher M, Antonarakis SE, Motulsky A eds. "Vogel & Motulsky's Human Genetics" 4th ed Springer Verlag (ISBN 978-3-540-37653-8) pp 365-401, 2010.

558-OA Borel C, Gagnebin M, Gehrig C, Kriventseva EV, Zdobnov EM, **Antonarakis SE.** Mapping of small RNAs in the human ENCODE regions. **American Journal of Human Genetics** 82:971-981, 2008 [PMID: 18394580] [IF 12.65].

559-OA Fokstuen S, Lyle R, Munoz A, Gehrig C, Lerch R, Perrot A, Osterziel KJ, Geier C, Beghetti M, Mach F, Sztajzel J, Sigwart U, **Antonarakis SE,** Blouin JL. A DNA Resequencing array for pathogenic Mutation Detection in Hypertrophic Cardiomyopathy. **Human Mutation** 29: 879-885, 2008 [PMID: 18409188] [IF 7.92].

560-OA Martinet D, Filges I, Besuchet Schmutz N, Morris MA, Gaide AC, Dahoun S, Bottani A, Addor MC, **Antonarakis SE,** Beckmann JS, Béna F. Subtelomeric 6p Deletion: clinical and array-CGH characterization in two patients. **American Journal of Medical Genetics** 146A: 2094-2102, 2008 [Epub Jul 15, 2008] [PMID: 18629875] [IF 2.44].

561-OA Friedli M, Spitz F, Nikolaev S, Lyle R, Arcangeli M, Duboule D, **Antonarakis SE.** Characterisation of mouse Dactylaplasia mutations, a model for human ectrodactyly SHFM3. **Mammalian Genome** 19: 272-278, 2008 [Epub Apr 5, 2008] [PMID: 18392654] [IF 2.29].

562-OA Djebali S, Kapranov P, Foissac S, Lagarde J, Reymond A, Ucla C, Wyss C, Drenkow J, Dumais E, Murray RR, Lin C, Szeto D, Denoeud F, Calvo M, Frankish A, Harrow J, Makrythanasis P, Vidal M, Salehi-Ashtiani K, **Antonarakis SE,** Gingeras TR, Guigó R. Efficient targeted transcript discovery via array-based normalization of RACE libraries. **Nature Methods** 5: 629-635, 2008 [Epub May 25, 2008] [PMID: 18500348] [IF 15.47].

563-Book Series **Antonarakis SE,** Fantini B. Special issue - History of Central Dogma of Moleculary Biology and its Epistemological Status today - Geneva, February 22-23, 2007 (ISSN: 0391-9714). **History and Philosophy of the Life Sciences** 28: 487-490, 2006.

564-OA Bonnefont J*, Nikolaev S*, Perrier AL, Guo S, Cartier L, Sorce S, Laforge T, Aubry L, Khaitovich P, Peschanski M, **Antonarakis SE,** Krause KH. Evolutionary forces shape the human RFPL1,2,3 genes toward a role in brain cortical development. (*equally contributed). **American Journal of Human Genetics** 83: 208-218, 2008 [Epub Jul 24, 2008] [PMID: 18656177] [IF 12.65].

565-OA Dahoun D, Gagos S, Gagnebin M, Gehrig C, Burgi C, Simon F, Vieux C, Lyle R, Morris MA, **Antonarakis SE,** Béna F, Blouin JL. Meiotic and mitotic non-disjunctions in monozygotic twins discordant for trisomy 21 and maternal 21qter inheritance. **American Journal of Medical Genetics** 146A: 2086-2093, 2008 [Epub Jul 14, 2008] [PMID: 18627064] [IF 2.44].

566-OA Lopes Pereira P, Magnol L, Sahún Abizanda I, Brault V, Duchon A, Prandini P, Gruart A, Bizot JC, Chadefaux-Vekemans B, Deutsch S, Trovero F, Delgado-García JM, **Antonarakis SE,** Dierssen M, Herault Y. A new mouse model for the trisomy of the Abcg1-U2af1 region reveals the complexity of the combinatorial genetic code of Down syndrome. **Human Molecular Genetics** 18: 4756-4769, 2009 [Epub Sep 26, 2009] [PMID: 19783846] [IF 7.25].

567-R Borel C, **Antonarakis SE.** Functional genetic variation of human miRNAs and phenotypic consequences. **Mammalian Genome** 19: 503-509, 2008. [Epub Sep 12, 2008] [PMID: 18787897] [IF 2.29].

568-OA Kazadi K, Loeuillet C, Deutsch S, Beckmann JS, Moradpour D, **Antonarakis SE,** Telenti A. Genomic determinants of the efficiency of internal ribosomal entry sites of viral and cellular origin. **Nucleic Acids Research** 36: 6918-6925, 2008 [Epub Oct 31, 2008] [PMID: 18978018] [IF 7.47].

569-OA Hubert FX, Kinkel SA, Crewther PE, Cannon PZF, Webster KE, Link M, Uibo R, O'Bryan MK, Meager A, Forehan SP, Smyth GK, Mittaz L, **Antonarakis SE,** Peterson P, Heath WR, Scott HS. Aire-deficient C57BL/6 mice mimicking the common human 13-bp deletion mutation present with only a mild autoimmune phenotype. **Journal of Immunology** 182: 3902-3918, 2009 [PMID: 19265170] [IF 6.07].

570-R Beckmann JS, Sharp AJ, **Antonarakis SE.** CNVs and Genetic Medicine (excitement and consequences of a rediscovery). **Cytogenetics & Genome Research** 123: 7-16, 2008 [Epub Mar 11, 2009] [PMID: 19287134] [IF 2.40].

571-OA Mefford HC*, Sharp AJ*, Baker C, Itsara A, Jiang Z, Buysse K, Huang S, Maloney VK, Crolla JA, Baralle D, Collins A, Mercer C, Norga K, de Ravel T, Devriendt K, Bongers EM, de Leeuw N, Reardon W, Gimelli S, Bena F, Hennekam RC, Male A,

Gaunt L, Clayton-Smith J, Simonic I, Park SM, Mehta SG, Nik-Zainal S, Woods CG, Firth HV, Parkin G, Fichera M, Reitano S, Giudice ML, Li KE, Casuga I, Broomer A, Conrad B, Schwerzmann M, Räber L, Gallati S, Striano P, Coppola A, Tolmie JL, Tobias ES, Lilley C, Armengol L, Spysschaert Y, Verloo P, De Coene A, Goossens L, Mortier G, Speleman F, van Binsbergen E, Nelen MR, Hochstenbach R, Poot M, Gallagher L, Gill M, McClellan J, King MC, Regan R, Skinner C, Stevenson RE, **Antonarakis SE**, Chen C, Estivill X, Menten B, Gimelli G, Gribble S, Schwartz S, Sutcliffe JS, Walsh T, Knight SJ, Sebat J, Romano C, Schwartz CE, Veltman JA, de Vries BB, Vermeesch JR, Barber JC, Willatt L, Tassabehji M, Eichler EE. Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. (*equally contributed). **New England Journal of Medicine** 359: 1685-1699, 2008 [Epub Sep 10, 2008] [PMID: 18784092] [IF 52.58].

572-OA Lyle R, Béna F, Gagos S, Gehrig C, Lopez G, Schinzel A, Lespinasse J, Bottani A, Dahoun S, Taine L, Doco-Fenzy M, Cornillet-Lefèbvre P, Pelet A, Lyonnet S, Toutain A, Colleaux L, Horst J, Kennerknecht I, Wakamatsu N, Descartes M, Franklin JC, Florentin-Arar L, Kitsiou S, Aït Yahya-Graison E, Costantine M, Sinet PM, Delabar JM, **Antonarakis SE.** Genotype-phenotype correlations in Down syndrome identified by array CGH in 30 cases of partial trisomy and partial monosomy chromosome 21. **European Journal of Human Genetics** 17: 454-466, 2008. [Epub Nov 12, 2008] [PMID: 19002211].

573-OA Moldrich RX, Dauphinot L, Laffaire J, Vitalis T, Hérault Y, Beart PM, Rossier J, Vivien D, Gehrig C, **Antonarakis SE**, Lyle R, Potier MC. Proliferation deficits and gene expression dysregulation in Down's syndrome (Ts1Cje) neural progenitor cells cultured from neurospheres. **Journal of Neuroscince Research** 87: 3143-3152, 2009 [Epub May 26, 2009] [PMID: 19472221] [IF 3.27].

574-R Harrow J, Nagy A, Reymond A, Alioto T, Patthy L, **Antonarakis SE**, Guigó R. Identifying Protein-Coding Genes in Genomic Sequences. **Genome Biology** 10: 201, 2009 [Epub Jan 30, 2009] [PMID: 19226436] [IF 6.59] (abstract).

575-OA Lespinasse J, Gimelli S, Béna F, **Antonarakis SE**, Ansermet F, Paoloni-Giacobino A. Characterization of an interstitial deletion 6q13-q14.1 in a girl with mental retardation, speech delay, and minor dysmorphisms. **European Journal of Medical Genetics** 52: 49-52, 2009 [Epub Oct 21, 2008] [PMID: 18992376].

576-OA Nikolaev S*, Deutsch S*, Genolet R, Borel C, Ucla C, Schütz F, Duriaux G, Dupré Y, Jaquier-Gubler P, Araud T, Conne B, Delorenzi M, Descombes P, Vassalli JD, Curran J, **Antonarakis SE.** Transcriptional and post-transcriptional profile of human chromosome 21. (*equally contributed). **Genome Research** 19: 1471-1479, 2009 [Epub Jul 6, 2009] [PMID: 19581486] [IF 13.59].

577-OA Bedard K, Attar H, Bonnefont J, Jaquet V, Borel C, Plastre O, Stasia MJ, **Antonarakis SE**, Krause KH. Three common polymorphisms in the CYBA gene form a haplotype associated with decreased ROS generation. **Human Mutation** 30: 1123-1133, 2009 [Epub Mar 31, 2009] [PMID: 19388116] [IF 6.27].

578-E Ballabio A, **Antonarakis SE.** Welcome to PathoGenetics. **Pathogenetics** 1:1, 2008. [PMID: 19014665].

579-E **Antonarakis SE.** 2008 ASHG Awards; Allan Award Introduction: Haig H. Kazazian Jr. **American Journal of Human Genetics** 84: 103–104, 2009.

580-OA The Bovine Genome Sequencing and Analysis Consortium, Elsik CG, Tellam RL, Worley KC, Gibbs RA, Muzny DM, Weinstock GM, Adelson DL, Eichler EE, Elnitski L, Guigó R, Hamernik DL, Kappes SM, Lewin HA, Lynn DJ, Nicholas FW, Reymond A, Rijnkels M, Skow LC, Zdobnov EM, Schook L, Womack J, Alioto T, **Antonarakis SE**, Astashyn A, Chapple CE, Chen HC, Chrast J, Câmara F, Ermolaeva O, Henrichsen CN, Hlavina W, Kapustin Y, Kiryutin B, Kitts P, Kokocinski F, Landrum M, Maglott D, Pruitt K, Sapojnikov V, Searle SM, Solovyev V, Souvorov A, Ucla C, Wyss C, Anzola JM, Gerlach D, Elhaik E, Graur D, Reese JT, Edgar RC, McEwan JC, Payne GM, Raison JM, Junier T, Kriventseva EV, Eyras E, Plass M, Donthu R, Larkin DM, Reecy J, Yang MQ, Chen L, Cheng Z, Chitko-McKown CG, Liu GE, Matukumalli LK, Song J, Zhu B, Bradley DG, Brinkman FS, Lau LP, Whiteside MD, Walker A, Wheeler TT, Casey T, German JB, Lemay DG, Maqbool NJ, Molenaar AJ, Seo S, Stothard P, Baldwin CL, Baxter R, Brinkmeyer-Langford CL, Brown WC, Childers CP, Connelley T, Ellis SA, Fritz K, Glass EJ, Herzig CT, Iivanainen A, Lahmers KK, Bennett AK, Dickens CM, Gilbert JG, Hagen DE, Salih H, Aerts J, Caetano AR, Dalrymple B, Garcia JF, Gill CA, Hiendleder SG, Memili E, Spurlock D, Williams JL, Alexander L, Brownstein MJ, Guan L, Holt RA, Jones SJ, Marra MA, Moore R, Moore SS, Roberts A, Taniguchi M, Waterman RC, Chacko J, Chandrabose MM, Cree A, Dao MD, Dinh HH, Gabisi RA, Hines S, Hume J, Jhangiani SN, Joshi V, Kovar CL, Lewis LR, Liu YS, Lopez J, Morgan MB, Nguyen NB, Okwuonu GO, Ruiz SJ, Santibanez J, Wright RA, Buhay C, Ding Y, Dugan-Rocha S, Herdandez J, Holder M, Sabo A, Egan A, Goodell J, Wilczek-Boney K, Fowler GR, Hitchens ME, Lozado RJ, Moen C, Steffen D, Warren JT, Zhang J, Chiu R, Schein JE, Durbin KJ, Havlak P, Jiang H, Liu Y, Qin X, Ren Y, Shen Y, Song H, Bell SN, Davis C, Johnson AJ, Lee S, Nazareth LV, Patel BM, Pu LL, Vattathil S, Williams RL Jr, Curry S, Hamilton C, Sodergren E, Wheeler DA, Barris W, Bennett GL, Eggen A, Green RD, Harhay GP, Hobbs M, Jann O, Keele JW, Kent MP, Lien S, McKay SD, McWilliam S, Ratnakumar A, Schnabel RD, Smith T, Snelling WM, Sonstegard TS, Stone RT, Sugimoto Y, Takasuga A, Taylor JF, Van Tassell CP, Macneil MD, Abatepaulo AR, Abbey CA, Ahola V, Almeida IG, Amadio AF, Anatriello E, Bahadue SM, Biase FH, Boldt CR, Carroll JA, Carvalho WA, Cervelatti EP, Chacko

E, Chapin JE, Cheng Y, Choi J, Colley AJ, de Campos TA, De Donato M, Santos IK, de Oliveira CJ, Deobald H, Devinoy E, Donohue KE, Dovc P, Eberlein A, Fitzsimmons CJ, Franzin AM, Garcia GR, Genini S, Gladney CJ, Grant JR, Greaser ML, Green JA, Hadsell DL, Hakimov HA, Halgren R, Harrow JL, Hart EA, Hastings N, Hernandez M, Hu ZL, Ingham A, Iso-Touru T, Jamis C, Jensen K, Kapetis D, Kerr T, Khalil SS, Khatib H, Kolbehdari D, Kumar CG, Kumar D, Leach R, Lee JC, Li C, Logan KM, Malinverni R, Marques E, Martin WF, Martins NF, Maruyama SR, Mazza R, McLean KL, Medrano JF, Moreno BT, Moré DD, Muntean CT, Nandakumar HP, Nogueira MF, Olsaker I, Pant SD, Panzitta F, Pastor RC, Poli MA, Poslusny N, Rachagani S, Ranganathan S, Razpet A, Riggs PK, Rincon G, Rodriguez-Osorio N, Rodriguez-Zas SL, Romero NE, Rosenwald A, Sando L, Schmutz SM, Shen L, Sherman L, Southey BR, Lutzow YS, Sweedler JV, Tammen I, Telugu BP, Urbanski JM, Utsunomiya YT, Verschoor CP, Waardenberg AJ, Wang Z, Ward R, Weikard R, Welsh TH Jr, White SN, Wilming LG, Wunderlich KR, Yang J, Zhao FQ. The genome sequence of Taurine Cattle: A window to ruminant biology and evolution. **Science** 324: 522-528, 2009 [PMID: 19390049] [IF 28.1].

581-OA Dimas AS, Deutsch S, Stranger BE, Montgomery S, Borel C, Ingle C, Beazley C, Gutierrez Arcelus M, Attar-Cohen H, Sekowska M, Gagnebin M, Nisbett J, Deloukas P, Dermitzakis ET, **Antonarakis SE.** Most common regulatory variation impacts gene expression in a tissue-dependent manner. **Science** 325: 1246-1250, 2009 [Epub Jul 30, 2009] [PMID: 19644074] [IF 28.1].

582-OA Nelis M, Esko T, Mägi R, Zimprich F, Zimprich A, Toncheva D, Karachanak S, Piskácková T, Balascák I, Peltonen L, Jakkula E, Rehnström K, Lathrop M, Heath S, Galan P, Schreiber S, Meitinger T, Pfeufer A, Wichmann HE, Melegh B, Polgár N, Toniolo D, Gasparini P, D'Adamo P, Klovins J, Nikitina-Zake L, Kucinskas V, Kasnauskiene J, Lubinski J, Debniak T, Limborska S, Khrunin A, Estivill X, Rabionet R, Marsal S, Julià A, **Antonarakis SE**, Deutsch S, Borel C, Attar H, Gagnebin M, Macek M, Krawczak M, Remm M, Metspalu A. **PLoS ONE** 4: e5472, 2009. [Epub May 8, 2009] [PMID: 19424496] [IF 4.35].

583-OA Makrythanasis P, Kapranov P, Bartoloni L, Reymond A, Deutsch S, Guigó R, Denoeud F, Drenkow J, Rossier C, Ariani F, Capra V, Excoffier L, Renieri A, Gingeras TR, **Antonarakis SE.** Variation in novel exons (RACEfrags) of the MECP2 gene in Rett and controls. **Human Mutation** 30: E866-879, 2009 [Epub Jun 26, 2009] [PMID: 19562714] [IF 7.03].

584-OA Diez Roux G, Banfi S, Sultan M, Geffers L, Anand A, Rozado D, Magen A, Canidio E, Pagani M, Peluso I, Lin-Marq N, Koch M, Mundlos S, Bilio M, Cantiello I, Verde R, De Masi C, Bianchi S, Cicchini J, Perroud E, Mehmeti S, Dagand E, Schrinner S, Nürnberger A, Schmidt K, Metz K, Zwingmann C, Brieske N, Springer C, Martinez-Hernandez A, Herzog S, Grabbe F, Sieverding C, Fischer B, Schrader K, Bürsing M, Schubert S, Helbig C, Alunni V, Battaini MA, Mura C, Henrichsen CN, Garcia-Lopez R, Echevarria D, Puelles E, Calero E, Kruse S, Uhr M, Kauck C, Feng G, Milyaev N, Kee Ong C, Kumar L, Lam M, Semple C, Gyenesei A, Radelof U, Lehrach H, Sarmientos P*, Reymond A, Davidson DR*, Dollé P*, **Antonarakis SE***, Yaspo ML*, Martinez S*, Baldock RA*, Eichele G*, Ballabio A*. A high-resolution anatomical atlas of the murine transcriptome at embryonic stage E14.5. (*labs equally contributed). **PLoS Biology** 9: e1000582, 2011 [doi: 10.1371/journal.pbio.1000582] [Epub Jan 18, 2011] [PMID: 21267068] [IF 14.79].

585-OA D'Haene B*, Attanasio C*, Beysen D*, Dostie J, Lemire E, Bouchard P, Field M, Jones K, Lorenz B, Menten B, Buysse K, Pattyn F, Friedli M, Ucla C, Rossier C, Wyss C, Speleman F, De Paepe A, Dekker J, **Antonarakis SE**, De Baere E. A distant deletion of a 7.4 kb fragment that harbours long-range cis-regulators and interacts with the FOXL2 promoter causes BPES. (*equally contributed). **PLoS Genetics** 5: e1000522, 2009 [Epub Jun 19, 2009] [doi:10.1371/journal.pgen.1000522] [PMID: 19543368] [IF 8.88].

586-OA Fellay J, Ge D, Shianna KV, Colombo S, Ledergerber B, Cirulli ET, Urban TJ, Zhang K, Gumbs CE, Smith JP, Castagna A, Cozzi-Lepri A, De Luca A, Easterbrook P, Günthard HF, Mallal S, Mussini C, Dalmau J, Martinez-Picado J, Miro JM, Obel N, Wolinsky SM, Martinson JJ, Detels R, Margolick JB, Jacobson LP, Descombes P, **Antonarakis SE**, Beckmann JS, O'Brien SJ, Letvin NL, McMichael AJ, Haynes BH, Carrington M, Feng S, Telenti A, Goldstein DB for the NIAID Center for HIV/AIDS Vaccine Immunology (CHAVI). Common genetic variation and the control of HIV-1 in humans. **PLoS Genetics** 5: e1000791, 2009. [Epub Dec 24, 2009] [PMID: 20041166] [IF 8.88].

587-OA Nikolaev S*, Iseli C*, Sharp AJ*, Robyr D, Rougemont J, Gehrig C, Farinelli L, **Antonarakis SE.** Detection of genomic variation by selection of a 9 Mb DNA region and high throughput sequencing. (*equally contributed). **PLoS ONE** 4: e6659, 2009 [doi:10.1371/journal.pone.0006659] [PMID: 19684856] [IF 4.35].

588-OA Friedli M, Barde I, Arcangeli M, Quazzola A, Verp S, Zakany J, Lin-Marq N, Robyr D, Attanasio C, Spitz F, Duboule D, Trono D, **Antonarakis SE.** High throughput enhancer screen using lentivector-mediated transgenesis identifies evolutionary conserved and non-conserved functional elements. **PLoS ONE** 5: e15741, 2010 [Epub Dec 29, 2010] [doi:10.1371/journal.pone.0015741] [PMID: 21206754] [IF 4.35].

589-OA Borel C, Deutsch S, Letourneau A, Migliavacca E, Montgomery SB, Dimas AS, Vejnar C, Attar H, Gagnebin M,

Gehrig C, Falconnet E, Dupré Y, Dermitzakis ET, **Antonarakis SE.** Identification of cis- and trans- regulatory variation modulating microRNA expression levels in human fibroblasts. **Genome Research** 21: 68-73, 2011 [Epub Dec 8, 2010] [PMID: 21147911] [IF 13.59].

590-OA Makrythanasis P, Moix I, Gimelli S, Fluss J, **Antonarakis SE,** Morris MA, Béna F, Bottani A. De novo MECP2 duplication in a girl with mild mental retardation and no obvious dysmorphic features. **Clinical Genetics** 78: 175-180, 2010 [Epub Jan 5, 2010] [PMID: 20236124].

591-BC Beckmann JS, **Antonarakis SE.** Lessons from the Genome-wide Association Studies for complex multifactorial disorders and traits. in Speicher M, Antonarakis SE, Motulsky A eds. "Vogel & Motulsky's Human Genetics" 4th ed Springer Verlag (ISBN 978-3-540-37653-8) pp 287-298, 2010.

592-OA Robyr D, Friedli M, Gehrig C, Arcangeli M, Marin M, Farinelli L, Quazzola A, Verp S, Trono D, **Antonarakis SE.** Chromosome Conformation Capture Uncovers Potential Genome-Wide Interactions between Human Conserved Non-Coding Sequences. **PLoS ONE** 6: e17634, 2011 [Epub Mar 7, 2011] (doi:10.1371/journal.pone.0017634) [PMID: 21408183] [IF 4.35] (abstract).

593-BC **Antonarakis SE.** Human Genome Sequence and Variation. in Speicher M, Antonarakis SE, Motulsky A eds. "Vogel & Motulsky's Human Genetics" 4th ed Springer Verlag (ISBN 978-3-540-37653-8) pp 31-53, 2010.

594-OA Djebali S, Lagarde J, Kapranov P, Lacroix V, Ribeca P, Mudge J, Ucla C, Foissac S, Howald C, Chrast J, Murry RR, Yang X, Ghamsari L, Lin C, Bell I, Dumais E, Drenkow J, van Berkum NL, Lajoie BR, Vidal M, Stamatoyannopoulos J, Harrow J, Hubbard T, Dekker J, Frankish A, Salehi-Ashtiani K, Reymond A, **Antonarakis SE***, Guigo R*, Gingeras TR*. Evidence for transcript networks composed of chimeric RNAs in human cells. (*labs equally contributed). **PLoS ONE** 7: e28213, 2012 [Epub Jan 4, 2012] (doi:10.1371/journal.pone.0028213) [PMID: 22238572] [IF 4.35] (abstract).

595-OA Béna F, Gimelli S, Migliavacca E, Brun-Druc N, Buiting K, **Antonarakis SE,** Sharp AJ. A recurrent 14q32.2 microdeletion mediated by expanded TGG repeats. **Human Molecular Genetics** 19: 1967-1973, 2010 [Epub Mar 2, 2010] [PMID: 20179077] [IF 7.25].

596-Book Speicher MR, **Antonarakis SE,** Motulsky AG. "Vogel and Motulsky's Human Genetics; problems and Approaches" 4th edition. Springer-Verlag, Berlin, Heidelberg (ISBN 978-3-540-37653-8) pp 981, 343 illus, 2010.

597-OA Sharp AJ, Migliavacca E, Dupre Y, Stathaki E, Sailani MR, Baumer A, Schinzel A, Mackay D, Robinson DO, Cobellis G, Cobellis L, Brunner HG, Steiner B, **Antonarakis SE.** Methylation profiling in individuals with uniparental disomy identifies novel differentially methylated regions on chromosome 15. **Genome Research** 20: 1271-1278, 2010 [Epub Jul 14, 2010] [PMID: 20631049] [IF 13.59].

598-OA De Cegli R, Romito A, Iacobacci S, Mao L, Lauria M, Fedele A, Klose J, Borel C, Descombes P, **Antonarakis SE,** di Bernardo D, Banfi S, Ballabio A, Cobellis G. A mouse embryonic stem cell bank for inducible overexpression of human chromosome 21 genes. **Genome Biology** 11: R64, 2010 [Epub Jun 22, 2010] [PMID: 20569505] [IF 6.13].

599-OA Fort A*, Borel C*, Migliavacca E, **Antonarakis SE,** Fish RJ, Neerman-Arbez M. Regulation of fibrinogen production by microRNAs. (*equally contributed). **Blood** 116: 2608-2615, 2010 [Epub Jun 22, 2010] [PMID: 20570858] [IF 10.55].

600-CR Lochmüller H, Aymé S, Pampinella F, Melegh B, Kuhn K, **Antonarakis SE,** Meitinger T. The Role of Biobanking in Rare Diseases: European Consensus Expert Group Report. **Biopreservation & Biobanking** 7: 155-156, 2009 (doi: 10.1089/bio.2010.0703.bdr) [PMID: 248358827].

601-N&V **Antonarakis SE,** Chakravarti A, Cohen JC, Hardy J. Mendelian disorders and multifactorial traits: the big divide or one for all? **Nature Reviews Genetics** 11: 380-384, 2010 (doi:10.1038/nrg2793) [PMID: 20395971] [IF 24.18].

602-OA Kapranov P, Ozsolak F, Kim SW, Foissac S, Huang J, Lipson D, Hart C, Roels S, Borel C, **Antonarakis SE,** Monaghan AP, John B, Milos PM. Novel class of human RNA species associated with gene termini suggests an uncharacterized RNA copying mechanism. **Nature** 466: 642-646, 2010 [PMID: 20671709] [IF 31.43].

603-OA Marini M, Bocciardi R, Gimelli S, Di Duca M, Divizia MT, Baban A, Gaspar H, Mammi I, Garavelli L, Cerone R, Emma F, Bedeschi MF, Tenconi R, Sensi A, Salmaggi A, Bengala M, Mari F, Colussi G, Szczaluba K, **Antonarakis SE,** Seri M, Lerone M, Ravazzolo R. A spectrum of LMX1B mutations in Nail-Patella Syndrome: new point mutations, deletion, and evidence of mosaicism in unaffected parents. **Genetics in Medicine** 12: 431-439, 2010 [Epub Jun 7, 2010] [PMID: 20531206].

604-OA Glaser B, Debbane M, Ottet MC, Zesiger P, Vuilleumier P, **Antonarakis SE**, Eliez S. Eye gaze during face processing in children and adolescents with 22q11.2 deletion syndrome (22q11DS). **Journal of American Academy of Child & Adolescent Psychiatry** (JAACAP) 49: 665-674, 2010 [Epub Jun 2, 2010] [PMID: 20610136] [IF 4.98].

605-BC Clark AG, Dermitzakis ET, **Antonarakis SE.** Trisomic Phase Inference. In Istrail S et al. Eds: "SNPs and Haplotype Inference" Springer-Verlag Heidelberg. LNBI 2983, pp. 1-8, 2004.

606-OA Hovatta O, Jaconi M, Töhönen V, Béna F, Gimelli S, Bosman A, Holm F, Wyder S, Zdobnov EM, Irion O, Andrews PW, **Antonarakis SE**, Zucchelli M, Kere J, Feki A. A Teratocarcinoma-Like Human Embryonic Stem Cell (hESC) Line and Four hESC Lines Reveal Potentially Oncogenic Genomic Changes. **PLoS ONE** 5: e10263, 2010 [Epub Apr 23, 2010] [PMID: 20428235] (doi:10.1371/journal.pone.0010263) [IF 4.35].

607-OA Gimelli S, Makrythanasis P, Stouder C, **Antonarakis SE**, Bottani A, Béna F. A de novo 12q13.11 microdeletion in a patient with severe mental retardation, cleft palate, and high myopia. **European Journal of Medical Genetics** 54: 94-96, 2010 [Epub Oct 6, 2010] [PMID: 20933621].

608-OA Sharp AJ, Stathaki E, Migliavacca E, Brahmachary M, Montgomery S, Dupre Y, Jacobs PA, **Antonarakis SE.** DNA methylation profiles of human active and inactive X chromosomes. **Genome Research** 21: 1592-1600, 2011 [Epub Aug 23, 2011] [PMID: 21862626] [IF 13.59].

609-OA Gimelli S, Divizia MT, Lerone M, Bricco L, Béna F, **Antonarakis SE**, Ravazzolo R, Gimelli G. Array-CGH analysis in a patient with WAGR syndrome and a reciprocal translocation t(2;11) inherited from the normal father with double translocation. **American Journal of Medical Genetics** A. 152A: 2130-2133, 2010 [Epub Jul 15, 2010] [PMID: 20635368].

610-OA Valsesia A, Rimoldi D, Martinet D, Ibberson M, Benaglio P, Quadroni M, Waridel P, Gaillard M, Pidoux M, Rapin B, Rivolta C, Xenarios I, Simpson AJ, **Antonarakis SE**, Beckmann JS, Jongeneel CV, Iseli C, Stevenson BJ. Network-Guided Analysis of Genes with Altered Somatic Copy Number and Gene Expression Reveals Pathways Commonly Perturbed in Metastatic Melanoma. **PLoS ONE** 6: e18369, 2011 [Epub Apr 8, 2011] [PMID: 21494657] [IF 4.35].

611-R Gardiner K, Herault Y, Lott IT, **Antonarakis SE**, Reeves R, Dierssen M. Down Syndrome: From Understanding the Neurobiology to Therapy. **Journal of Neuroscience** 30: 14943-14945, 2010 (doi 10.1523/JNEUROSCI.3728-10.2010) [Epub Nov 10, 2010] [PMID: 21068296] (abstract).

612-OA Fokstuen S, Munoz A, Melacini P, Iliceto S, Perrot A, Özcelik C, Jeanrenaud X, Rieubland C, Farr M, Faber L, Sigwart U, Mach F, Lerch R, **Antonarakis SE**, Blouin JL. Rapid detection of genetic variants in hypertrophic cardiomyopathy by custom DNA resequencing array in clinical practice. **Journal of Medical Genetics** 48: 572-576, 2011 [PMID: 21239446] [Epub Jan 14, 2011] (abstract).

613-OA Radhakrishna U, Nath SK, McElreavey K, Ratnamala U, Sun C, Maiti AK, Gagnebin M, Béna F, Newkirk HL, Sharp AJ, Everman DB, Murray JC, Schwartz CE, **Antonarakis SE**, Butler MG. Genome-wide linkage and copy number variation analysis reveals 710 kb duplication on chromosome 1p31.3 responsible for autosomal dominant omphalocele. **Journal of Medical Genetics** 49: 270-276, 2012 [PMID: 22499347].

614-R Liu C, Belichenko P, Zhang L, Fu D, Kleschevnikov A, **Antonarakis SE**, Mobley WC and Yu YE. Animal models for Down syndrome-associated developmental cognitive disabilities. Dev. Neurosci. 33: 404-413, 2011 [Epub Aug 25, 2011] [PMID: 21865664] [IF 2.70].

615-R Hamamy H, **Antonarakis SE**, Cavalli-Sforza LL, Temtamy S, Romeo G, Ten Kate LP, Bennett RL, Shaw A, Megarbane A, van Duijn C, Bathija H, Fokstuen S, Engel E, Zlogotora J, Dermitzakis E, Bottani A, Dahoun S, Morris MA, Arsenault S, Aglan MS, Ajaz M, Alkalamchi A, Alnaqeb D, Alwasiyah MK, Anwer N, Awwad R, Bonnefin M, Corry P, Gwanmesia L, Karbani GA, Mostafavi M, Pippucci T, Ranza-Boscardin E, Reversade B, Sharif SM, Teeuw ME, Bittles AH. Consanguineous Marriages, Pearls and Perils: Geneva International Consanguinity Workshop Report. **Genetics in Medicine** 13: 841-847, 2011 [Epub May 6, 2011] [PMID: 21555946] [IF 3.92].

616-OA Jacquemont S, Zufferey F, Harewood L, Kutalik Z, Walters RG, Martinet D, Shen Y, Valsesia A, Beckmann ND, Thorleifsson G, Belfiore M, Bouquillon S, Campion D, de Leeuw N, de Vries BBA, Fernandez BA, Fernández-Aranda F, Gratacòs M, Guilmatre A, Hoyer J, Kurg A, Le Caignec C, Männik K, Ounap K, Platt OS, Sanlaville D, Van Haelst MM, Villatoro Gomez S, Vulto-van Silfhout AT, Walha F, Wu BL, Yu Y, Aboura A, Alembik Y, **Antonarakis SE**, Arveiler B, Barth M, Bednarek N, Béna F, Bergmann S, Beri M, Bernardini L, Blaumeiser B, Bottani A, Boute O, Brunner HG, Cailley D, Callier P, Chiesa J,

Chrast J, Coutton C, Cuisset JM, Cuvellier JC, David A, de Freminville B, Delobel B, Delrue MA, Demeer B, Descamps D, Didelot G, Dieterich K, Disciglio V, Doco-Fenzy M, Drunat S, Duban-Bedu B, Dubourg C, El-Sayed Moustafa J, Faas BHW, Faivre L, Faudet A, Fellmann F, Real JM, Fisher R, Flori E, Forer L, Gaillard D, Gerard M, Gieger C, Gimelli S, Gimelli G, Grabe HJ, Guichet A, Guillin O, Gustafsson O, Hartikainen AL, Heron D, Hippolite L, Holder M, Homuth G, Isidor B, Jaillard S, Jaros Z, Jarvelin MR, Joly Helas G, Jonveaux P, Kaksonen S, Keren B, Kloss-Brandstätter A, VAM Knoers NVAM, Koolen DA, Kooy FR, Kroisel PM, Kronenberg F, Labalme A, Landais E, Lapi E, Layet V, Legallic S, Leheup B, Leube B, Lewis S, Lucas J, MacDermot KD, Magnusson P, Mathieu-Dramard M, Meitinger T, Mencarelli MA, Merla G, Moerman A, Moser V, Mucciolo M, Nauck M, Ndiaye C, Nordgren A, Pasquier L, Petit F, Picard F, Plessis G, Rajcan-Separovic E, Ramelli GP, Rauch A, Ravazzolo R, Renieri A, Richart C, Ried J, Rieubland C, Roetzer KM, Rooryck C, Rossi M, Saemundssen E, Satre V, Schurman C, Sigurdsson E, Stefansson H, Tensgröm C, Tinahones FJ, Touraine R, Vallée L, van Binsbergen E, Van der Aa N, Vincent-Delorme C, Visvikis-Siest S, Vollenweider P, Völzke H, Waeber G, Wallgren-Pettersson C, Witwicki RM, Zwolinksi S, Andrieux J, Estivill X, Gusella JF, Metspalu A, Scherer SW, Stefansson K, Thorsteinsdóttir U, Blakemore AIF, Beckmann JS, Reymond A, Froguel P. Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. **Nature** 478: 97-102, 2011 [Epub Aug 31, 2011] [PMID: 21881559] [IF 31.43] (abstract).

617-OA Ratnamala U, Lyle R, Raval R, Singh R, Vishnupriya S, Himabindu P, Rao VV, Aggarwal S, Paluru P, Bartoloni L, Young TL, Paoloni-Giacobino A, Morris MA, Nath S, **Antonarakis SE**, Radhakrishna U. Refinement of the X-linked nonsyndromic high-grade myopia locus (MYP1) on Xq28 and exclusion of thirteen known positional candidate genes by direct sequencing. **Invest Ophthalmol Visual Sciences IOVS** 52: 6814-6819, 2011 [Epub Feb 25, 2011] [PMID: 21357393] [IF 3.43].

618-N&V Makrythanasis P, **Antonarakis SE.** From sequence to functional understanding: the difficult road ahead. **Genome Medicine** 3: 21-24, 2011 [Epub Apr 6, 2011] [PMID: 21470392].

619-OA Fasquelle L, Scott HS, Lenoir M, Wang J, Rebillard G, Gaboyard S, Venteo S, François F, Mausset-Bonnefont AL, **Antonarakis SE**, Neidhart E, Chabbert C, Puel JL, Guipponi M, Delprat B. Tmprss3, a Transmembrane Serine Protease Deficient in Human DFNB8/10 Deafness, is Critical for Cochlear Hair Cell Survival at the Onset of Hearing. **Journal of Biological Chemistry** 286: 17383-17397, 2011 [Epub Mar 21, 2011] [PMID: 21454591] [IF 6.35] (abstract).

620-OA Nikolaev SI, Rimoldi D, Iseli C, Valsesia A, Robyr D, Gehrig C, Harshman K, Guipponi M, Bukach O, Zoete V, Michielin O, Muehlethaler K, Speiser D, Beckmann JS, Xenarios I, Halazonetis TD, Jongeneel CV, Stevenson BJ, **Antonarakis SE.** Exome sequencing identifies recurrent MAP2K1 and MAP2K2 mutations in melanoma. **Nature Genetics** 44:133-139, 2012 [Epub Dec 25, 2011] (doi:10.1038/ng.1026) [PMID: 22197931] [IF 36.38] (abstract).

621-OA The ENCODE Project Consortium: Myers RM, Stamatoyannopoulos J, Snyder M, Dunham I, Hardison RC, Bernstein BE, Gingeras TR, Kent WJ, Birney E, Wold B, Crawford GE, Epstein CB, Shoresh NS, Ernst J, Mikkelsen TS, Kheradpour P, Zhang X, Wang L, Issner R, Coyne MJ, Durham T, Ku M, Truong T, Ward LD, Altshuler RC, Lin MF, Kellis M, Davis CA, Kapranov P, DobinA, Zaleski C, Schlesinger F, Batut P, Chakrabortty S, Jha S, Lin W, Drenkow J, Wang H, Bell K, Gao H, Bell I, Dumais E, Dumais J, **Antonarakis SE**, Ucla C, Borel C, Guigo R, Djebali S, Lagarde J, Kingswood C, Ribeca P, Sammeth M, Alioto T, Merkel A, Tilgner H, Carninci P, Hayashizaki Y, Lassmann T, Takahashi H, Abdelhamid RF, Hannon G, Fejes-Toth K, Preall J, Gordon A, Sotirova V, Reymond A, Howald C, Aït Yahya Graison E, Chrast J, Ruan Y, Ruan X, Shahab A, Ting Poh W, Wei CL, Furey TS, Boyle AP, Sheffield NC, Song L, Shibata Y, Vales T, Winter D, Zhang Z, London D, Wang T, Keefe D, Iyer VR, Lee BK, McDaniell RM, Liu Z, Battenhouse A, Bhinge AA, Lieb JD, Grasfeder LL, Showers KA, Giresi PG, Kim SKC, Shestak C, Pauli F, Reddy TE, Gertz J, Partridge EC, Jain P, Sprouse RO, Bansal A, Pusey B, Muratet MA, Varley KE, Bowling KM, Newberry KM, Nesmith AS, Dilocker JA, Parker SL, Waite LL, Thibeault K, Roberts K, Absher DM, Mortazavi A, Williams B, Marinov G, Trout D, Pepke S, King B, McCue K, Kirilusha A, DeSalvo G, Fisher-Aylor K, Amrhein H, Vielmetter J, Sherlock G, Sidow A, Batzoglou S, Rauch R, Kundaje A, Libbrecht M, Margulies EM, Parker SCJ, Elnitski L, Green ED, Hubbard T, Harrow J, Searle S, Kokocinski F, Aken B, Frankish A, Hunt T, Despacio-Reyes G, Kay M, Mukherjee G, Bignell A, Saunders G, Boychenko V, Brent M, Van Baren MJ, Brown RH, Gerstein M, Khurana E, Balasubramanian S, Zhang Z,, Lam H, Cayting P, Robilotto R, Lu Z, Guigo R, Derrien T, Tanzer A, Knowles DG, Mariotti M, Harte R, Diekhans M, Lin M, Kheradpour P, Valencia A, Tress M, Rodriguez JM, Landt SG, Raha D, Shi M, Euskirchen G, Grubert F, Kasowski M, Lian J, Cayting P, Lacroute P, Xu Y, Monahan H, Patacsil D, Slifer T, Yang X, Charos A, Reed B, Wu L, Auerbach RK, Habegger L, Hariharan M, Rozowsky J, Abyzov A, Weissman SM, Gerstein M, Struhl K, Lamarre-Vincent N, Lindahl-Allen M, Miotto B, Moqtaderi Z, Fleming JD, Newburger P, Farnham PJ, Frietze S, O'Geen H, Xu X, Blahnik KR, Cao AR, Iyengar S, Kaul R, Thurman RE, Wang H, Navas PA, Sandstrom R, Sabo PJ, Weaver M, Canfield T, Lee K, Neph S, Roach V, Reynolds A, Johnson A, Rynes E, Giste E, Vong S, Neri J, Frum T, Johnson EM, Nguyen ED, Ebersol AK, Sanchez ME, Sheffer HH, Lotakis D, Haugen E, Humbert R, Kutyavin T, Shafer T, Dekker J, Lajoie BR, Sanyal A, Rosenbloom KR, Dreszer TR, Raney BJ, Barber GP, Meyer LR, Sloan CA, Malladi VS, Cline MS, Learned K, Swing VK, Zweig AS, Rhead B, Fujita PA, Roskin K, Karolchik D, Kuhn RM, Haussler D, Wilder SP, Keefe D, Sobral D, Herrero J, Beal K, Lukk M, Brazma A, Vaquerizas JM, Luscombe NM, Bickel PJ, Boley N, Brown JB, Li Q, Huang H, Gerstein M, Habegger L, Sboner A, Auerbach RK, Yip KY, Cheng C, Yan KK, Bhardwaj N, Wang J, Lochovsky L, Jee J, Gibson T, Leng J, Du J, Harris RS, Song G, Miller W, Roskin K, Suh B, Wang T, Paten B, Noble WS, Hoffman MM, Buske OJ, Weng Z, Dong X, Wang

J, Xi H, Tenenbaum SA, Doyle F, Penalva LO, Chittur S, Tullius TD, Parker SCJ, White KW, Karmakar S, Victorsen A, Jameel N, Bild N, Grossman RL, Landt SG, Yang X, Patacsil D, Slifer T, Dekker J, Lajoie BR, Sanyal A, Weng Z, Whitfield TW, Wang J, Collins PJ, Trinklein ND, Partridge EC, Myers RM, Giddings MC, Chen X, Khatun J, Maier C, Yu Y, Gunawardena H, Risk B, Feingold EA, Lowdon RF, Dillon LAL, Good PJ. A User's Guide to the Encyclopedia of DNA Elements (ENCODE). **PLoS Biology** 9: e1001046, 2011 [Epub Apr 19, 2011] [PMID: 21526222] [IF 14.79].

622-OA Klopocki E, Lohan S, Doelken SD, Stricker S, Ockeloen CW, Soares Thiele de Aguiar RS, Lezirovitz K, Mingroni-Netto RC, Jamsheer A, Shah H, Kurth I, Habenicht R, Warman M, Devriendt K, Kordaß U, Rajab A, Mäkitie O, Naveed M, Radhakrishna U, **Antonarakis SE**, Horn D, Mundlos S. Duplications of BHLHA9 are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. **Journal of Medical Genetics** 49:119-125, 2012. (doi: 10.1136/jmedgenet-2011-100409) [Epub Dec 6, 2011].

623-R Bousquet J, Anto JM, Sterk PJ, Adcock IM, Chung KF, Roca J, Agusti A, Brightling C7, Cambon-Thomsen A, Cesario A, Abdelhak S, **Antonarakis SE**, Avignon A, Ballabio A, Baraldi E, Baranov A, Bieber T, Bockaert J, Brahmachari S, Brambilla C, Bringer J, Dauzat M, Ernberg I, Fabbri L, Froguel P, Galas D, Gojobori T, Hunter P, Jorgensen C, Kauffmann F, Kourilsky P, Kowalski ML, Lancet D, Le Pen C, Mallet J, Mayosi B, Mercier J, Metspalu A, Nadeau JH, Ninot G, Noble D, Öztürk M, Palkonen S, Préfaut C, Rabe K, Renard E, Roberts RG, Samolinski B, Schünemann HJ, Simon HU, Soares MB, Superti-Furga G, Tegner J, Verjovski-Almeida S, Wellstead P, Wolkenhauer O, Wouters E, Balling R, Brookes AJ,Charron D, Pison C, Chen Z, Hood L, Auffray C. Systems medicine and integrated care to combat chronic noncommunicable diseases. **Genome Medicine** 3:43, 2011 [Epub Jul 6, 2011] [PMID: 21745417].

624-R Letourneau A, **Antonarakis SE**. Genomic determinants in the phenotypic variability of Down syndrome. In "Down Syndrome: From Understanding the Neurobiology to Therapy" ed. M Dierssen. **Progress in Brain Research** 197: 15-28, 2012 [PMID: 22541286].

625-OA Grad I, Hibaoui Y, Jaconi M, Chicha L, Bergström-Tengzelius R, Sailani MR, Pelte MF, Dahoun S, Mitsiadis TA, Töhönen V, Bouillaguet S, **Antonarakis SE**, Zucchelli M, Hovatta O, Feki A. Nanog priming before full reprogramming may generate germ cell tumors. **European Cell Mater eCM** 22: 258-274, 2011 [PMID: 22071697] [IF 9.65].

626-B **Antonarakis SE**, Bartholdi D, Gallati S, Imhasly P, Müller HJ, Papassotiropoulos A, Porz R. La Génétique dans la Médecine au quotidien. 2nd ed. Booklet published by the Swiss Academy of Medical Sciences, Basel pgs 90, 2011.

627-OA Makrythanasis P, Gimelli S, Béna F, Morris MA, Dahoun S, **Antonarakis SE**, Bottani A. Homozygous deletion of a gene-free region of 4p15 in a child with multiple anomalies: could biallelic loss of conserved, non-coding elements lead to a phenotype ? **European Journal of Medical Genetics** 55: 63-66, 2012 [Epub Nov 10, 2011].

628-OA Cossec JC, Lavaur J, Berman DE, Rivals I, Hoischen A, Stora S, Ripoll C, Mircher C, Grattau Y, Olivo-Marin JC, de Chaumont F, Lecourtois M, **Antonarakis SE**, Veltman JA, Delabar JM, Duyckaerts C, Di Paolo G, Potier MC. Trisomy for Synaptojanin1 in Down syndrome is functionally linked to the enlargement of early endosomes. **Human Molecular Genetics** 21: 3156-3172, 2012 [Epub Apr 17, 2012] [PMID: 22511594] [IF 8.06] (abstract).

629-OA The ENCODE Project Consortium. Dunham I, Kundaje A, Aldred SF, Collins PJ, Davis CA, Doyle F, Epstein CB, Frietze S, Harrow J, Kaul R, Khatun J, Lajoie BR, Landt SG, Lee BK, Pauli F, Rosenbloom KR, Sabo P, Safi A, Sanyal A, Shoresh N, Simon JM, Song L, Trinklein ND, Altshuler RC, Brown JB, Cheng C, Djebali S, Dong X, Ernst J, Furey TS, Gerstein M, Giardine B, Greven M, Hardison RC, Harris RS, Herrero J, Hoffman MM, Iyer S, Kellis M, Khatun J, Kheradpour P, Kundaje A, Lassmann T, Li Q, Lin X, Marinov GK, Merkel A, Mortazavi A, Parker SCJ, Reddy TE, Rozowsky J, Schlesinger F, Thurman RE, Wang J, Ward LD, Whitfield TW, Wilder SP, Wu WS, Xi HS, Yip K, Zhuang J, Green ED, Gunter C, Snyder M, Pazin MJ, Lowdon RF, Dillon LAL, Adams LB, Kelly CJ, Zhang J, Wexler JR, Good PJ, Feingold EA, Bernstein BE, Birney W, Crawford GE, Dekker J, Elnitski L, Farnham PJ, Giddings MC, Gingeras TR, Green ED, Guigo R, Hubbard TJ, Kent WJ, Lieb JD, Margulies EH, Myers RM, Stamatoyannopoulos JA, Tenenbaum SA, Weng Z, White KP, Wold B, Jainab Khatun J, Yanbao Yu Y, John Wrobel J, Brian A. Risk BA, Gunawardena H, Kuiper HC, Maier CW, Xie L, Chen X, Giddings MC, Epstein CB, Shoresh N, Ernst J, Kheradpour P, Mikkelsen TS, Gillespie S, Goren A, Ram O, Zhang X, Wang L, Issner R, Coyne MJ, Durham T, Ku M, Truong T, Ward LD, Altshuler RC, Eaton ML, Djebali S, Davis CA, Merkel A, Dobin A, Lassmann T, Mortazavi A, Tanzer A, Lagarde J, Lin W, Schlesinger F, Xue C, Marinov GK, Khatun J, Williams BA, Zaleski C, Rozowsky J, Roder M, Kokocinski F, Abdelhamid RF, Alioto T, Antoshechkin I, Baer MT, Batut P, Bell I, Bell K, Chakrabortty S, Chen X, Chrast J, Curado J, Derrien T, Drenkow J, Dumais E, Dumais J, Duttagupta R, Fastuca M, Fejes-Toth K, Ferreira P, Foissac S, Fullwood MJ, Gao H, Gonzalez D, Gordon A, Gunawardena H, Howald C, Jha S, Johnson R, Kapranov P, King B, Kingswood C, Li G, Luo OJ, Park E, Preall JB, Presaud K, Ribeca P, Risk BA, Robyr D, Ruan X, Sammeth M, Sandhu KS, Schaeffer L, See LH, Shahab A, Skancke J, Suzuki AM, Takahashi H, Tilgner H, Trout D, Walters N, Wang H, Wrobel J, Yu YB, Hayashizaki Y, Harrow J, Gerstein M, Reymond A, **Antonarakis SE**, Hannon GJ, Giddings MC, Ruan YJ, Wold B, Carninci P, Guigo R, Gingeras TR, Learned K, Malladi VS, Rosenbloom KR, Sloan

CA, Wong MC, Barber GP, Cline MS, Dreszer TR, Heitner SG, Karolchik D, Kent WJ, Kirkup VM, Meyer LR, Long JC, Maddren M, Raney BJ, Furey TS, Song L, Grasfeder LL, Giresi PG, Lee BK, Battenhouse A, Sheffield NC, Simon JM, Showers KA, Safi A, London D, Bhinge AA, Shestak C, Schaner MR, Kim SK, Zhang ZZ, Mieczkowski PA, Mieczkowska JO, Liu Z, McDaniell RM, Ni Y, Rashid NU, Kim MJ, Adar S, Zhang ZC, Wang TY, Winter D, Keefe D, Iyer VR, Lieb JD, Crawford GE, Li G, Sandhu KS, Zheng M, Wang P, Luo OJ, Shahab A, Fullwood MJ, Ruan X, Ruan Y, Pauli F, Williams BA, Gertz J, Marinov GK, Reddy TE, Vielmetter J, Partridge EC, Trout D, Varley KE, Gasper C, Bansal A, Pepke S, Jain P, Amrhein H, Bowling KM, Anaya M, Cross MK, Brandon King B, Muratet MA, Antoshechkin I, Newberry KM, McCue K, Nesmith AS, Fisher-Aylor KI, Pusey B, DeSalvo G, Parker SL, Balasubramanian S, Davis NS, Meadows SK, Eggleston T, Gunter C, Newberry JS, Levy SE, Absher DM, Mortazavi A, Wong WH, Wold B, Blow MJ, Visel A, Pennachio LA, Parker SCJ, Petrykowska HM, Abyzov A, Aken A, Barrell D, Barson G, A, Bignell A, Boychenko V, Bussotti G, Chrast J, Davidson C, Derrien T, Despacio-Reyes G, Diekhans M, Ezkurdia I, Frankish A, Gilbert J, Gonzalez JM, Griffiths E, Harte R, Hendrix DA, Howald C, Hunt T, Jungreis I, Kay M, Khurana E, Kokocinski F, Leng JJ, Lin MF, Loveland J, Lu Z, Manthravadi D, Mariotti M, Mudge J, Mukherjee G, Notredame C, Pei B, Rodriguez JM, Saunders G, Sboner A, Searle S, Sisu C, Snow C, Steward C, Tanzer A, Tapanari E, Tress ML, van Baren MJ, Walters N, Washietl S, Wilming L, Zadissa A, Zhang Z, Brent M, Haussler D, Valencia A, Landt SG, Frietze S, Abyzov A, Addleman N, Alexander RP, Auerbach RK, Balasubramanian S, Bettinger K, Bhardwaj N, Boyle AP, Cao AR, Cayting P, Charos A, Cheng Y, Cheng C, Eastman C, Euskirchen G, Fleming JD, Grubert F, Habegger L, Hariharan M, Harmanci A, Iyengar S, Jin VX, Karczewski KJ, Kasowski M, Lacroute P, Lam H, Lamarre-Vincent N, Leng JJ, Lian J, Lindahl-Allen M, Min R, Miotto B, Monahan H, Moqtaderi Z, Mu XJ, O'Geen H, Ouyang ZQ, Patacsil D, Pei B, Raha D, Ramirez L, Reed B, Sboner A, Shi M, Sisu C, Slifer T, Witt H, Wu L, Xu X, Yan KK, Yang X, Yip K, Zhang ZD, Struhl K, Weissman SM, Gerstein M, Farnham PJ, Tenenbaum SA, Penalva LO, Doyle F, Karmakar S, Landt SG, Bhanvadia RR, Choudhury A, Domanus M, Ma L, Moran J, Patacsil D, Slifer T, Victorsen A, Yang X, White KP, Auer T, Centanin L, Eichenlaub M, Gruhl F, Heermann S, Inoue D, Kellner T, Kirchmaier S, Mueller C, Reinhardt R, Schertel L, Schneider S, Sinn R, Wittbrodt B, Wittbrodt J, Weng Z, Whitfield TW, Wang J, Collins PJ, Aldred SF, Trinklein AD, Partridge EC, Jain G, Lajoie BR, Sanyal A, Balasundaram G, Bates DL, Byron R, Canfield TK, Diegel MJ, Dunn D, Ebersol AK, Tristan Frum T, Garg K, Gist E, Hansen RS, Boatman L, Haugen E, Humbert R, Jain G, Johnson AK, Johnson EM, Kutyavin TV, Lajoie BR, Lee K, Lotakis D, Maurano MT, Neph SJ, Neri FV, Nguyen ED, Qu H, Reynolds A, Roach V, Rynes E, Sabo P, Sanchez ME, Sandstrom RS, Sanyal A, Shafer AO, Stergachis AB, Thomas S, Thurman RE, Vernot B, Vierstra J, Vong S, Wang H, Weaver MA, Yan Y, Zhang MH, Akey JM, Bender M, Dorschner MO, Groudine M, MacCoss MJ, Navas P, Stamatoyannopoulos G, Kaul R, Dekker J, Dunham I, Beal K, Brazma A, Flicek P, Herrero J, Johnson N, Keefe D, Lukk M, Luscombe NM, Sobral D, Vaquerizas JM, Wilder SP, Batzoglou S, Sidow A, Hussami N, Kyriazopoulou-Panagiotopoulou S, Libbrecht MW, Schaub MA, Kundaje A, Hardison RC, Miller W, Giardine B, Harris RS, Wu WS, Bickel PJ, Banfai B, Boley NP, Brown JB, Huang H, Li Q, Li JJ, Noble WS, Bilmes JA, Buske OJ, Hoffman MM, Sahu AD, Kharchenko PV, Park PJ, Weng ZP, Iyer S, Dong XJ, Greven M, Lin XY, Wang J, Xi HS, J Zhuang J, Gerstein M, Alexander RP, Balasubramanian S, Cheng C, Harmanci A, Lochovsky L, Min RQ, Mu XM, Rozowsky J, Yan KK, Yip K, Birney E. An Integrated Encyclopedia of DNA Elements in the Human Genome. **Nature** 489: 57–74, 2012 (doi:10.1038/nature11247) [PMID: 22955616] [IF 36.28](abstract and PDF).

630-BC **Antonarakis SE**, Cooper DN. Human Gene Mutation in Inherited Disease: Molecular Mechanisms and Clinical Consequences. Chapter 7 in Rimoin DL, Connor JM, Pyeritz RE, Korf BR, eds. "Emery and Rimoin's Principles and Practice of Medical Genetics" 6th ed. Churchill Livingstone, 2012.

631-OA Djebali S, Davis CA, LaGarde J, Dobin A, Zaleski C, Merkel A, Lassmann T, Lin W, Schlesinger F, Xue C, Tanzer A, Mortizavi A, Khatun J, Kokocinski F, Abdelhamid RF, Alioto T, Baer MT, Batut P, Bell K, Bell I, Chakrabortty S, Chen X, Chrast J, Curado J, Derrien T, Drenkow J, Dumais E, Dumais J, Duttagupta R, Fastuca M, Fejes-Toth K, Ferreira P, Foissac S, Fullwood MJ, Gao H, Gonzalez D, Gordon A, Gunawardena H, Howald C, Johnson R, Kapranov P, Kingswood C, Luo OJ, Marinov GK, Park E, Preall JB, Ribeca P, Risk B, Robyr D, Roeder M, Sammeth M, See LH, Shahab A, Skancke J, Suzuki AM, Takahashi H, Tilgner H, Trout D, Walters N, Wang H, Williams BA, Wrobel J, Yu Y, Ruan X, Hayashizaki Y, Harrow J, Hubbard T, Reymond A, **Antonarakis SE**, Hannon G, Giddings MC, Ruan Y, Wold B, Carninci P, Guigo R, Gingeras TR. Landscape of transcription in human cells. **Nature** 489: 101–108, 2012 (doi:10.1038/nature11233) [PMID: 22955620] [IF 36.28] (abstract and PDF).

632-OA Tlili A, Hoischen A, Ripoll C, Benabou E, Badel A, Ronan A, Touraine R, Grattau Y, Stora S, van Bon B, de Vries B, Menten B, Bockaert N, Gecz J, **Antonarakis SE**, Campion D, Potier MC, Bléhaut H, Delabar JM, Janel N. BDNF and DYRK1A are variable and inversely correlated in lymphoblastoid cell lines from Down syndrome patients. Molec. Neurobiol. 46: 297-303, 2012 [Epub Jun 5, 2012] [PMID: 22669612] (abstract).

633-OA Borel C, Migliavacca E, Letourneau A, Gagnebin M, Béna F, Sailani MR, Dermitzakis ET, Sharp AJ, **Antonarakis SE**. Tandem repeat sequence variation as causative cis eQTLs for protein-coding gene expression variation: the case of CSTB. **Human Mutation** 33: 1302-1309, 2012 [Epub Jun 15, 2012] [PMID: 22573514] (abstract).

634-OA Makrythanasis P, van Bon BW, Steehouwer M, Rodríguez-Santiago B, Simpson M, Dias P, Anderlid BM, Arts P, Bongers EM, Frissyra E, Gilissen C, Hoyer J, Ijntema H, C Kets CM, Koolen DA, Marcelis CL, de Munnik S, Roscioli T, Ruiterkamp-Veersteg M, Schuurs-Hoeijmakers JH, Revencu N, Schoumans J, van der Burgt I, van Lier B, Zweier C, Reymond A, Trembath RC, Perez-Jurado L, Dupont JM, de Vries B, Brunner HG, Veltman JA, Merla G, **Antonarakis SE**[§], Hoischen A[§]

([§]labs equally contributed). MLL2 mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. Clinical Genetics 84: 539-545, 2013 (doi: 10.1111/cge.12081) [Epub Jan 16, 2013] [PMID: 23320472] (abstract)

635-OA Schneider M, Van der Linden M, Glaser B, Rizzi E, Dahoun SP, Hinard C, Bartoloni L, **Antonarakis SE**, Debbané M, Eliez S. Preliminary structure and predictive value of attenuated negative symptoms in 22q11.2 deletion syndrome. Psychiatry Research 196: 277-284, 2012 [Epub Feb 27, 2012] [PMID: 22377578] (abstract).

636-L The BLUEPRINT Consortium: Adams D, Altucci L, **Antonarakis SE**, Ballesteros J, Beck S, Bird A, Bock C, Boehm B, Campo Guerri E, Caricasole A, Dahl F, Dermitzakis ET, Enver T, Esteller Badosa M, Estivill X, Ferguson-Smith A, Fitzgibbon J, Flicek P, Giehl C, Graf T, Grosveld F, Guigo R, Gut I, Helin K, Jarvius J, Küppers R, Lehrach H, Lengauer T, Lernmark A, Leslie D, Loeffler M, Macintyre E, Mai A, Martin Subero JI, Minucci S, Ouwehand W, Pelicci PG, Pendeville H, Rakyan V, Reik W, Schübeler D, Seifert M, Siebert R, Simmons D, Soranzo N, Spicuglia S, Stratton M, Stunnenberg H, Tanay A, Torrents D, Valencia A, Vellenga E, Vingron M, Walter J, Willcocks S. BLUEPRINT to decode the epigenetic signature written in blood. Nature Biotechnology 30: 224-226, 2012 (doi: 10.1038/nbt.2153) [PMID: 22398613].

637-OA Radhakrishna U, Ratnamala U, Deutsch S, Bartoloni L, Kuracha MR, Singh R, Banwait J, Bastola DK, Johar K, Nath SK, **Antonarakis SE**. Novel homozygous, heterozygous and hemizygous FRMD7 gene mutations segregated in the same consanguineous family with congenital X-linked nystagmus. European Journal of Human Genetics 20: 1032-1036, 2012 (doi: 10.1038/ejhg.2012.60) [Epub Apr 11, 2012] [PMID: 22490987] (abstract).

638-OA Attar H, Bedard K, Migliavacca E, Gagnebin M, Dupré Y, Descombes P, Borel C, Deutsch S, Prokisch H, Meitinger T, Mehta D, Wichmann E, Delabar JM, Dermitzakis ET, Krause KH, **Antonarakis SE**. Extensive natural variation for cellular hydrogen peroxide release is genetically controlled. PLoS ONE 7: e43566, 2012 (doi:10.1371/journal.pone.0043566) [Epub Aug 29, 2012] [PMID: 22952707] (abstract).

639-OA Nikolaev SI, Sotiriou SK, Pateras IS, Santoni F, Sougioultzis S, Edgren H, Almusa H, Robyr D, Guipponi M, Saarela J, Gorgoulis VG, **Antonarakis SE**[§], Halazonetis TD[§] ([§]co-corresponding authors). A Single-Nucleotide Substitution Mutator Phenotype Revealed by Exome Sequencing of Human Colon Adenomas. Cancer Research 72: 6279-6289, 2012 [PMID: 23204322] (abstract).

640-OA Popadin KY, Nikolaev SI, Junier T, Baranova M, **Antonarakis SE**. Purifying selection in mammalian mitochondrial protein-coding genes is highly effective and concordant with evolution of nuclear genes. Molecular Biology & Evolution 30: 347-355, 2013 (doi: 10.1093/molbev/mss219) [Epub Sep 14, 2012] [PMID: 22983951] [IF 5.55] (abstract).

641-OA Esko T, Massimo Mezzavilla M, Nelis M, Borel C, Debniak T, Jakkula E, Julia A, Karachanak S, Khrunin A, Kisfali P, Krulisova V, Kučinskienė ZA, Rehnström K, Traglia M, Nikitina-Zake L, Zimprich F, **Antonarakis SE**, Estivill X, Glavač D, Gut I, Klovins J, Krawczak M, Kučinskas V, Lathrop M, Macek M, Marsal S, Meitinger T, Melegh B, Limborska S, Lubinski J, Paolotie A, Schreiber S, Toncheva D, Toniolo D, Wichmann H, Zimprich A, Metspalu M, Gasparini P, Metspalu A, D'Adamo P.Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. European Journal of Human Genetics 21:659-665, 2013 (doi: 10.1038/ejhg.2012.229) [Epub Dec 19, 2012] [PMID: 23249956] (abstract).

642-OA Nica AC, Ongen H, Irminger JC, Bosco D, Berney T, **Antonarakis SE**, Halban PH, Dermitzakis ET. Cell-type, allelic, and genetic signatures in the human pancreatic beta cell transcriptome. Genome Research 23:1554-1562, 2013 [Epub May 28, 2013] [PMID: 23716500] [IF 13.61] (abstract).

643-OA Gutierrez-Arcelus M, Montgomery SB, Lappalainen T, Ongen H, Yurovsky A, Bryois J, Buil A, Giger T, Romano L, Planchon A, Falconnet E, Padioleau I, Borel C, Letourneau A, Makrythanasis P, Guipponi M, Gehrig C, **Antonarakis SE**[§], Dermitzakis ET[§] ([§]co-corresponding authors). Passive and active DNA methylation and the interplay with genetic variation in gene regulation. eLife 2: e00523, 2013 (doi: 10.7554/eLife.00523) [Epub Jun 4, 2013] [PMID: 23755361] (abstract and PDF).

644-OA Sailani MR, Makrythanasis P, Valsesia A, Santoni F, Deutsch S, Popadin K, Borel C, Migliavacca E, Sharp AJ, Duriaux Sail G, Falconnet E, Rabionet K, Serra-Juhe C, Vicari S, Laux D, Grattau Y, Dembour G, Megarbane A, Touraine R, Stora S, Kitsiou S, Fryssira H, Chatzisevastou-Loukidou C, Kanavakis E, Merla G, Bonnet D, Pérez-Jurado LA, Estivill X, Delabar JM, **Antonarakis SE**. The complex SNP and CNV genetic architecture of the increased risk of Congenital Heart Defects in Down syndrome. Genome Research 23: 1410-1421, 2013 [Epub Jun 19, 2013] (doi: 10.1101/gr.147991.112) [PMID: 23783273] [IF 13.61] (abstract).

645-R Makrythanasis P, **Antonarakis SE**. High Throughput Sequencing and Rare Genetic Diseases. Molecular Syndromology 3: 197-203, 2012 [Epub Nov 9, 2012] [PMID: 23293577] (abstract).

646-OA De Iaco A, Santoni F, Vannier A, Guipponi M, **Antonarakis SE**, Luban J. TNPO3 protects HIV-1 replication from CPSF6-mediated capsid stabilization in the host cell cytoplasm. **Retrovirology** 10: 20, 2013 [Epub Feb 15, 2013] [PMID: 23414560] [IF 6.47] (abstract).

647-OA Béna F, Bruno DL, Eriksson M, van Ravenswaaij-Arts C, Stark Z, Dijkhuizen T, Gerkes E, Gimelli S, Ganesamoorthy D, Thuresson AC, Labalme A, Till M, Bilan F, Pasquier L, Kitzis A, Dubourg C, Rossi M, Bottani A, Gagnebin M, Sanlaville D, Gilbert-Dussardier B, Guipponi M, van Haeringen A, Kriek M, Ruivenkamp C, **Antonarakis SE**, Anderlid BM, Slater HR, Schoumans J. Molecular and clinical characterization of 25 individuals with exonic deletions of NRXN1 and comprehensive review of the literature. **American Journal of Medical Genetics B Neuropsychiatric Genetics** 162B: 388-403, 2013 (doi: 10.1002/ajmg.b.32148) [Epub Mar 26, 2013] [PMID: 23533028] (abstract).

648-OA Giorgio E, Robyr D, Spielmann M, Ferrero E, Di Gregorio E, Imperiale D, Vaula G, Stamoulis G, Santoni F, Atzori C, Gasparini P, Ferrera D, Canale C, Guipponi M, Pennacchio LA, **Antonarakis SE**, Brussino A, Brusco A. A large genomic deletion leads to enhancer adoption by the lamin B1 gene: a second path to autosomal dominant leukodystrophy (ADLD). **Human Molecular Genetics** 24: 3143-3154, 2015 [Epub Feb 20, 2015] [PMID: 25701871] (abstract).

649-OA Lappalainen T, Sammeth M, Friedländer MR, Hoen PAC, Monlong J, Rivas MA, Gonzàlez-Porta M, Kurbatova N, Griebel T, Ferreira PG, Barann M, Wieland T, Greger L, van Iterson M, Almlöf J, Ribeca P, Pulyakhina I, Esser D, Giger T, Tikhonov A, Marc Sultan M, Bertier G, MacArthur DG, Lek M, Lizano E, Buermans HPI, Padioleau I, Schwarzmayr T, Karlberg O, Ongen H, Beltran S, Gut M, Kahlem K, Amstislavskiy V, Pirinen M, Montgomery SB, Donnelly P, McCarthy MI, Flicek P, Strom TM, The Geuvadis Consortium, Lehrach H, Schreiber S, Sudbrak R, Carracedo A, **Antonarakis SE**, Häsler R, Syvänen AC, van Ommen GJ, Brazma A, Meitinger T, Rosenstiel P, Guigó R, Gut IG, Estivill X, Dermitzakis ET. Transcriptome and genome sequencing uncovers functional variation in human populations. **Nature** 501: 506-511, 2013 (doi: 10.1038/nature12531) [Epub Sep 15, 2013] [PMID: 24037378] (abstract).

650-OA Hibaoui Y, Grad I, Letourneau A, Sailani MR, Dahoun S, Santoni FA, Gimelli S, Pelte MF, Béna F, **Antonarakis SE**[§], Feki A[§] ([§]co-corresponding authors). Modelling neurodevelopment in Down syndrome using induced pluripotent stem cells derived from monozygotic twins discordant for trisomy 21. **EMBO Molecular Medicine** 6: 259-277, 2014 [Epub Dec 27, 2013] [PMID: 24375627] [IF 7.79] (abstract).

650a-OA Hibaoui Y, Grad I, Letourneau A, Santoni FA, Antonarakis SE, Feki A. Data in brief: Transcriptome analysis of induced pluripotent stem cells from monozygotic twins discordant for trisomy 21. **Genomics Data** 2: 226–229, 2014 (doi:10.1016/j.gdata.2014.07.006).

651-CR MacArthur DG, Manolio TA, Dimmock DP, Rehm HL, Shendure J, Abecasis G, Adams D, Altman R, **Antonarakis SE**, Ashley E, Barrett JC, Biesecker L, Conrad DF, Cooper G, Cox N, Daly MJ, Gerstein M, Goldstein D, Hirschhorn J, Leal S, Pennacchio L, Stamatoyannopoulos J, Sunyaev S, Valle D, Voight B, Winckler W, Gunter C. Investigating causality of genome sequence variants in human disease. **Nature** 508: 469-476, 2014 (doi: 10.1038/nature13127) [Epub Apr 24, 2014] [PMID: 24759409] (abstract and PDF)

652-OA Makrythanasis P, Santoni FA, Guipponi M, Vannier A, Béna F, Gimelli S, Stathaki E, Temtamy S, Mégarbané A, Masri A, Aglan MS, Zaki MS, Bottani A, Fokstuen S, Gwanmesia L, Aliferis K, Bustamante ME, Stamoulis G, Psoni S, Kitsiou-Tzeli S, Fryssira H, Kanavakis E, Al-Allawi N, Sefiani A, Al-Hait S, Elalaoui SC, Jalkh N, Al-Gazali L, Al-Jasmi F, Chaabouni Bouhamed H, Abdalla E, Cooper DN, Hamamy H, **Antonarakis SE**. Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families. **Human Mutation** 35: 1203-1210, 2014 (doi: 10.1002/humu.22617) [Epub Jul 17, 2014] [PMID: 25044680] (abstract).

653-OA Coppola A, Romito A, Borel C, Altucci L, Banfi S, **Antonarakis SE**, Minchiotti G, Cobellis G. Early cardiomyogenesis is controlled by the let-7c/miR-99a cluster and epigenetic modifications. **Stem Cell Research** 12: 323-337, 2013 (doi: 10.1016/j.scr.2013.11.008) [Epub Nov 28, 2013] [PMID: 24365598] (abstract).

654-OA Nikolaev SI, Santoni F, Vannier A, Falconnet E, Giarin E, Basso G, Hoischen A, Veltman JA, Groet J, Nizetic D, **Antonarakis SE**. Exome sequencing identifies drivers of progression of Transient Myeloproliferative Disorder to AMKL in Down Syndrome. **Blood** 122: 554-561, 2013 [Epub Jun 3, 2013] [PMID: 23733339] (abstract).

655-OA Fokstuen S, Makrythanasis P, Nikolaev S, Santoni F, Robyr D, Munoz A, Bevillard J, Farinelli L, Iseli C, **Antonarakis SE**, Blouin JL. Multiplex Targeted High Throughput Sequencing for mendelian cardiac disorders. **Clinical Genetics** 85: 365-370, 2014 (doi: 10.1111/cge.12168) [Epub Apr 16, 2013] [PMID: 23590259] (abstract).

656-OA Popadin K, Gutierrez-Arcelus M, Dermitzakis ET, **Antonarakis SE.** Genetic and epigenetic regulation of human LincRNAs gene expression variation. American Journal of Human Genetics 93: 1015-1026, 2013. (doi: 10.1016/j.ajhg.2013.10.022) [Epub Nov 19, 2013] [PMID: 24268656] (abstract).

657-OA Moore DM, Onoufriadis A, Shoemark A, Simpson MA, de Castro SC, zur Lage PI, Bartoloni L, Gallone G, Petridi S, Woollard WJ, Antony D, Schmidts M, Bevillard J, Didonna T, Santoni F, Makhrythanasis P, Mongan NP, Djakow J, Pals G, Lucas JS, Marthin JK, Nielsen KG, Guiponni M, **Antonarakis SE**, Hogg C, Emes RD, Chung EMK, Greene NDE, Blouin JL, Jarman AP, Mitchison HM. Mutations in ZMYND10, a gene essential for proper axonemal assembly of inner and outer dynein arms in human and fly, cause primary ciliary dyskinesia. American Journal of Human Genetics 93: 346-356, 2013 (doi: 10.1016/j.ajhg.2013.07.009) [Epub Jul 24, 2013] [PMID: 23891471] (abstract).

658-BF **Antonarakis SE.** Foreword to a book entitled "Genomic Medicine: Principles and Practice, 2nd ed." (Oxford Monographs on Medical Genetics) edited by Kumar D, and Eng C. Oxford University Press, New York. (ISBN 978-0-19-989602-8) pgs vii-viii, 2014.

659-R Makrythanasis P, **Antonarakis SE.** Pathogenic variants in Non-Coding Elements. Clinical Genetics 84: 422-428, 2013 (doi: 10.1111/cge.12272) [Epub Sep 5, 2013] [PMID: 24007299] (abstract).

660-OA 't Hoen PA, Friedländer MR, Almlöf J, Sammeth M, Pulyakhina I, Anvar SY, Laros JF, Buermans HP, Karlberg O, Brännvall M; The GEUVADIS Consortium, van Ommen GJ, Estivill X, Guigó R, Syvänen AC, Gut IG, Dermitzakis ET, **Antonarakis SE**, Brazma A, Flicek P, Schreiber S, Rosenstiel P, Meitinger T, Strom TM, Lehrach H, Sudbrak R, Carracedo A, Pulyakhina I, Anvar SY, Laros JF, Buermans HP, van Iterson M, Friedländer MR, Monlong J, Lizano E, Bertier G, Ferreira PG, Sammeth M, Almlöf J, Karlberg O, Brännvall M, Ribeca P, Griebel T, Beltran S, Gut M, Kahlem K, Lappalainen T, Giger T, Ongen H, Padioleau I, Kilpinen H, Gonzàlez-Porta M, Kurbatova N, Tikhonov A, Greger L, Barann M, Esser D, Häsler R, Wieland T, Schwarzmayr T, Sultan M, Amstislavskiy V, den Dunnen JT, van Ommen GJ, Gut IG, Guigó R, Estivill X, Syvänen AC, Dermitzakis ET, Lappalainen T. Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. Nature Biotechnology 31: 1015-1022, 2013 (doi: 10.1038/nbt.2702) [Epub Sep 15, 2013] [PMID: 24037425] (abstract).

661-OA Steijger T, Abril JF, Engström PG, Kokocinski F, RGASP Consortium (Wu TD, Wold BJ, Trapnell C, Reymond A, Derrien T, Djebali S, Gonzalez D, Lagarde J, Alioto T, Ribeca P, Sammeth M, Kahles A, Rätsch G, Solovyev V, Mortazavi A, **Antonarakis SE**, Du J, Habegger L, Leng J, Sboner A, Gerstein M, Dudoit S, Lefebvre G, Rougemont J, Behr J, Bohnert R, Sonnenburg S, Thalheim L, Zeller G, Stanke M, Salzberg S, Gingeras TR, Lewis S, Richard R, Schulz MH, Weese D, Iseli C, Diaz N, Solorzano N, Stevenson B, Stockinger H, Valsesia A, Bucher P, Ambrosini G, Kokocinski F, Searle MJ, White S), Hubbard TJ, Guigó R, Harrow J, Bertone P. Assessment of transcript reconstruction methods for RNA-seq. Nature Methods 10: 1177-1184, 2013 (doi: 10.1038/nmeth.2714) [Epub Nov 3, 2013] [PMID: 24185837] (abstract).

662-OA Letourneau A, Santoni FA, Bonilla X, Sailani MR, Gonzalez D, Popadin K, Kind J, Chevalier C, Thurman R, Sandstrom RS, Hibaoui Y, Falconnet E, Gagnebin M, Gehrig C, Vannier A, Guipponi M, Farinelli L, Robyr D, Garieri M, Migliavacca E, Borel C, Deutsch S, Feki A, Stamatoyannopoulos JS, Herault Y, van Steensel B, Guigo R, **Antonarakis SE.** Domains of genome-wide gene expression dysregulation in Down's syndrome. Nature 508: 345-350, 2014 (doi: 10.1038/nature13200) [Epub Apr 17, 2014] [PMID: 24740065] (abstract).
N+V: Up and down in Down's syndrome. Pope BD, Gilbert DM. Nature 508: 323-324, 2014 [PMID: 24740061].

663-OA Guipponi M, Santoni FA, Setola V, Gehrig C, Rotharmel M, Cuenca M, Guillin O, Dikeos D, Georgantopoulos G, Papadimitriou G, Curtis L, Méary A, Schürhoff F, Jamain S, Avramopoulos D, Leboyer M, Rujescu D, Pulver A, Campion D, Siderovski DP, **Antonarakis SE.** Exome Sequencing in 53 Sporadic Cases of Schizophrenia Identifies 18 Putative Candidate Genes. PLoS ONE 9:e112745, 2014 (doi: 10.1371/journal.pone.0112745) [Epub Nov 24, 2014] [PMID: 25420024] (abstract)

664-OA Gutierrez-Arcelus M, Ongen H, Lappalainen T, Montgomery SB, Buil A, Yurovsky A, Bryois J, Padioleau I, Romano L, Planchon A, Falconnet E, Bielser D, Gagnebin M, Giger T, Borel C, Letourneau A,, Makrythanasis P, Guipponi M, Gehrig C, **Antonarakis SE**[§], Dermitzakis ET[§] ([§]co-corresponding authors) Tissue-specific effects of genetic and epigenetic variation on gene regulation and splicing. PLoS Genetics 11: e1004958, 2015 (doi: 10.1371/journal.pgen.1004958) [Epub Jan 29, 2015] [PMID: 25634236] [IF 8.16] (abstract and PDF).
Research highlights in Nature Reviews Genetics (here)

665-OA Santoni FA, Makrythanasis P, Nikolaev S, Guipponi M, Robyr D, Bottani A, **Antonarakis SE.** VariantMaster is a robust tool for the identifiction of causative variants from HTS data in familial, denovo and somatic genetic disorders including cancer. Genome Research 24: 349-355, 2014 [Epub Jan 3, 2014] [PMID: 24389049] (abstract).
Research highlights in Nature Reviews Genetics (here).

666-OA Picard F, Makrythanasis P, Navarro V, Ishida S, de Bellescize J, Ville D, Weckhuysen S, Fosselle E, Suls A, De Jonghe P, Vasselon Raina M, Lesca G, Depienne C, An-Gourfinkel I, Vlaicu M, Baulac M, Mundwiller E, Couarch P, Combi R, Ferini-Strambi L, Gambardella A, **Antonarakis SE**, Leguern E, Steinlein O, Baulac S. DEPDC5 mutations in autosomal dominant nocturnal frontal lobe epilepsy. **Neurology** 82: 2101-2106, 2014 [Epub May 9, 2014] [PMID: 24814846] [IF 8.25] (abstract).

667-Let Makrythanasis P, Hamamy H, **Antonarakis SE**, Mauron A, Hurst SA. Analysis of the Born in Bradford birth cohort. **Lancet** 383: 123, 2014 (doi: 10.1016/S0140-6736(14)60019-2) [PMID: 24411966].

668-OA Callier P, Calvel P, Matevossian A, Makrythanasis P, Bernard P, Kurosaka H, Vannier A, Thauvin-Robinet C, Borel C, Mazaud-Guittot S, Rolland A, Desdoits-Lethimonier C, Guipponi M, Zimmermann C, Stévant I, Kuhne F, Conne B, Santoni F, Lambert S, Huet F, Mugneret F, Jaruzelska J, Faivre L, Wilhelm D, Jégou B, Trainor PA, Resh MD, **Antonarakis SE**, Nef S. Loss of function mutation in the palmitoyl-transferase HHAT leads to syndromic 46,XY disorder of sex development by impeding Hedgehog protein palmitoylation and signaling. **PLoS Genetics** 10: e1004340, 2014 (doi: 10.1371/journal.pgen.1004340) [Epub May 1, 2014] [PMID: 24784881] (abstract and PDF).

669-OA Nikolaev S, Garieri M, Santoni F, Falconnet E, Ribaux P, Guipponi M, Murray A, Groet J, Giarin E, Basso G, Nizetic D, **Antonarakis SE**. Frequent RAS mutations in acute lymphoblastic leukaemia in Down syndrome are almost mutually exclusive with JAK2 mutations. **Nature Communications** 5: 4654, 2014 (doi: 10.1038/ncomms5654) [Epub Aug 8, 2014] [PMID: 25105841] (abstract).

670-OA Nikolaev S, Santoni F, Garieri M, Makrythanasis P, Falconnet E, Guipponi M, Vannier A, Clement V, Bena F, Schaller K, Dutoit V, Dietrich PY, **Antonarakis SE**. Extrachromosomal driver mutations in glioblastoma and low grade glioma. **Nature Communications** 5: 5690, 2014 (doi: 10.1038/ncomms6690) [Epub Dec 4, 2014] [PMID: 25471132] (abstract).

671-N+V **Antonarakis SE**. MOOCs: La fin des Facultes de Medecine conventionelles?. **Revue Medicale Suisse** 10: 719-720, 2014 [PMID: 24783740] (pdf).

672-OA Popadin KY, Gutierrez-Arcelus M, Lappalainen T, Buil A, Nikolaev SI, Lukowski SW, Bazykin GA, Seplyarskiy VB, Ioannidis P, Zdobnov E, Dermitzakis ET, **Antonarakis SE**. Gene age predicts the strength of purifying selection acting on gene expression variation in humans. **American Journal of Human Genetics** 95: 660-674, 2014. (doi: 10.1016/j.ajhg.2014.11.003) [Epub Dec 4, 2014] [PMID: 25480033] (abstract).
Research highlights in Nature Reviews Genetics (here)

673-OA Makrythanasis P, Temtamy S, Aglan MS, Otaify GA, Hamamy H, **Antonarakis SE**. A novel homozygous mutation in FGFR3 causes tall stature, severe lateral tibial deviation, scoliosis, hearing impairment, camptodactyly and arachnodactyly. **Human Mutation** 35: 959-963, 2014 (doi: 10.1002/humu.22597) [Epub May 26, 2014] [PMID: 24864036] (abstract).

674-OA Sahún I, Marechal D, Lopes Pereira P, Nalesso V, Gruart A, Delgado Garcia JM, **Antonarakis SE**, Dierssen M, Herault Y. Cognition and hippocampal plasticity in the mouse is altered by monosomy of a genomic region implicated in Down syndrome. **Genetics** 197: 899-912, 2014 [Epub Apr 21, 2014] [PMID: 24752061] [IF 4.39] (abstract).

675-OA Hamamy H, Makrythanasis P, Al-Allawi N, Muhsin AA, **Antonarakis SE**. Recessive thrombocytopenia likely due to a homozygous pathogenic variant in the FYB gene: Case Report. **BMC Medical Genetics** 15: 135, 2014 [Epub Dec 17, 2014] [PMID: 25516138] (abstract).

676-OA Bosman A, Letourneau A, Sartiani L, Del Lungo M, Ronzoni F, Kuziakiv R, Tohonen V, Zucchelli M, Santoni F, Guipponi M, Dumevska B, Hovatta O, **Antonarakis SE**, Jaconi ME. Perturbations of Heart Development and Function in Cardiomyocytes from hESC with Trisomy 21. **Stem Cells** 33: 1434-1446, 2015 (doi: 10.1002/stem.1961) [Epub Jan 31, 2015] [PMID: 25645121] [IF 7.13] (abstract).

677-OA Letourneau A, Cobellis G, Fort A, Santoni F, Garieri M, Falconnet E, Ribaux P, Vannier A, Guipponi M, Carninci P, Borel C, **Antonarakis SE**. Single-minded 2 (SIM2), potentially involved in Down syndrome, has properties of super-enhancers and pioneer transcription factors in pluripotent mouse ES cells. **PLoS ONE** 10: e0126475, 2015 (doi: 10.1371/journal.pone.0126475) [Epub May 8, 2015] [PMID: 25955728] (abstract and PDF).

678-OA Brault V, Duchon A, Romestaing C, Sahun I, Pothion S, Karout M, Borel C, Dembele D, Bizot JC, Messaddeq N, Sharp AJ, Roussel D, **Antonarakis SE**, Dierssen M, Herault Y. Opposite Phenotypes of Muscle Strength and Locomotor Function in Mouse Models of Partial Trisomy and Monosomy 21 for the Proximal Hspa13-App Region. **PLoS Genetics** 11: e1005062, 2015 (doi: 10.1371/journal.pgen.1005062) [Epub Mar 24, 2015] [PMID: 25803843] [IF 8.16] (abstract and PDF).

679-OA Borel C, Ferreira PG, Santoni F, Delaneau O, Fort A, Popadin KY, Garieri M, Falconnet E, Ribaux P, Guipponi M, Padioleau I, Carninci P, Dermitzakis ET, **Antonarakis SE**. Biased allelic expression in human primary fibroblast single cells. **American Journal of Human Genetics** 96: 70-80, 2014 (doi: 10.1016/j.ajhg.2014.12.001) [Epub Dec 31, 2014] [PMID: 25557783] (abstract).

680-OA Gannagé-Yared MH, Makrythanasis P, Chouery E, Sobacchie C, Mehawej C, Santoni FA, Guipponi M, **Antonarakis SE**, Hamamy H, Mégarbané A. Exome sequencing reveals a mutation in DMP1 in a family with familial sclerosing bone dysplasia. **Bone** 68: 142-145, 2014 (doi: 10.1016/j.bone.2014.08.014) [Epub Aug 30, 2014] [PMID: 25180662] (abstract).

681-OA Fairfield H, Srivastava A, Ananda G, Liu R, Kircher M, Lakshminarayana A, Harris B, Karst SY, Dionne LA, Kane CC, Curtain M, Berry ML, Ward-Bailey PF, Greenstein I, Byers C, Czechanski A, Sharp J, Palmer K, Gudis P, Martin W, Tadenev A, Bogdanik L, Pratt CH, Chang B, Schroeder DG, Cox GA, Cliften P, Milbrandt J, Murray S, Burgess R, Bergstrom DE, Donahue LR, Hamamy H, Masri A, Santoni FA, Makrythanasis P, **Antonarakis SE**, Shendure J, Reinholdt LG. Exome sequencing reveal pathogenic mutations in 91 strains of mice with Mendelian disorders. **Genome Research** 25: 948-957, 2015 (doi: 10.1101/gr.186882.114) [Epub Apr 27, 2015] [PMID: 25917818] (abstract).

682-OA Greger L, Su J, Rung J, Ferreira PG; Geuvadis consortium of 49 authors including **Antonarakis SE**, Lappalainen T, Dermitzakis ET, Brazma A. Tandem RNA chimeras contribute to transcriptome diversity in human population and are associated with intronic genetic variants. **PLoS ONE** 9: e104567, 2014 (doi: 10.1371/journal.pone.0104567) [Epub Aug 18, 2014] [PMID: 25133550] (abstract).

683-OA Murray A, Letourneau A, Canzonetta C, Stathaki E, Gimelli S, Sloan-Bena F, Abrehart R, Goh P, Lim S, Baldo C, Dagna-Bricarelli F, Hannan S, Mortensen M, Ballard D, Syndercombe Court D, Fusaki N, Hasegawa M, Smart TG, Bishop C, **Antonarakis SE**, Groet J, Nizetic D. Isogenic induced pluripotent stem cell lines from an adult with mosaic Down Syndrome model accelerated neuronal ageing and neurodegeneration. **Stem Cells** 33: 2077-2084, 2015 (doi: 10.1002/stem.1968) [Epub Feb 19, 2015] [PMID: 25694335] [IF 7.13] (abstract).

684-OA Sailani MR, FA, Letourneau A, Borel C, Makrythanasis P, Hibaoui Y, Popadin K, Bonilla X, Guipponi M, Gehrig C, Vannier A, Carre-Pigeon F, Feki A, Nizetic D, **Antonarakis SE**. DNA methylation differences in trisomy 21 using monozygotic twins. **PLoS ONE** 10: e0135555, 2015. (doi: 10.1371/journal.pone.0135555) [Epub Aug 28, 2015] [PMID: 26317209] (abstract and PDF).

685-OA Calvel P, Kusz-Zamelczyk K, Janecki D, Makrythanasis P, Borel C, Conne B, Vannier A, Béna F, Gimelli S, Fichna P, **Antonarakis SE**, Nef S, Jaruzelska J. A case of Wiedemann-Steiner syndrome associated with 46,XY DSD gonadal dysgenesis. Sex Development 9: 289-295, 2015. [Epub Nov 7, 2015] [PMID: 26544196] (abstract).

686-OA Guipponi M, Chentouf A, Webling KEB, Freimann K, Crespel A, Nobile C, Lemke JR, Hansen J, Dorn T, Lesca G, Ryvlin P, Hirsch E, Rudolf G, Rosenberg DS, Becker F, Helbig I, Muhle H, Salzmann A, Chaouch M, Oubaiche ML, Ziglio S, Gehrig C, Santoni F, Pizzato M, Langel U, **Antonarakis SE**. Galanin pathogenic mutations in temporal lobe epilepsy. **Human Molecular Genetics** 24: 3082-3091, 2015 [Epub Feb 17, 2015] [PMID: 25691535] [IF 6.67] (abstract).

687-OA Dieudonné FX, O'Connor PB, Gubler-Jaquier P, Yasrebi H, Conne B, Nikolaev S, **Antonarakis SE**, Baranov PV, Curran J. The effect of heterogeneous transcription start sites (TSS) on the translatome: Implications for the cellular phenotype. **BMC Genomics** 16: 986, 2015. [Epub Nov 21, 2015] [PMID: 26589636] (abstract).

688-OA Fokstuen S, Makrythanasis P, Hammar E, Guipponi M, Ranza E, Varvagiannis K, Santoni FA, Albarca-Aguilera M, Poleggi ME, Couchepin F, Brockmann C, Mauron A, Hurst SA, Moret C, Gehrig C, Bevillard J, Araud T, Gimelli S, Stathaki E, Paoloni-Giacobino A, Bottani A, Sloan-Béna F, D'Amato-Sizonenko L, Mostafavi M, Hamamy H, Nouspikel T, Blouin JL, **Antonarakis SE**. Experience of a multidisciplinary task force with exome sequencing for Mendelian disorders. **Human Genomics** 10: 24, 2016 (doi: 10.1186/s40246-016-0080-4) [Epub Jun 28, 2016] [PMID: 27353043] (abstract).

689-OA Rosa A, Chande A, Ziglio S, De Sanctis V, Bertorelli R, Goh SL, McCauley SM, Nowolsielska A, **Antonarakis SE**, Luban J, Santoni FA, Pizzato M. HIV1 Nef promotes infection by preventing SERINC5 incorporation into the virion membrane. **Nature** 526: 212-217, 2015 (doi: 10.1038/nature15399) [Epub Sep 30, 2015] [PMID: 26416734] (abstract).

690-OA Rivas MA, Pirinen M, Conrad DF, Lek M, Tsang EK, Karczewski KJ, Maller JB, Kukurba KR, DeLuca DS, Fromer M, Ferreira PG, Smith KS, Zhang R, Zhao F, Banks E, Poplin R, Ruderfer DM, Purcell SM, Tukiainen T, Minikel EV, Stenson PD, Cooper DN, Huang KH, Sullivan TJ, Nedzel J, (GTEx Consortium; Geuvadis Consortium), Segre AV, Young TR, Gelfand ET, Trowbridge CA, Ward LD, Kheradpour P, Iriarte B, Meng Y, Palmer CD, Esko T, Winckler W, Hirschhorn J, Kellis M, Getz G, Shablin AA, Li G, Zhou YH, Nobel AB, Rusyn I, Wright FA, Battle A, Mostafavi S, Mele M, Reverter F, Goldmann J, Koller D, Gamazon ER, Im HK, Konkashbaev A, Nicolae DL, Cox NJ, Flutre T, Wen X, Stephens M, Pritchard JK, Tu Z, Zhang B, Huang T,

Long Q, Lin L, Yang J, Zhu J, Liu J, Brown A, Mestichelli B, Tidwell D, Lo E, Salvatore M, Shad S, Thomas JA, Lonsdale JT, Choi RC, Karasik E, Ramsey K, Moser MT, Foster BA, Gillard BM, Syron J, Fleming J, Magazine H, Hasz R, Walters GD, Bridge JP, Miklos M, Sullivan S, Barker LK, Traino H, Mosavel M, Siminoff LA, Valley DR, Rohrer DC, Jewel S, Branton P, Sobin LH, Barcus M, Qi L, Hariharan P, Wu S, Tabor D, Shive C, Smith AM, Buia SA, Undale AH, Robinson KL, Roche N, Valentino KM, Britton A, Burges R, Bradbury D, Hambright KW, Seleski J, Korzeniewski GE, Erickson K, Marcus Y, Tejada J, Taherian M, Lu C, Robles BE, Basile M, Mash DC, Volpi S, Struewing JP, Temple GF, Boyer J, Colantuoni D, Little R, Koester S, Carithers LJ, Moore HM, Guan P, Compton C, Sawyer SJ, Demchok JP, Vaught JB, Rabiner CA, Lockhart NC, Friedlander MR, 't Hoen PA, Monlong J, Gonzàlez-Porta M, Kurbatova N, Griebel T, Barann M, Wieland T, Greger L, van Iterson M, Almlof J, Ribeca P, Pulyakhina I, Esser D, Giger T, Tikhonov A, Sultan M, Bertier G, Lizano E, Buermans HP, Padioleau I, Schwarzmayr T, Karlberg O, Ongen H, Kilpinen H, Beltran S, Gut M, Kahlem K, Amstislavskiy V, Stegle O, Flicek P, Strom TM, Lehrach H, Schreiber S, Sudbrak R, Carracedo A, **Antonarakis SE**, Hasler R, Syvanen AC, van Ommen GJ, Brazma A, Meitinger T, Rosenstiel P, Gut IG, Estivill X.Bustamante CD, Li JB, Daly MJ, Guigo R, Donnelly P, Ardlie K, Sammeth M, Dermitzakis ET, McCarthy MI, Montgomery SB, Lappalainen T, MacArthur DG. Effect of predicted protein-truncating genetic variants on the human transcriptome. **Science** 348: 666-669, 2015 (doi: 10.1126/science.1261877) [Epub May 8, 2015] [PMID: 25954003] (abstract).

691-OA Seplyarskiy VB, Soldatov RA, Popadin KY, **Antonarakis SE**, Bazykin GA, Nikolaev SI. APOBEC-induced mutations in human cancers are strongly enriched on the lagging DNA strand during replication. **Genome Research** 26: 174-182, 2016 [Epub Jan 11, 2016] [PMID: 26755635] (abstract).

692-OA Santoni FA, Makrythanasis P, **Antonarakis SE**. CATCHing putative causative variants in consanguineous families. **BMC Bioinformatics** 16: 310, 2015 (doi: 10.1186/s12859-015-0727-5) [Epub Sep 28, 2015] [PMID: 26415661] (abstract)

693-OA Bonilla X, Parmentier L, King B, Bezrukov F, Kaya G, Seplyarskiy VB, Zoete V, Sharpe HJ, McKee T, Letourneau A, Ribaux PG, Santoni FA, Popadin K, Basset-Seguin N, Ben Chaabene R, Andrianova MA, Guipponi M, Garieri M, Verdan C, Grosdemange K, Sumara O, Eilers M, Aifantis I, Michielin O, de Sauvage FJ, **Antonarakis SE**[§], Nikolaev SI[§] ([§]*corresponding authors*). Genomic analysis reveals novel secondary drivers and progression pathways in skin basal cell carcinoma. **Nature Genetics** 48: 398-406, 2016 (doi: 10.1038/ng.3525) [Epub Mar 7, 2016] [PMID: 26950094] (abstract).

694-OA Chentouf A, Dahdouh A, Guipponi M, Oubaiche ML, Chaouch M, Hamamy H, **Antonarakis SE**. Familial epilepsy in Algeria: Clinical features and inheritance profiles. **Seizure** 31: 12–18, 2015 (doi: 10.1016/j.seizure.2015.06.015) [Epub Jul 3, 2015]. [PMID: 26362371] (abstract).

695-OA Pule GD, Ngo Bitoungui VJ, Chemegni BC, Kengne AP, **Antonarakis SE**, Wonkam A. Association between variants at BCL11A erythroid-specific enhancer and foetal haemoglobin levels among Sickle Cell Disease Patients in Cameroon: implications for future therapeutic interventions. **OMICS Journal of Integrative Biology** 19: 627-631, 2015 (doi: 10.1089/omi.2015.0124) [Epub Sep 22, 2015] [PMID: 26393293] (abstract).

696-OA Makrythanasis P, Kato M, Zaki M, Saitsu H, Nakamura K, Santoni F, Miyatake S, Nakashima M, Issa MY, Guipponi M, Letourneau A, Logan C, Roberts N, Parry DA, Johnson CA, Matsumoto N, Hamamy H, Sheridan E, Kinoshita T, **Antonarakis SE**[§], Murakami Y[§] ([§]labs equally contributed). Pathogenic variants in PIGG cause intellectual disability with seizures and hypotonia. **American Journal of Human Genetics** 98: 615-626, 2016 (doi: 10.1016/j.ajhg.2016.02.007) [Epub Mar 15, 2016] [PMID: 26996948] (abstract).

697-OA Nicastro N, Ranza E, **Antonarakis SE**, Horvath J. Pure progressive ataxia and palatal tremor (PAPT) associated with a new polymerase gamma (POLG) mutation. **Cerebellum** 15:829-831, 2016 [Epub Nov 25, 2015] [PMID: 26607151] (abstract).

698-OA Guipponi M, Santoni F, Schneider M, Gehrig C, Bonilla X, Kates W, Morrow B, Armando M, Vicara S, Shashi V, Hooper SR, Eliez S, **Antonarakis SE**. No evidence for the presence of genetic variants predisposing to psychotic disorders on the non-deleted 22q11.2 allele of VCFS patients. **Translational Psychiatry** 7: e1039, 2017 (doi: 10.1038/tp.2016.258) [Epub Feb 21, 2017] [PMID: 28221368] (abstract).

699-OA Joshi R, Garg P, Zaitlen N, Lappalainen T, Watson CT, Azam N, Ho D, **Antonarakis SE**, Brunner HG, Buiting K, Cheung SW, Coffee B, Eggermann T, Francis D, Geraedts JP, Gimelli G, Jacobson SG, Le Caignec C, de Leeuw N, Liehr T, Mackay DJ, Montgomery SB, Pagnamenta AT, Papenhausen P, Robinson DO, Ruivenkamp C, Schwartz C, Steiner B, Stevenson DA, Surti U, Wassink T, Sharp AJ. DNA methylation profiling of uniparental disomy cases reveals novel imprinted loci in the human genome. **American Journal of Human Genetics** 99: 555-566, 2016 (doi: 10.1016/j.ajhg.2016.06.032) [Epub Aug 24, 2016] [PMID: 27569549] (abstract).

700-CR den Dunnen JT, Dalgleish R, Maglott DR, Hart RK, Greenblatt MS, McGowan-Jordan J, Roux AF, Smith T,

Antonarakis SE, Taschner PEM, on behalf of the Human Genome Variation Society (HGVS), the Human Variome Project (HVP) and the Human Genome Organisation (HUGO). HGVS Recommendations for the Description of Sequence Variants: 2016 Update. **Human Mutation** 37: 564-569, 2016 (doi: 10.1002/humu.22981) [PMID: 26931183] (abstract)

701-OA Ecker S, Chen L, Bagger FO, Pancaldi V, Sidiropoulos N, Rapin N, Fernández JM, Watt S, Carrillo de Santa Pau E, Juan D, Merkel A, Albers CA, Amstislavskiy V, Ashford S, Bomba L, Bourque G, Bujold D, Burden F, Busche S, Caron M, Chen SH, Cheung W, Clarke L , Datta A, Delaneau O, Elding H, Farrow S, Garrido D, Ge B, Iotchkova V, Kundu K, Kwan T, Lambourne JJ, Lowy E, Mann AL, Mead D, Pourfarzad F, Redensek A, Rehnstrom K, Rendon A, Richardson D, Risch T, Rowlston S, Shao X, Simon MM, Sultan M, Walter K, Wilder SP, Yang Y, **Antonarakis SE**, Dermitzakis ET, Flicek P, Guigo R, Lehrach H, Marten JHA, Yaspo ML, Stunnenberg HG, Ouwehand WH, Frontini M, Downes K, Pastinen T, Kuijpers TW, Rico D, Valencia A, Beck S, Soranzo N, Paul DS. Epigenetic and Transcriptional Variability Shapes Phenotypic Plasticity of Human Immune Cells. **Genome Biology** 18: 18, 2017 (doi: 10.1186/s13059-017-1156-8) [Epub Jan 26, 2017] [PMID: 28126036] (abstract).

702-OA Chen L, Ge B, Casale FP, Vasquez L, Kwan T, Garrido-Martín D, Watt S, Yan Y, Kundu K, Ecker S, Datta A, Richardson D, Burden F, Mead D, Mann AL, Fernandez JM, Rowlston S, Wilder SP, Farrow S, Shao X, Lambourne JJ, Redensek A, Albers CA, Amstislavskiy V, Ashford S, Berentsen K, Bomba L, Bourque G, Bujold D, Busche S, Caron M, Chen SH, Cheung W, Delaneau O, Dermitzakis ET, Elding H, Colgiu I, Bagger FO, Flicek P, Habibi E, Iotchkova V, Janssen-Megens E, Kim B, Lehrach H, Lowy E, Mandoli A, Matarese F, Maurano MT, Morris JA, Pancaldi V, Pourfarzad F, Rehnstrom K, Rendon A, Risch T, Sharifi N, Simon MM, Sultan M, Valencia A, Walter K, Wang SY, Frontini M, **Antonarakis SE**, Clarke L, Yaspo ML, Beck S, Guigo R, Rico D, Martens JH, Ouwehand WH, Kuijpers TW, Paul DS, Stunnenberg HG, Stegle O, Downes K, Pastinen T, Soranzo N. Genetic Drivers of Epigenetic and Transcriptional Variation in Human Immune Cells. **Cell** 167: 1398-1414, 2016 (doi: 10.1016/j.cell.2016.10.026) [Epub Nov 17, 2016] [PMID: 27863251] (abstract).

703-OA Fort A, Panousis NI, Garieri M, **Antonarakis SE**, Lappalainen T, Dermitzakis ET, Delaneau O. MBV: a method to solve sample mislabeling and detect technical bias in large combined genotype and sequencing assay data sets. **Bioinformatics** in press 2017 (doi: 10.1093/bioinformatics/btx074) [Epub Feb 10, 2016] [PMID: 28186259].

704-R **Antonarakis SE.** Down syndrome and the complexity of genome dosage imbalance. **Nature Reviews Genetics** 18: 147-163, 2017 (doi: 10.1038/nrg.2016.154) [Epub Dec 28, 2016] [PMID: 28029161] (abstract).

705-OA Popadin K, Peischl S, Garieri M, Sailani MR, Letourneau A, Santoni F, Lukowski SW, Bazykin G, Nikolaev S , Meyer D, Excoffier L, Reymond A, **Antonarakis SE.** Slightly deleterious genomic variants and transcriptome perturbations contribute to the embryonic selection of Down syndrome. **Genome Research** 28:1-10, 2018 (doi: 10.1101/gr.228411.117) [Epub Dec 13, 2017] [PMID: 29237728] (abstract).

706-OA Olcese C, Patel MP, Shoemark A, Kiviluoto S, Legendre M, Williams HJ, Vaughan C, Hayward J, Goldenberg A, Emes RD, Munye M, Dyer L, Cahill T, Bevillard J, Gehrig C, Guipponi M, Chantot S, Duquesnoy P, ThomasL, Jeanson L, Copin B, Tamalet A, Thauvin C, Papon JF, Garin A, Pin I, Fassad M, Jenkins L, Boustred C, Cullup T, Dixon M, Onoufriadis A, Bush A, Chung EMK, **Antonarakis SE**, Loebinger MR, Wilson R, Armengot M, Escudier E, Hogg C, UK10K, Amselem S, Sun Z, Bartoloni L, Blouin JL, Mitchison HM. X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor PIH1D3. **Nature Communications** 8: 14279, 2017 (doi: 10.1038/ncomms14279) [Epub Feb 8, 2017] [PMID: 28176794] (abstract).

707-OA Makrythanasis P, Guipponi M, Santoni FA, Zaki M, Issa MY, Ansar M , Hamamy H, **Antonarakis SE.** Exome sequencing discloses KALRN homozygous variant as likely cause of intellectual disability and short stature in a consanguineous pedigree. **Human Genomics** 10: 26, 2016 (doi: 10.1186/s40246-016-0082-2) [Epub Jul 16, 2016] [PMID: 27421267] (abstract).

708-Book Kumar D & **Antonarakis SE** eds. Medical and Health Genomics. 1st Edition. Academic Press; (ISBN: 9780124201965) Pgs 358, 2016.

709-BC **Antonarakis SE.** Content and Variation of the Human Genome. In Kumar D and Antonarakis SE "Medical and Health Genomics". 1st Ed. Academic Press; (ISBN: 9780124201965) pp 161-177, 2016.

710-OA Santoni FA, Stamoulis G, Garieri M, Falconnet E, Ribaux P, Borel C, **Antonarakis SE.** Detection of Imprinted Genes by Single-Cell Allele-Specific Gene Expression. **American Journal of Human Genetics** 100: 444-453, 2017 (doi: 10.1016/j.ajhg.2017.01.028) [Epub Feb 6, 2017] [PMID: 28190458] (abstract).

711-OA Sartori A, Cheng P, Falconnet E, Ribaux P, Aubry-Lachainaye JP, Levesque MP, **Antonarakis SE,** Matthes T, Borel C. Single-cell mRNA profiles of leukemic blasts reveal a transcriptional regulatory circuitry with potential value for the

classification and prognosis of acute myeloid leukemia. Biorxiv.org posted May 31, 2017 (doi: https://doi.org/10.1101/141754) (abstract).

712-OA Stévant I, Neirjinck Y, Borel C, Escoffier J, Smith LB, **Antonarakis SE**, Dermitzakis ET, Nef S. Deciphering Cell Lineage Specification during Male Sex Determination with Single-Cell RNA Sequencing. **Cell Reports** 22: 1589-1599, 2018 (doi: 10.1016/j.celrep.2018.01.043) [Epub Feb 6, 2018] [PMID: 29425512] (abstract).
Posted in www.biorxiv.org/content/early/2017/09/18/190264.

713-OA Liu Y, Borel C, Li L, Muller T, Williams EW, Germain PL, Buljan M, Sajic T, Boersema P, Shao W, Faini M, Testa G, Beyer A, **Antonarakis SE**[§], Aebersold R[§] ([§]co-corresponding authors). Systematic proteome and proteostasis profiling in human Trisomy 21 fibroblast cells. **Nature Communications** 8: 1212, 2017 (doi: 10.1038/s41467-017-01422-6) [Epub Oct 31, 2017] [PMID: 29089484] (abstract).

714-OA Ferreira PG, Oti M, Barann M, Wieland T, Ezquina S, Friedländer MR, Rivas MA, Esteve-Codina A, The GEUVADIS Consortium (Estivill X, Dermitzakis E, **Antonarakis SE**, Meitinger T, Palotie A, Deleuze JF, Sudbrak R, Lerach H, Gut I, Syvänen AC, Gyllensten U, Schreiber S, Brunner H, Veltman J Hoen PCT, van Ommen GJ, Carracedo A, Brazma A, Flicek P, Cambon-Thomsen A, Mangion J, Bentley D, Hamosh A), Rosenstiel P, Strom TM, Lappalainen T, Guigó R, Sammeth M. Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing. **Scientific Reports** 6: 32406, 2016. (doi: 10.1038/srep32406) [Epub Sep 12, 2016] [PMID: 27617755] (abstract).

715-OA Guo T, Repetto G, McGinn DM, Chung JH, Nomaru H, Campbell CL, Blonska A, Bassett AS, Chow EWC, Mlynarski EE, Swillen A, Vermeesch J, Devriendt K, Gothelf D, Carmel M, Michaelovsky E, Schneider M, Eliez S, **Antonarakis SE**, Coleman K, Tomita-Mitchell A, Mitchell ME, Digilio MC, Dallapiccola B, Marino B, Philip N, Busa T, Kushan L, Barden CE, Piotrowicz M, Hawuła W, Roberts AE, Tassone F, Simon TJ, van Duin E, van Amelsvoort TA, Kates WR, Zaikai E, Johnston HR, Cutler DJ, Agopian AJ, Mitchell LE, Goldmuntz E, Wang T, Emanuel BS, Morrow BE and the International 22q11.2 Consortium/Brain and Behaviour Consortium. Genome-wide association study of modifiers for tetralogy of Fallot in 22q11.2DS identifies variants in the GPR98 locus on 5q14.3. **Circulation: Cardiovascular Genetics** 10: e001690, 2017 (doi: 10.1161/CIRCGENETICS.116.001690) [PMID: 29025761] (abstract).

716-OA Nikolaev SI, Vetiska S, Bonilla X, Boudreau E, Jauhiainen S, Jahromi BR, Khyzha N, Kinnunen S, Kiehl TR, Mendes Pereira V, Krings T , Andrade H, Tung T, Valiante T, Zadeh G, Tymianski M, Rauramaa T, Ylä-Herttuala S, **Antonarakis SE**, Frösen J, Fish JE, Radovanovic I. Brain arteriovenous malformations are caused by somatic activating KRAS mutations in endothelial cells. **New England Journal of Medicine** 378: 250-261, 2018 (doi: 10.1056/NEJMoa1709449) [Epub Jan 3, 2018] [PMID: 29298116] (abstract).

717-OA Gaspar L, Howald C, Popadin K, Maier B, Mauvoisin D, Moriggi E, Gutierrez-Arcelus M, Falconnet E, Borel C, Kunz D, Kramer A, Gachon F, Dermitzakis ET, **Antonarakis SE**, Brown SA. The genomic landscape of human cellular circadian variation points to a novel role for the human signalosome. **Elife**. 6: e24994, 2017 (doi: 10.7554/eLife.24994) [Epub Sep 4, 2017] [PMID: 28869038] (abstract and PDF).

718-R Stunnenberg HG; International Human Epigenome Consortium (228 collaborators: Abrignani S, Adams D, de Almeida M, Altucci L, Amin V, Amit I, **Antonarakis SE**, Aparicio S, Arima T, Arrigoni L, Arts R, Asnafi V, Badosa ME, Bae JB, Bassler K, Beck S, Berkman B, Bernstein BE, Bilenky M, Bird A, Bock C, Boehm B, Bourque G, Breeze CE, Brors B, Bujold D, Burren O, Bussemakers MJ, Butterworth A, Campo E, Carrillo-de-Santa-Pau E, Chadwick L, Chan KM, Chen W, Cheung TH, Chiapperino L, Choi NH, Chung HR, Clarke L, Connors JM, Cronet P, Danesh J, Dermitzakis M, Drewes G, Durek P, Dyke S, Dylag T, Eaves CJ, Ebert P, Eils R, Eils J, Ennis CA, Enver T, Feingold EA, Felder B, Ferguson-Smith A, Fitzgibbon J, Flicek P, Foo RS, Fraser P, Frontini M, Furlong E, Gakkhar S, Gasparoni N, Gasparoni G, Geschwind DH, Glažar P, Graf T, Grosveld F, Guan XY, Guigo R, Gut IG, Hamann A, Han BG, Harris RA, Heath S, Helin K, Hengstler JG, Heravi-Moussavi A, Herrup K, Hill S, Hilton JA, Hitz BC, Horsthemke B, Hu M, Hwang JY, Ip NY, Ito T, Javierre BM, Jenko S, Jenuwein T, Joly Y, Jones SJ, Kanai Y, Kang HG, Karsan A, Kiemer AK, Kim SC, Kim BJ, Kim HH, Kimura H, Kinkley S, Klironomos F, Koh IU, Kostadima M, Kressler C, Kreuzhuber R, Kundaje A, Küppers R, Larabell C, Lasko P, Lathrop M, Lee DH, Lee S, Lehrach H, Leitão E, Lengauer T, Lernmark Å, Leslie RD, Leung GK, Leung D, Loeffler M, Ma Y, Mai A, Manke T, Marcotte ER, Marra MA, Martens JH, Martin-Subero JI, Maschke K, Merten C, Milosavljevic A, Minucci S, Mitsuyama T, Moore RA, Müller F, Mungall AJ, Netea MG, Nordström K, Norstedt I, Okae H, Onuchic V, Ouellette F, Ouwehand W, Pagani M, Pancaldi V, Pap T, Pastinen T, Patel R, Paul DS, Pazin MJ, Pelicci PG, Phillips AG, Polansky J, Porse B, Pospisilik JA, Prabhakar S, Procaccini DC, Radbruch A, Rajewsky N, Rakyan V, Reik W, Ren B, Richardson D, Richter A, Rico D, Roberts DJ, Rosenstiel P, Rothstein M, Salhab A, Sasaki H, Satterlee JS, Sauer S, Schacht C, Schmidt F, Schmitz G, Schreiber S, Schröder C, Schübeler D, Schultze JL, Schulyer RP, Schulz M, Seifert M, Shirahige K, Siebert R, Sierocinski T, Siminoff L, Sinha A, Soranzo N, Spicuglia S, Spivakov M, Steidl C, Strattan JS, Stratton M, Südbeck P, Sun H, Suzuki N, Suzuki Y, Tanay A, Torrents D, Tyson FL, Ulas T, Ullrich S, Ushijima T, Valencia A, Vellenga E, Vingron M, Wallace C, Wallner S, Walter J, Wang H, Weber S, Weiler N, Weller A, Weng A, Wilder S, Wiseman SM, Wu AR, Wu Z, Xiong J, Yamashita Y, Yang X, Yap DY, Yip KY, Yip S, Yoo JI, Zerbino D, Zipprich G), Hirst M. The International

Human Epigenome Consortium: A Blueprint for Scientific Collaboration and Discovery. **Cell** 167: 1145-1149, 2016 (doi: 10.1016/j.cell.2016.11.007) [Epub Nov 17, 2016] [PMID: 27863232] (abstract).

719-OA Assoum M, Philippe C, Isidor B, Perrin L, Makrythanasis P, Sondheimer N, Paris C, Douglas J, Lesca G, **Antonarakis SE**, Hamamy H, Jouan T, Duffourd Y, Auvin S, Saunier A, Begtrup A, Nowak C, Chatron N, Ville D, Mireskandari K, Milani P, Jonveaux P, Lemeur G, Milh M, Amamoto M, Kato M, Nakashima M, Miyake N, Matsumoto N, Masri A, Thauvin-Robinet C, Rivière JB, Faivre L, Thevenon J. Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. **American Journal of Human Genetics** 99: 1368-1376, 2016 (doi: 10.1016/j.ajhg.2016.10.009) [Epub Nov 16, 2016] [PMID: 27889060] (abstract).

720-OA Andrianova M, Chetan GK, Sibin MK, Mckee T, Merkler D, Narasinga Rao KVL, Ribaux P, Blouin JL, Makrythanasis P, Seplyarskiy VB, **Antonarakis SE**, Nikolaev SI. Germline PMS2 and somatic POLEexo mutations cause hypermutability of the leading DNA strand in the bMMRD syndrome brain tumors. **Journal of Pathology** 243: 331-341, 2017 (doi: 10.1002/path.4957) [Epub Sep 28, 2017] [PMID: 28805995] (abstract)

721-OA Garieri M, Stamoulis G, Blanc X, Falconnet E, Ribaux P, Borel C, Santoni F, **Antonarakis SE.** Extensive cellular heterogeneity of X inactivation revealed by single-cell allele-specific expression in human fibroblasts. **Proceedings of the National Academy of Sciences USA** 115: 13015-13020, 2018 (doi: 10.1073/pnas.1806811115)[Epub Dec 3, 2018] [PMID: 30510006] (abstract) . Biorxiv.org posted Apr 10, 2018 (doi: https://doi.org/10.1101/298984).

722-OA Ansar M, Riazuddin S, Makrythanasis P, Paracha SA, Iqbal Z, Sarwar MT, Khan J, Assir MZ, Hussain M, Razzaq A, Lopo Polla D, Suhail A, Holmgren A, Misceo D, Iwaszkiewicz J, de Brouwer APM, Guipponi M, Santoni FA, Hanquinet S, Zoete V, Frengen E, Ahmed J, van Bokhoven H, Riazuddin S, **Antonarakis SE.** Biallelic variants in LINGO1 are associated with autosomal recessive intellectual disability, speech and motor delay, and microcephaly. **Genetics in Medicine** 20: 778-784, 2018 (doi: 10.1038/gim.2017.113) [Epub Aug 24, 2017] [PMID: 28837161] (abstract).

723-Let **Antonarakis SE.** Genomic databases, a WHO affair! **Science** 356: 812-813, 2017 (doi: 10.1126/science.aan4717) [Epub May 26, 2016] [PMID: 28546180].

724-OA Ranza E, Garcia-Tarodo S, Varvagiannis K, Guipponi M, Lobrinus JA, Bottani A, Kern I, Kurian M, Pittet MP, **Antonarakis SE**, Fluss J, Korff CM. SERPINI1 pathogenic variants: an emerging cause of childhood-onset progressive myoclonic epilepsy. **American Journal of Medical Genetics** 173: 2456-2460, 2017 (doi: 10.1002/ajmg.a.38317) [Epub Jun 20, 2017] [PMID: 28631894] (abstract).

725-OA Garieri M, Delaneau O, Santoni F, Fish R, Mull D, Carninci P, Dermitzakis ET, **Antonarakis SE**, Fort A. The effect of genetic variation on promoter usage and enhancer activity. **Nature Communications** 8: 1358, 2017 (doi: 10.1038/s41467-017-01467-7) [Epub Nov 7, 2017] [PMID: 29116076] (abstract).

726-OA Miranda AM, Herman M, Cheng R, Nahmani E, Barrett G, Micevska E, Fontaine G, Potier MC, Head E, Schmitt FA, Lott IT, Jimanez-Velazquez IZ, **Antonarakis SE**, Di Paolo G, Lee JH, Hussaini SA, Marquer C. Excess Synaptojanin 1 Contributes to Place Cell Dysfunction and Memory Deficits in the Aging Hippocampus in Three Types of Alzheimer's Disease. **Cell Reports** 23: 2967-2975, 2018 (doi: 10.1016/j.celrep.2018.05.011) [Epub Jun 5, 2018] [PMID: 29874583] (abstract).

727-OA Makrythanasis P, Maroofian R, Stray-Pedersen A, Musaev D, Zaki MS, Mahmoud IG, Selim L, Elbadawy A, Jhangiani SN, Coban Akdemir ZH, Gambin T, Sorte HS, Heiberg A, McEvoy-Venneri J, James KN, Stanley V, Belandres D, Guipponi M, Santoni FA, Ahangari N, Tara F, Doosti M, Iwaszkiewicz J, Zoete V, Backe PH, Hamamy H, Gleeson JG, Lupski JR, Karimiani EG, **Antonarakis SE.** Biallelic variants in KIF14 cause intellectual disability with microcephaly. **European Journal of Human Genetics** 26: 330-339, 2018 (doi: 10.1038/s41431-017-0088-9) [Epub Jan 17, 2018] [PMID: 29343805] (abstract).

728-E Auffray C; Sagner M; Abdelhak S; Adcock I; Agusti A; Amaral M; **Antonarakis SE**; Arena R; Argoul F; Balling R; Barabasi AL; Beckmann J; Bjartell A; Blomberg N; Bourgeron T; Boutron B; Brahmachari S; Bréchot C; Brightling C; Cascante M; Cesario A; Charron C; Chen SJ; Chen Z; Chung F; Clément K; Conesa A; Cozzone A; de Jong M; Deleuze JF; Demotes J; di Meglio A; Djukanovic R; Dogrusoz U; Epel E; Fischer A; Gelemanovic A; Goble C; Gojobori T; Goldman M; Goossens H; Gros F; Guo YK; Hainaut P; Harrison D; Hoffmann H; Hood L; Hunter P; Jacob Y; Kitano H; Klingmüller U; Knoppers B; Kolch W; Koopmans M; Lancet D; Laville M; Lehn JM; Lévi F; Lisistsa A; Lotteau V; Magnan A; Mayosi B; Metspalu A; Moreau Y; N'Dow J; Nicod L; Noble D; Nogueira MM; Norrby-Teglund A; Nottale L; Openshaw P; Oztürk M; Palkonen S; Parodi S; Pellet J; Polasek O; Price N; Pristipino C; Radstake T; Raes M; Roca J; Rozman D; Sabatier P; Sasson S; Schmeck B; Serageldin I; Simonds A; Soares B; Sterk P; Superti-Furga G; Supple D; Tegner J; Uhlen M; van der Werf S; Villoslada P; Vinciguerra M; Volpert V; Webb S; Wouters E; Sanz F; Nobrega F. Viva Europa, a Land of Excellence in Research and Innovation for Health and Wellbeing. **Progress in Preventive Medicine**. In press 2017.

729-OA Delaneau O, Zazhytska M, Borel C, Howald C, Kumar S, Ongen H, Popadin K, Marbach D, Bielser D, Hacker D, Romano-Palumbo L, Ribaux P, Wiederkehr M, Falconnet E, Bucher P, Bergmann S, **Antonarakis SE**, Reymond A, Dermitzakis ET. Intra- and inter-chromosomal chromatin interactions mediate genetic effects on gene regulation. **Science** 364: 452, 2019. (pii: eaat8266. doi: 10.1126/science.aat8266) [Epub May 3, 2019] [PMID: 31048460] (abstract).
Biorxiv posted http://www.biorxiv.org/content/early/2017/08/03/171694.

730-CR Florin MV, Escher G, **Antonarakis SE**, Beretta I, Busshoff U, Chiaooerino L, Fellay J, Galasso I, Garden H, Hafen E, Hanselman D, Hurst S, Johnson R, Lovis C, Renn O, Rion Logean S. Policy Brief: A RoadMap for the development of Precision Medicine. EPFL International Risk Governance Center IRGC 2017.

731-OA Varvagiannis K, Hanquinet S, Billieux MH, De Luca R, Rimensberger P, Lidgren M, Guipponi M, Makrythanasis P, Blouin JL, **Antonarakis SE**, Steinfeld R, Kern I, Poretti A, Fluss J, Fokstuen S. Congenital Neuronal Ceroid Lipofuscinosis with a Novel CTSD Gene Mutation: A Rare Cause of Neonatal-Onset Neurodegenerative Disorder. **Neuropediatrics** 49: 150-153, 2018 (doi: 10.1055/s-0037-1613681) [Epub Dec 28, 2017] [PMID: 29284168] (abstract).

732-OA Lambert N, Dauve C, Ranza E, Makrythanasis P, Santoni F, Sloan-Béna F, Gimelli S, Blouin JL, Guipponi M, Bottani A, **Antonarakis SE**, Kosel M, Fluss J, Giacobino A. Novel NEXMIF pathogenic variant in a boy with severe autistic features, intellectual disability and epilepsy, and his mildly affected mother. **Journal of Human Genetics** 63: 847-850, 2018 (doi: 10.1038/s10038-018-0459-2) [Epub May 1, 2018] [PMID: 29717186] (abstract).

733-OA Ansar M, Chung H, Waryah YM, Makrythanasis P, Falconnet E, Rao AR, Guipponi M, Narsani AK, Fingerhut R, Santoni FA, Ranza E, Waryah AM, Bellen HJ, **Antonarakis SE**. Visual impairment and progressive phthisis bulbi caused by recessive pathogenic variant in MARK3. **Human Molecular Genetics** 27: 2703–2711, 2018 (doi: 10.1093/hmg/ddy180) [Epub May 6, 2018] [PMID: 29771303] (abstract).

734-OA Liu Y, Mi Y, Mueller T, Kreibich S, Williams EG, Van Drogen A, Borel C, Germain PL, Frank M, Bludau I, Mehnert M, Seifert M, Emmenlauer M, Bezrukov F, Sloan Bena F, Zhou H, Testa G, Saez-Rodriguez J, **Antonarakis SE**, Hardt WD, Aebersold R. Genomic, proteomic and phenotypic heterogeneity in HeLa cells across laboratories: Implications for reproducibility of research results. **Nature Biotechnology** 37: 314-322, 2019 (doi.org/10.1038/s41587-019-0037-y) [Epub Feb 18, 2019] [PMID: 30778230] (abstract).
Biorxiv.org posted April 30, 2018 (doi: https://doi.org/10.1101/307421).

735-OA Ranza E, Z'Graggen W, Lidgren M, Beghetti M, Guipponi M, **Antonarakis SE**, Absoud M, Goyal S, Pal DK, Korff CM. SCN8A heterozygous variants are associated with Anoxic-Epileptic Seizures. **American Journal of Medical Genetics** 182: 1209-1216, 2020 (doi: 10.1002/ajmg.a.61513) [Epub Feb 10, 2020] [PMID: 32040247] (abstract).

736-OA Stamoulis G, Garieri M, Makrythanasis P, Letourneau A, Guipponi M, Panousis N, Falconnet E, Ribaux P, Borel C, Santoni F, **Antonarakis SE**. Single cell transcriptome in aneuploidies reveals mechanisms of gene dosage imbalance. **Nature Communications** 10: 4495, 2019 (doi: 10.1038/s41467-019-12273-8), [Epub Oct 3, 2019] [PMID: 31582743] (abstract).
Biorxiv.org posted Sep 23, 2018 (doi: https://doi.org/10.1101/424887).

737-R Faundez V, De Toma I, Bardoni B, Bartesaghi R, Nizetic D, de la Torre R, Cohen Kadosh R, Herault Y, Dierssen M, Potier MC; Down Syndrome and Other Genetic Developmental Disorders ECNP Network (**Antonarakis SE**, Contestabile A, Coppus T, De Deyn P, Dekker A, Delabar JM, Fisher E, Martinez-Cué C, Strydom A). Translating molecular advances in Down syndrome and Fragile X syndrome into therapies. **European Neuropsychopharmacology** in press 2018 (doi: 10.1016/j.euroneuro.2018.03.006) [Epub Jun 8, 2018] [PMID: 29887288] (abstract).

738-OA Rehman AU, Najafi M, Kambouris M, Al Gazali L, Makrythanasis P, Aboulfazl R, Maroofian R, Rajab A, Hunter JV, Bakey Z, Tokita M, He W, Vetrini F, Petersen A, Wu K, Al Jasmi F, Helmstädter M, Xia F, Liu P, Ghayoor Karimiani E, Karami Madani GR, El-Shanti H, Eng CM, **Antonarakis SE**, Hertecant J, Walkiewicz M, Schmidts M, Yang Y. Biallelic loss of function variants in PPP1R21 impair endocytotic functions and cause a neurodevelopmental syndrome reminiscent of a lysosomal storage disorder. **Human Mutation** 40: 267-280, 2019. (doi: 10.1002/humu.23694) [Epub Dec 6, 2018] [PMID: 30520571] (abstract).

739-OA Ansar M, Chung H, Taylor R, Nazir A, Imtiaz S, Sarwar MT, Manousopoulou A, Makrythanasis P, Qureshi S, Falconnet E, Guipponi M, Pournaras CJ, Ranza E, Santoni FA, Ahmed J, Shah I, Gul K, Black G, Bellen H, **Antonarakis SE**. Biallelic loss-of-function variants in DNMBP cause infantile cataract. **American Journal of Human Genetics** 103: 568-578, 2018 (doi: 10.1016/j.ajhg.2018.09.004) [Epub Oct 4, 2018] [PMID: 30290152] (abstract).

740-BC **Antonarakis SE**, Cooper DN. Human Genomic Variants and Inherited Disease; Molecular Mechanisms and Clinical Consequences. Chapter 7 in Connor JM, Pyeritz RE, Korf BR, eds. "Emery and Rimoin's Principles and Practice of Medical

Genetics" 7th ed. Churchill Livingstone, in press 2019.

741-OA Ansar M, Aziz Paracha SA, Serretti A, Sarwar MT, Khan J, Ranza E, Falconnet E, Iwaszkiewicz J, Fahim Shah S, Ali A, Santoni FA, Zoete V, Megarbane A, Ahmed J, Colombo R, Makrythanasis P, **Antonarakis SE.** Biallelic loss-of-function variants in FBXL3 cause intellectual disability, delayed motor development and short stature. **Human Molecular Genetics** 28: 972–979, 2019 (doi: 10.1093/hmg/ddy406) [Epub Nov 16, 2018] [PMID: 30481285] (abstract).

742-BC **Antonarakis SE.** Genetic Medicine. Chapter on the Swiss Academies Communications booklet, "Personalized Medicine" Vol 14, #6, pgs 36-59, 2019. ISSN 2297-1823 (doi http://doi.org/10.5281/zenodo.3271401). A Rauch et al editors.

743-R **Antonarakis SE.** Carrier screening for Recessive Disorders. **Nature Reviews Genetics** 20: 549-561, 2019 (doi: 10.1038/s41576-019-0134-2) [Epub May 29, 2019] [PMID: 31142809] (abstract).

744-OA Ansar M, Ullah F, Adams DJ, Lai A, Paracha SA, Sarwar MT, Iwaszkiewicz J, Agha Z, Pais L, Millan F, Falconnet E, Ranza E, Zoete V, Makrythanasis P, Santoni FA, Ahmed J, Katsanis N, Walsh C, Davis EE, **Antonarakis SE.** Recessive variants in DYNC1I2 cause syndromic microcephaly with intellectual disability, cerebral malformations and dysmorphic facial features. **American Journal of Human Genetics** 104: 1073-1087, 2019 (doi: 10.1016/j.ajhg.2019.04.002) [Epub Apr 28, 2019] [PMID: 31079899] (abstract).

745-R **Antonarakis SE,** Skotko BG, Rafii MS, Strydom A, Pape S, Bianchi DW, Sherman SL, Reeves RH. Down Syndrome. **Nature Reviews Disease Primers** 6: 9, 2020 (doi: 10.1038/s41572-019-0143-7). [Epub Feb 6, 2020] [PMID: 32029743] Review (abstract).

746-OA Humbert* J, Salian* S, Makrythanasis* P, Lemire G, Rousseau J, Ehresmann S, Garcia T, Alasiri R, Bottani A, Hanquinet S, Beaver E, Heeley J, Smith ACM, Berger SI, **Antonarakis SE,** Yang XJ, Côté J, Campeau P. De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. American Journal of Human Genetics 107: 564-574, 2020 (doi: 10.1016/j.ajhg.2020.08.002) [Epub Aug 21, 2020] [PMID: 32822602] (abstract).

747-OA *Vaclavik V, Holzer FJ, Ranza E, Guipponi M, **Antonarakis SE,** Rossillion B, Munier FL, Schorderet DF. Novel compound heterozygote mutations in CYP2U1 can cause maculopathy with or without hereditary spastic paraplegia HSP56. Submitted 2020.*

748-OA Ansar M, Chung HL, Al-Otaibi A, Elagabani MN, Ravenscroft TA, Paracha SA, Scholz R, Magid TA, Sarwar MT, Shah SF, Ali Qaisar A, Makrythanasis P, Marcogliese PC, Kamsteeg EJ, Falconnet E, Ranza E, Santoni FA, Aldhalaan H, Al-Asmari A, Ali Faqeih E, Ahmed J, Kornau HC, Bellen H, **Antonarakis SE.** Biallelic variants in IQSEC1 cause intellectual disability, developmental delay and short stature. **American Journal of Human Genetics** 105: 907-920, 2019 (doi: 10.1016/j.ajhg.2019.09.013) [Epub Sep 27, 2019] [PMID: 31607425] (abstract).

749-OA Ansar M, Ranza E, Shetty M, Paracha SA, Azam M, Kern I, Iwaszkiewicz J, Farooq O, Pournaras CJ, Malcles A, Kecik M, Muzaffar W, Qurban A, Ali L, Aggoun Y, Rivolta C, Santoni FA, Makrythanasis P, Ahmed J, Qamar R, Sarwar MT, Henry LK, **Antonarakis SE.** Taurine treatment of retinal degeneration and cardiomyopathy in a consanguineous family with SLC6A6 taurine transporter deficiency. **Human Molecular Genetics** 29: 618–623, 2020 (doi: 10.1093/hmg/ddz303) [Epub Dec 31, 2019] [PMID: 31903486] (abstract).

750-OA Manole A, Efthymiou S, O'Connor E, Mendes MI, Jennings M, Jenkins D, Rodriguez Lopez M, Maroofian R, Salpietro V, Harripaul R, Badalato L, Francklyn CS, Athanasiou-Fragkouli A, Sullivan R, Davagnanam I, Mankad K, Seda M, Desai S, Baranano K, Zafar F, Rana N, Ilyas M, Horga A, Kara M, Mattioli F, Goldenberg A, Griffin H, Piton A, Henderson LB, Kara B, Dilruba Aslanger A, Raaphorst J, Pfundt R, Portier R, Shinawi M, Kirby A, Christensen KM, Wang L, Rosti RO, Cytrynbaum C, Paracha SA, Sarwar MT, SYNAPS Study Group, Ahmed J, Santoni FA, Ranza E, Iwaszkiewicz J, Weksberg R, Person RE, Si Y, Telegrafi A, Andrews MV, Baldridge D, Gabriel H, Mohr J, Oehl-Jaschkowitz B, Debard S, Senger B, Fischer F, van Ravenwaaij C, Fock AJM, Stevens SJC, Bähler J, Nasar A, Mantovani JF, Manzur A, Sarkozy A, Francklyn C, Smith DEC, Salomons GS, Ahmed ZM, Riazuddin S, Riazuddin S, Usmani MA, Ansar M, Ansar M, **Antonarakis SE,** Vincent JB, Ayub M, Grimmel M, Jelsig AM, Duelund Hjortshøj T, Gásdal Karstensen H, Haack TB, Distelmaier F, Horvath R, Gleeson JG, Becker H, Mandel JL, Koolen DA, Houlden H. De-novo and biallelic pathogenic variants in NARS1 cause neurodevelopmental delay due to dominant negative and partial loss-of-function effect. **American Journal of Human Genetics** 107: 311-324, 2020 (doi: 10.1016/j.ajhg.2020.06.016) [Epub Jul 31, 2020] [PMID: 32738225] (abstract).

751-OA Zhao Y, Diacou D, Johnston HR, Musfee FI, McDonald-McGinn DM, McGinn D, Crowley TB, Repetto GM, Swillen A, Breckpot J, Vermeesch JR, Kate WR, M. Digilio MC, Unolt M, Marino B, Pontillo M, Armando M, Di Fabio F, Vicari S, van den Bree M, Moss H, Owen M, Murphy KC, Murphy C, Murphy D, Schoch K, Shashi V, Tassone F, Simon TJ, Shprintzen RJ,

Campbell L, Philip N, Heine-Suñer D, García-Miñaúr S, Fernández L, International 22q11.2 Brain and Behavior Consortium (39 additional authors including **Antonarakis SE**), Bearden CE, Vingerhoets C, van Amelsvoort T, Eliez S, Schneider M, Vorstman J, Gothelf D, Zackai E, Agopain AJ, Gur RE, Bassett AS, Emanuel BS, Goldmuntz E, Mitchell LE, Wang T, Morrow BE. Complete sequence of the 22q11.2 allele in 1,053 subjects 1 with 22q11.2 deletion syndrome reveals modifiers of conotruncal heart defects. **American Journal of Human Genetics** 106: 26-40, 2020 (doi: 10.1016/j.ajhg.2019.11.010) [Epub Dec 20, 2019] [PMID: 31870554] (abstract).

752-OA Küry S, Zhang J, Besnard T, Caro-Llopis A, Zeng X, Robert SM, Josiah SS, Kiziltug E, Denommé-Pichon AS, Cogné B, Kundishora AJ, Hao LT, Li H, Stevenson RE, Louie RJ, Deb W, Torti E, Vignard V, McWalter K, Raymond FL, Rajabi F, Ranza E, Grozeva D, Coury SA, Blanc X, Brischoux-Boucher E, Keren B, Õunap K, Reinson K, Ilves P, Wentzensen IM, Barr EE, Heide Guihard S, Charles P, Seaby EG, Monaghan KG, Rio M, van Bever Y, van Slegtenhorst M, Chung WK, Wilson A, Quinquis D, Bréhéret F, Retterer K, Lindenbaum P, Scalais E, Rhodes L, Stouffs K, Pereira EM, Berger SM, Milla SS, Jaykumar AB, Cobb MH, Panchagnula S, Duy PQ, Vincent M, Mercier S, Gilbert-Dussardier B, Le Guillou X, Audebert-Bellanger S, Odent S, Schmitt S, Boisseau P, Bonneau D, Toutain A, Colin E, Pasquier L, Redon R, Bouman A, Rosenfeld JA, Friez AJ, Pérez-Peña H, Raza Akhtar Rizvi S, Haider S, **Antonarakis SE**, Schwartz CE, Martínez F, Bézieau S, Kahle KT, Isidor B. Rare pathogenic variants in WNK3 cause X-linked intellectual disability. **Genetics in Medicine** 24:1941-1951, 2022 (DOI: 10.1016/j.gim.2022.05.009) [Epub Jun 9, 2022] [PMID: 35678782] (abstract)

753-OA Taylor MR, Martin EA, Sinnen B, Trilokekar R, Ranza E, **Antonarakis SE**, Williams ME. KIRREL3 mediated synapse formation is attenuated by disease-associated missense variants. **Journal of Neuroscience** 40: 5376-5388, 2020 (doi: 10.1523/JNEUROSCI.3058-19.2020) [Epub Jun 5, 2020] [PMID: 32503885] (abstract)
Biorxiv.org posted Dec 30, 2019 (doi: https://doi.org/10.1101/2019.12.30.891085).

754-OA Ansar M, Ebstein F, Özkoç H, Paracha SA, Iwaszkiewicz J, Gesemann M, Zoete V, Ranza E, Santoni FA, Sarwar MT, Ahmed J, Krüger E, Bachmann-Gagescu R, **Antonarakis SE.** Biallelic variants in PSMB1 encoding the proteasome subunit β6 cause impairment of proteasome function, microcephaly, intellectual disability, developmental delay and short stature. **Human Molecular Genetics** 29: 1132–1143, 2020 (doi: 10.1093/hmg/ddaa032) [Epub Mar 4, 2020] [PMID: 32129449] (abstract).

755-OA Raftopoulou C, Roumelioti FM, Dragona E, Gimelli S, Sloan-Béna F, Gorgoulis V, **Antonarakis SE**, Gagos S. Karyotypic Flexibility of the Complex Cancer Genome and the Role of Polyploidization in Maintenance of Structural Integrity of Cancer Chromosomes. **Cancers** 2020 pii: E591 (doi: 10.3390/cancers12030591) [Epub Mar 5, 2020] [PMID: 32150835] (abstract).

756-OL **Antonarakis SE.** Taurine newborn screening to prevent one form of retinal degeneration. European Journal of Human Genetics 28: 1479-1480, 2020 (doi: 10.1038/s41431-020-0671-3) [Epub Jun 22, 2020] [PMID: 32572200] (abstract).

757-E **Antonarakis SE.** 2019 William Allan Award: Know Thy Genome. **American Journal of Human Genetics** 106, 289-294, 2020 [Epub Mar 6, 2020] (doi.org/10.1016/j.ajhg.2020.02.005).
See 2019 Allan Award Introduction: Stylianos Antonarakis by Haig H. Kazazian Jr. American Journal of Human Genetics 106: 288, 2020 (doi.org/10.1016/j.ajhg.2020.01.006)

758-OA Polla DL, Farazi Fard MA, Tabatabaei Z, Habibzadeh P, Nikuei P, Makrythanasis P, Schuurs-Hoeijmakers JHM, Shahzad M, Fatima F, Fatima N, Donker Kaat L, Bruggenwirth HT, Fleming LR, Condie J, Ploski R, Pollak A, Pilch J, Venselaar H, Masri AT, Hamamy H, Santoni FA, Linda K, Ahmed ZM, Nadif Kasri N, de Brouwer APM, Ansar M, Riazuddin S, Riazuddin S, Silawi M, Ruggeri G, Pirozzi F, Eftekhar E, Taghipour Sheshdeh A, Bahramjahan S, Mirzaa GM, **Antonarakis SE**, Ali Faghihi M, van Bokhoven H. Biallelic variants in TMEM222, encoding a neuronal vesicular transmembrane protein, cause a new autosomal recessive neurodevelopmental disorder. **Genetics in Medicine** 23: 1246-1254, 2021 (doi: 10.1038/s41436-021-01133-w) [Epub Apr 6, 2021] [PMID: 33824500] (abstract).

759-R Illouz T, Biragyn A, Frenkel-Morgenstern M, Weissberg O, Gorohovski A, Merzon E, Green I, Iulita F, Flores-Aguilar L, Del Mar Dierssen Sotos M, De Toma I, Lifshitz H, **Antonarakis SE**, Yu E, Herault Y, Potier MC, Botté A, Roper R, Sredni B, Sarid R, London J, Mobley W, Strydom A, Okun E. Specific Susceptibility to COVID-19 in Adults with Down Syndrome. **Neuromolecular Medicine** 23: 561-571, 2021 (doi: 10.1007/s12017-021-08651-5) [Epub Mar 4, 2021] [PMID: 33660221] (abstract).

760-OA Tremblay-Laganière C, Mai Nguyen TT, Maroofian R, Ghayoor Karimiani EG, Kirmani S, Akbar F, Ibrahim S, Afroze B, Doosti M, Ashrafzadeh F, Babaei M, Efthymiou S, Sultan T, Ladda RL, McLaughlin HM, Truty R, Mahida S, Cohen JS, Baranano K, Ismail FY, Patel MS, Lehman A, Edmondson AC, Nagy A, Walker MA, Mercimek-Andrews S, Maki Y, Sachdev R, Macintosh R, Palmer EE, Mancini GMS, Barakat TS, Steinfeld R, Rüsch C, Stettner G, Wagner M, Wortmann SB, Kini U, Brady AF, Stals KL, Ismayilova N, Ellard S, Bernardo D, Nugent K, McLean SD, **Antonarakis SE**, Houlden H, Kinoshita T, Campeau PM, Murakami Y. PIGG variant pathogenicity assessment reveals novel features within nineteen families. **Genetics in Medicine** 23: 1873-1881, 2021 (doi: 10.1038/s41436-021-01215-9) [Epub Jun 10, 2021] [PMID: 34113002] (abstract).

761-OA Itai T, Hamanaka K, Sasaki K, Wagner M, Kotzaeridou U, Bröesse I, Ries M, Kobayashi Y, Tohyama J, Kato M, Peitee Ong W, Chew HB, Rethanavelu K, Ranza E, Blanc X, Uchiyama Y, Tsuchida N, Fujita A, Azuma Y, Koshimizu E, Mizuguchi T, Takata A, Miyake N, Takahashi H, Miyagi E, Tsurusaki Y, Doi H, Taguri M, **Antonarakis SE**, Nakashima M, Saitsu H, Miyatake S, Matsumoto N. De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. **Human Mutation** 42: 66-76, 2021 (doi: 10.1002/humu.24130) [Epub Nov 1, 2020] [PMID: 33131106] (abstract)

762-OA *Ansar M, Pituello F, Paracha SA, Bel Vialar S, Ranza E, Santoni FA, Sarwar MT, Ahmed J, Agius E, **Antonarakis SE**. CDC25B biallelic variants cause short stature, microcephaly, intellectual disability, developmental delay, facial dysmorphism and microphthalmia. Submitted 2021.*

763-OA **Antonarakis SE**, Ales H, Rapti M, Rademaker J, Meylan J, Iwaszkiewicz J, Zoete V, Ansar M, Borel C, Menzel O, Kuželová K, Santoni FA. Dominant monoallelic variant in the PAK2 gene causes Knobloch syndrome type 2. **Human Molecular Genetics** 31: 1-9, 2021 (doi: 10.1093/hmg/ddab026) [Epub Mar 9, 2021] [PMID: 33693784] (abstract). Biorxiv posted October 7, 2020. https://doi.org/10.1101/2020.10.06.328419.

764-OA Chen N, Zhao S, Jolly A, Wang L, Pan H, Yuan J, Chen S, Koch A, Ma C, Tian W, Jia Z, Kang J, Zhao L, Qin C, Fan X, Rall K, Coban-Akdemir Z, Chen Z, Jhangiani S, Liang Z, Niu Y, Li X, Yan Z, Wu Y, Dong S, Song C, Qiu G, Zhang S, Liu P, Posey JE, Zhang F, Luo G, Wu Z on behalf of Deciphering Disorders Involving Scoliosis and COmorbidities (DISCO) study group, Su J, Zhang J, Chen E, Rouskas K, Glentis S, Bacopoulou F, Deligeoroglou , Chrousos G, Lyonnet S, Rosenberg CM, Dingeldein I, Bonilla X, Borel C, Gibbs RA, Dietrich JE, Dimas AS, **Antonarakis SE**, Brucker S, Lupski JR, Wu N, Zhu L. Perturbations of genes essential for Müllerian duct/Wölffian duct development in Mayer-Rokitansky-Küster-Hauser syndrome. **American Journal of Human Genetics** 108: 337-345, 2021 (doi: 10.1016/j.ajhg.2020.12.014) [Epub Jan 5, 2021] [PMID: 33434492] (abstract).

765-OA Schalk A, Cousin MA, Dsouza NR, Challman TD, Wain KE, Powis Z, Minks K, Trimouille A, Lasseaux E, Lacombe D, Angelini C, Michaud V, Van-Gils J, Spataro N, Ruiz A, Gabau E, Stolerman E, Washington C, Louie R, Lanpher BC, Kemppainen JL, Innes M, Kooy F, Meuwissen M, Goldenberg A, Lecoquierre F, Vera G, Diderich KEM, Sheidley B, El Achkar CM, Park M, Hamdan FF, Michaud JL, Lewis AJ, Zweier C, Reis A, Wagner M, Weigand H, Journel H, Keren B, Passemard S, Mignot C, van Gassen K, Brilstra EH, Itzikowitz G, O'Heir E, Allen J, Donald KA, Korf BR, Skelton T, Thompson M, Robin NH, Rudy NL, Dobyns WB, Foss K, Zarate YA, Bosanko KA, Alembik Y, Durand B, Tran Mau-Them F, Ranza E, Blanc X, **Antonarakis SE**, McWalter K, Torti E, Millan F, Dameron A, Tokita M, Zimmermann MT, Klee EW, Piton A, Gerard B. De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. **Journal of Medical Genetics** 59: 965-975, 2022 (doi: 10.1136/jmedgenet-2021-107751) [Epub Dec 15, 2021] [PMID: 34930816] (abstract).

766-R **Antonarakis SE**. Short arms of human acrocentric chromosomes and the completion of the human genome sequence. **Genome Research** 32: 599-607, 2022 (doi: 10.1101/gr.275350.121) [Epub Mar 31, 2022] [PMID: 35361624] (abstract).

767-R **Antonarakis SE**. History of the methodology of disease gene identification. **American Journal of Medical Genetics A** 185: 3266-3275, 2021 (doi: 10.1002/ajmg.a.62400) [Epub Jun 23, 2021] [PMID: 34159713] (abstract).

768-OA Rodan LH, Spillmann RC, Kurata HT, Lamothe SM, Maghera J, Jamra RA, Alkelai A, **Antonarakis SE**, Atallah I, Bar-Yosef O, Bilan F, Bjorgo K, Blanc X, Van Bogaert P, Bolkier Y, Burrage LC, Christ BU, Granadillo JL, Dickson P, Donald KA, Dubourg C, Eliyahu A, Emrick L, Engleman K, Veronika Gonfiantini MV, Good JM, Kalser J, Kloeckner C, Lachmeijer G, Macchiaiolo M, Nicita F, Odent S, O'Heir E, Ortiz-Gonzalez X, Pacio-Miguez M, Palomares-Bralo M, Pena L, Platzer K, Quinodoz M, Ranza E, Rosenfeld JA, Elane Roulet-Perez E, Santani A, Santos Simarro F, Pode-Shakked B, Skraban C, Slaugh R, Superti-Furga A, Thiffault I, van Jaabrsveld RH, Vincent M, Wang HG, Zacher P, Undiagnosed Diseases Network*, Rush E, Pitt G, Yee Billie Au PY, Shashi V. Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. **Genetics in Medicine** 23: 1922-1932, 2021 (doi: 10.1038/s41436-021-01232-8) [Epub Jun 23, 2021] [PMID: 34163037] (abstract).

769-OA Nurk S, Koren S, Rhie A, Rautiainen M, Bzikadze AV, Mikheenko A, Vollger MR, Altemose N, Uralsky L, Gershman A, Aganezov S, Hoyt SJ, Diekhans M, Logsdon GA, Alonge M, **Antonarakis SE**, Borchers M, Bouffard GG, Brooks SY, Caldas GV, Chen NC, Cheng H, Chin CS, Chow W, de Lima LG, Dishuck PC, Durbin R, Dvorkina T, Fiddes IT, Formenti G, Fulton RS, Fungtammasan A, Garrison E, Grady PGS, Graves-Lindsay TA, Hall IM, Hansen NF, Hartley GA, Haukness M, Howe K, Hunkapiller MW, Jain C, Jain M, Jarvis ED, Kerpedjiev P, Kirsche M, Kolmogorov M, Korlach J, Kremitzki M, Li H, Maduro VV, Marschall T, McCartney AM, McDaniel J, Miller DE, Mullikin JC, Myers EW, Olson ND, Paten B, Peluso P, Pevzner PA, Porubsky D, Potapova T, Rogaev EI, Rosenfeld JA, Salzberg SL, Schneider VA, Sedlazeck FJ, Shafin K, Shew CJ, Shumate A, Sims Y, Smit AFA, Soto DC, Sović I, Storer JM, Streets A, Sullivan BA, Thibaud-Nissen F, Torrance J, Wagner J, Walenz BP, Wenger A, Wood JMD, Xiao C, Yan SM, Young AC, Zarate S, Surti U, McCoy RC, Dennis MY, Alexandrov IA, Gerton JL, O'Neill RJ, Timp W, Zook JM, Schatz MC, Eichler EE, Miga KH, Phillippy AM. The complete sequence of a human genome.

Science 376: 44-53, 2022. (doi: 10.1126/science.abj6987) [Epub Mar 31, 2022] [PMID: 35357919] (abstract).
Biorxiv posted May 27, 2021, (doi.org/10.1101/2021.05.26.445798).

770-OA Mattioli F, Darvish H, Aziz Paracha S, Tafakhori A, Ghasemi Firouzabadi S, Chapi M, Azhar Baig HM, Reymond A, **Antonarakis SE**, Ansar M. Biallelic truncation variants in ATP9A are associated with a novel autosomal recessive neurodevelopmental disorder. **NPJ Genomic Medicine** 6: 94, 2021 (doi.org/10.1038/s41525-021-00255-z) [Epub Nov 11, 2021] [PMID: 34764295] (abstract).
Medrxiv posted Jun 4, 2021 (doi.org/10.1101/2021.05.31.21257832)

771-R Illouz T, Biragyn A, Iulita MF, Flores-Aguilar L, Dierssen M, De Toma I, **Antonarakis SE**, Yu E, Herault Y, Potier MC, Botté A, Roper R, Sredni B, London J, Mobley W, Strydom A, Okun E. Immune dysregulation and increased risk of complications and mortality following respiratory tract infections in adults with Down syndrome. **Frontiers in Immunology-Inflammation** 12: 621440, 2021 (DOI: 10.3389/fimmu.2021.621440) [Epub Jun 25, 2021] [PMID: 34248930].

772-R Lee C, **Antonarakis SE**, Hamosh A, Burn J. Three Decades of the Human Genome Organisation (HUGO). **American Journal of Medical Genetics A** 185: 3314-3321, 2021 (doi: 10.1002/ajmg.a.62512) [Epub Sep 28, 2021] [PMID: 34581472] (abstract).

773-OA Rapti M, Zouaghi Y, Meylan J, Ranza E, **Antonarakis SE**, Santoni FA. CoverageMaster: comprehensive CNV detection and visualization from NGS short reads for genetic medicine applications. **Briefings in Bioinformatics** 23: bbac049 2022 (doi: 10.1093/bib/bbac049). [Epub Feb 28, 2022] [PMID: 35224620] (abstract).

774-OA Meuwissen M, Verstraeten A, Ranza E, Iwaszkiewicz J, Bastiaansen M, Mateiu L, Nemegeer M, Meester J, Afenjar A, Amaral M, Ballhausen D, Barnett SS, Barth M, Asselbergh B, Spaas K, Heeman B, Bassetti J, Blackburn PR, Schaer M, Blanc X, Zoete V, Casas K, Courtin T, Doummar D, Guerry F, Keren B, Pappas J, Rabin R, Shinawi M, Vulto-van Silfhout AT, Wagner M, Ziegler A, Schaefer E,Gerard B, De Bie CI, Abbot MA, **Antonarakis SE***, Loeys B*. Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, cause a neurodevelopmental disorder. **Genetics in Medicine** 24: 1583-1591, 2022 (doi: 10.1016/j.gim.2022.04.003) [Epub May 2, 2022] [PMID: 35499524] (abstract).

775-OA Jolly A , Du H, Borel C, Chen N , Zhao S, Grochowski CM, Fatih JM, Dawood M, Jhangiani SN, Muzny DN, Koch A, Rouskas K, Glentis S, Bacopoulou F, Wise CA, Dietrich JE, Van den Veyver IB, Dimas AS, Brucker S, Sutton VR, Gibbs RA, **Antonarakis SE**, Wu N, Coban-Akdemir ZH, Zhu L, Posey JE, Lupski JR. Mutational Burden Analysis Supports GREB1L as a Contributory Driver Gene in the Etiology of Mayer-Rokitansky-Küster-Hauser Syndrome. **Human Genetics and Genomics (HGG) Advances** in press 2023.

776-OA Mao K#, Borel C#, Ansar M#, Jolly A, Makrythanasis P, Froehlich C, Iwaszkiewicz J, Wang B, Xu X, Li Q, Blanc X, Zhu H, Chen Q, Jin F, Ankamreddy H, Singh S, Zhang H, Wang X, Chen P, Ranza E, Paracha SA, Shah SF, Guida V, Piceci-Sparascio F, Melis D, Dallapiccola B, Digilio MC, Novelli A, Magliozzi M, Fadda MT, Streff H, Machol K, Lewis RA, Zoete V, Squeo GM, Prontera P, Mancano G, Gori G, Mariani M, Selicorni A, Psoni S, Fryssira H, Douzgou S, Marlin S, Biskup S, De Luca A, Merla G, Zhao S, Cox TC, Groves AK, Lupski JR, Zhang Q, Zhang YB*, **Antonarakis SE***. FOXI3 pathogenic variants cause one form of craniofacial macrosomia. **Nature Communications** 2023 Apr 11;14:2026, 2023 (doi: 10.1038/s41467-023-37703-6) [Epub Apr 11, 2023] [PMID: 37041148] (abstract).

777-E **Antonarakis SE**, Orkin SH. Haig H. Kazazian Jr., M.D. (1937-2022) Obituary. **Genome Research** 32: v-vi, 2022 (doi: 10.1101/gr.276712.122) [Epub Mar 21, 2022].

778-OA Ma C, Chen N, Jolly A, Zhao S, Coban-Akdemir Z, Tian W, Kang J, Ye Y, Wang Y, Koch A, Zhang Y, Qin C, Bonilla X, Borel C, Rall K, Chen Z, Jhangiani S, Niu Y, Li X, Qiu G, Zhang S, Luo G, Wu Z, Deciphering Disorders Involving Scoliosis and COmorbidities (DISCO) study group, Bacopoulou F, Deligeoroglou E, Zhang TJ, Rosenberg C, Gibbs RA, Dietrich JE, Dimas AS, Liu P, **Antonarakis SE**, Brucker SY, Posey JE, Lupski JR, Wu N, Zhu L. Functional characteristics of a broad spectrum of TBX6 variants in Mayer-Rokitansky-Küster-Hauser syndrome. **Genetics in Medicine** 24: 2262-2273, 2022 (doi: 10.1016/j.gim.2022.08.012) [Epub Sep 16, 2022] [PMID: 36112137] (abstract).

779-OA Cediel ML, Stawarski M, Blanc X, Nosková L, Magner M, Platzer K, Gburek-Augustat J, Baldridge D, Constantino JN, Ranza E, Bettler B, **Antonarakis SE**. GABBR1 Monoallelic De Novo Variants Linked to Neurodevelopmental Delay and Epilepsy. **American Journal of Human Genetics** 109: 1885-1893, 2022 (doi: 10.1016/j.ajhg.2022.08.010) [Epub Sep 13, 2022] [PMID: 36103875] (abstract).

780-OA Hiatt SM, Trajkova S, Rossi Sebastiano M, Partridge EC, Abidi FE, Anderson A, Ansar M, **Antonarakis SE**, Azadi A, Bachmann-Gagescu R, Bartuli A, Benech C, Berkowitz JL, Betti MJ, Brusco A, Cannon A, Caron G, Chen Y, Crenshaw MM, Cuisset L, Curry CJ, Darvish H, Demirdas S, Descartes M, Douglas J, Dyment DA, Zghal Elloumi H, Ermondi G, Faoucher M,

Farrow EG, Felker SR, Fisher H, Hurst ACE, Joset P, Kmoch S, Leadem BR, Macchiaiolo M, Magner M, Mandrile G, Mattioli F, McEown M, Meadows SK, Medne L, Meeks NJL, Montgomery S, Napier MP, Natowicz M, Newberry KM, Niceta M, Noskova L, Nowak C, Noyes AG, Osmond M, Pullano V, Quélin C, Rahimi-Aliabadi S, Rauch A, Redon S, Reymond A, Schwager CR, Sellars EA, Scheuerle A, Shukarova-Angelovska E, Skraban C, Sullivan B, Tartaglia M, Thiffault I, Uguen K, Umaña LA, van Bever Y, van der Crabben SN, van Slegtenhorst MA, Waisfisz Q, Myers RM, Cooper GM. Deleterious, protein-altering variants 1 in the X-linked transcriptional coregulatory ZMYM3 in 22 individuals with a neurodevelopmental delay phenotype. **American Journal of Human Genetics** 110: 215-227, 2023 (doi: 10.1016/j.ajhg.2022.12.007) [Epub Dec 21, 2022] [PMID: 36586412] (abstract).

781-OA Mattioli F, Worpenberg L, Li CT, Ibrahim N, Naz S, Sharif S, Ghasemi Firouzabadi S, Vosoogh S, Guex N, **Antonarakis SE**, Ansar M, Darvish H, Liu RJ, Roignant JY, Reymond A. Biallelic variants in NSUN6 cause an autosomal recessive neurodevelopmental disorder. **Genetics in Medicine** in press 2023.

782-OA Serey-Gaut M, Cortes M, Makrythanasis P, Suri M, Taylor AMR, Sullivan JA, Asleh AN, Mitra J, Dar MA, McNamara A, Shashi V, Dugan S, Song X, Rosenfeld JA, Cabrol C, Iwaszkiewicz J, Zoete V, Pehlivan D, Coban Akdemir Z, Roeder ER, Okashah Littlejohn R, Dibra HK, Byrd PJ, Stewart GS, Geckinli BB, Posey J, Westman R, Jungbluth C, Eason J, Sachdev R, Evans CA, Lemire G, VanNoy GE, O'Donnell-Luria A, Tran Mau-Them F, Juven A, Piard J, Yee Nixon C, Zhu Y, Ha T, Buckley MF, Thauvin C, Umanah GKE, Van Maldergem L, Lupski JR, Roscioli T , Dawson VL, Dawson TM, **Antonarakis SE.** Biallelic TTI1 variants cause an autosomal recessive neurodevelopmental disorder with microcephaly. **American Journal of Human Genetics** 110: 499-515, 2023 (doi: 10.1016/j.ajhg.2023.01.006). [Epub Jan 31, 2023] [PMID: 36724785] (abstract).

783-OA Rao AR, Nazir A, Imtiaz S, Paracha SA, Waryah YM, Ujjan ID, Anwar I, Iqbal A, Santoni FA, Shah I, Gul K, Baig HMA, Waryah AM, **Antonarakis SE,** Ansar M. Delineating the Spectrum of Genetic Variants Associated with Bardet-Biedl Syndrome in Consanguineous Pakistani Pedigrees. **Genes** (Basel). 14:404, 2023. (doi: 10.3390/genes14020404) [Epub Feb 3, 2023] [PMID: 36833331](abstract).

OA: original articles	584
BC: book chapters	68
R: reviews	63
N&V: news and views	10
Let: letters	6
OL: original letters with data	8
RFLP: polymorphism reports	12
CR: committee reports	13
E: Editorial	5
BF: Book forward	1
Book: Books	10

Bibliography in italics and gray not yet accepted for publication.

Google scholar

Citation indices 26-apr-2023

	All	*Since 2018*
Citations	129102	36173
h-index	159	75
i10-index	636	319

http://www.ncbi.nlm.nih.gov/pubmed/?term=antonarakis+s

UNIVERSITÉ
DE GENÈVE

FACULTÉ DE MÉDECINE
Department of Genetic
Medicine and Development

Stylianos E. Antonarakis
Professor Emeritus (active)

Direct line: +41 (0)79 691 2340
stylianos.antonarakis@unige.ch

Γενεύη, 26 Απριλίου 2023

Συμβούλιο Διοίκησης ΕΚΠΑ 2023

Αγαπητή/έ κυρία/κύριε
Επιθυμώ ευγενικά να σας εκφράσω την εκδήλωση ενδιαφέροντος για τη θέση εξωτερικού μέλους του συμβουλίου διοίκησης του Εθνικού και Καποδιστριακού Πανεπιστημίου Αθηνών (ΕΚΠΑ) σύμφωνα με την ανακοινωση σας και την ισχύουσα νομοθεσία.
Είμαι υπερήφανος που σπούδασα στην Ιατρική Σχολή του ΕΚΠΑ την περίοδο 1969-1975 και ιδιαίτερα ευγνώμων που έγινα επίτιμος διδάκτορας του ίδιου πανεπιστημίου το 2018. Θα ήθελα λοιπόν να χρησιμοποιήσω την εκτεταμένη και πολυετή ακαδημαϊκή μου εμπειρία (σε πανεπιστήμια της Ευρώπης και της Αμερικής) στη θέση του μέλους του συμβουλίου διοίκησης, για την καλή λειτουργία του ΕΚΠΑ και την διεθνή του ακτινοβολία.

Μετά τιμής

S.E. Antonarakis

Στυλιανός Ε. Αντωναράκης