Stanislas Lyonnet

List of Publications by Year
in descending order

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281 papers	27,663 citations	4641 85 h-index	6979 154 g-index
285 all docs	285 docs citations	285 times ranked	28050 citing authors

#	Article	IF	Citations
1	Biallelic alterations in PLXND1 cause common arterial trunk and other cardiac malformations in humans. Human Molecular Genetics, 2023, 32, 353-356.	1.4	3
2	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. Journal of Medical Genetics, 2022, 59, 559-567.	1.5	25
3	Using deep-neural-network-driven facial recognition to identify distinct Kabuki syndrome 1 and 2 gestalt. European Journal of Human Genetics, 2022, 30, 682-686.	1.4	4
4	Agonists of prostaglandin E ₂ receptors as potential first in class treatment for nephronophthisis and related ciliopathies. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2115960119.	3.3	13
5	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	3.9	24
6	Bi-allelic variants in IPO8 cause a connective tissue disorder associated with cardiovascular defects, skeletal abnormalities, and immune dysregulation. American Journal of Human Genetics, 2021, 108, 1126-1137.	2.6	14
7	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	2.6	17
8	Size matters: Large copy number losses in Hirschsprung disease patients reveal genes involved in enteric nervous system development. PLoS Genetics, 2021, 17, e1009698.	1.5	14
9	Prenatalâ€onset of congenital neuronal ceroid lipofuscinosis with a novel CTSD mutation. Birth Defects Research, 2021, 113, 1324-1332.	0.8	1
10	TALPID3/KIAA0586 Regulates Multiple Aspects of Neuromuscular Patterning During Gastrointestinal Development in Animal Models and Human. Frontiers in Molecular Neuroscience, 2021, 14, 757646.	1.4	3
11	Expanding the phenotype in Adamsâ€“Oliver syndrome correlating with the genotype. American Journal of Medical Genetics, Part A, 2020, 182, 29-37.	0.7	15
12	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. Genetics in Medicine, 2020, 22, 181-188.	1.1	30
13	Three Copies of Four Interferon Receptor Genes Underlie a Mild Type I Interferonopathy in Down Syndrome. Journal of Clinical Immunology, 2020, 40, 807-819.	2.0	44
14	Should autism spectrum disorder be considered part of CHARGE syndrome? A cross-sectional study of 46 patients. Orphanet Journal of Rare Diseases, 2020, 15, 136.	1.2	4
15	Bi-allelic Variations of SMO in Humans Cause a Broad Spectrum of Developmental Anomalies Due to Abnormal Hedgehog Signaling. American Journal of Human Genetics, 2020, 106, 779-792.	2.6	25
16	Phenotypic similarity for rare disease: Ciliopathy diagnoses and subtyping. Journal of Biomedical Informatics, 2019, 100, 103308.	2.5	17
17	PAICS deficiency, a new defect of de novo purine synthesis resulting in multiple congenital anomalies and fatal outcome. Human Molecular Genetics, 2019, 28, 3805-3814.	1.4	22
18	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30

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19	Anatomical and functional abnormalities on MRI in kabuki syndrome. NeuroImage: Clinical, 2019, 21, 101610.	1.4	17
20	MED13L lossâ€ofâ€function variants in two patients with syndromic Pierre Robin sequence. American Journal of Medical Genetics, Part A, 2018, 176, 181-186.	0.7	9
21	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscyphodysplasia. European Journal of Human Genetics, 2018, 26, 1611-1622.	1.4	18
22	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain, 2018, 141, 1998-2013.	3.7	67
23	Targeted therapy in patients with PIK3CA-related overgrowth syndrome. Nature, 2018, 558, 540-546.	13.7	374
24	Mutations in BOREALIN cause thyroid dysgenesis. Human Molecular Genetics, 2017, 26, ddw419.	1.4	37
25	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255.	9.4	88
26	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	2.6	61
27	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	3.8	72
28	Moyamoya syndrome in children with neurofibromatosis type 1: Italianâ€“French experience. American Journal of Medical Genetics, Part A, 2017, 173, 1521-1530.	0.7	36
29	De novo mutations in CBL causing early-onset paediatric moyamoya angiopathy. Journal of Medical Genetics, 2017, 54, 550-557.	1.5	33
30	Efficiency of prenatal diagnosis in Pierre Robin sequence. Prenatal Diagnosis, 2017, 37, 1169-1175.	1.1	18
31	Mutations in TUBB4B Cause a Distinctive Sensorineural Disease. American Journal of Human Genetics, 2017, 101, 1006-1012.	2.6	30
32	Phenotype and genotype analysis of a French cohort of 119 patients with CHARGE syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 417-430.	0.7	65
33	Altered SOX9 genital tubercle enhancer region in hypospadias. Journal of Steroid Biochemistry and Molecular Biology, 2017, 170, 28-38.	1.2	10
34	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	1.1	134
35	DCDC2Mutations Cause Neonatal Sclerosing Cholangitis. Human Mutation, 2016, 37, 1025-1029.	1.1	56
36	Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. American Journal of Human Genetics, 2016, 98, 971-980.	2.6	113

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37	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. Human Molecular Genetics, 2016, 25, ddw333.	1.4	38
38	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	2.6	39
39	Mutations in RIT1 cause Noonan syndrome with possible juvenile myelomonocytic leukemia but are not involved in acute lymphoblastic leukemia. European Journal of Human Genetics, 2016, 24, 1124-1131.	1.4	23
40	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	2.6	132
41	Mosaic parental germline mutations causing recurrent forms of malformations of cortical development. European Journal of Human Genetics, 2016, 24, 611-614.	1.4	33
42	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	1.1	125
43	RPL10 mutation segregating in a family with Xâ€linked syndromic Intellectual Disability. American Journal of Medical Genetics, Part A, 2015, 167, 1908-1912.	0.7	27
44	Prenatal diagnosis of Pierre Robin Sequence: accuracy and ability to predict phenotype and functional severity. Prenatal Diagnosis, 2015, 35, 853-858.	1.1	19
45	Functional Assessment of Disease-Associated Regulatory Variants In Vivo Using a Versatile Dual Colour Transgenesis Strategy in Zebrafish. PLoS Genetics, 2015, 11, e1005193.	1.5	31
46	Novel variants in GNAI3 associated with auriculocondylar syndrome strengthen a common dominant negative effect. European Journal of Human Genetics, 2015, 23, 481-485.	1.4	21
47	Functional Loss of Semaphorin 3C and/or Semaphorin 3D and Their Epistatic Interaction with Ret Are Critical to Hirschsprung Disease Liability. American Journal of Human Genetics, 2015, 96, 581-596.	2.6	118
48	Mutations in KIAA0586 Cause Lethal Ciliopathies Ranging from a Hydrolethalus Phenotype to Short-Rib Polydactyly Syndrome. American Journal of Human Genetics, 2015, 97, 311-318.	2.6	82
49	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531.	2.6	47
50	MMP21 is mutated in human heterotaxy and is required for normal left-right asymmetry in vertebrates. Nature Genetics, 2015, 47, 1260-1263.	9.4	65
51	Mutations of the ImprintedCDKN1CGene as a Cause of the Overgrowth Beckwith-Wiedemann Syndrome: Clinical Spectrum and Functional Characterization. Human Mutation, 2015, 36, 894-902.	1.1	62
52	Novel Mutation and Structural RNA Analysis of the Noncoding RNase <i>MRP</i> Gene in Cartilage-Hair Hypoplasia. Molecular Syndromology, 2015, 6, 77-82.	0.3	6
53	Incomplete penetrance and phenotypic variability of 6q16 deletions including SIM1. European Journal of Human Genetics, 2015, 23, 1010-1018.	1.4	35
54	Identification of a novel ARL13B variant in a Joubert syndrome-affected patient with retinal impairment and obesity. European Journal of Human Genetics, 2015, 23, 621-627.	1.4	48

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55	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 2015, 125, 3585-3599.	3.9	69
56	Extensive investigation of the IGF2/H19 imprinting control region reveals novel OCT4/SOX2 binding site defects associated with specific methylation patterns in Beckwith-Wiedemann syndrome. Human Molecular Genetics, 2014, 23, 5763-5773.	1.4	58
57	Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. Human Molecular Genetics, 2014, 23, 2279-2289.	1.4	98
58	Multiple congenital anomaliesâ€intellectual disability (MCAâ€ID) and neuroblastoma in a patient harboring a de novo 14q23.1q23.3 deletion. American Journal of Medical Genetics, Part A, 2014, 164, 1310-1317.	0.7	9
59	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	1.1	50
60	Clinical evidence for a mandibular to maxillary transformation in Auriculocondylar syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1850-1853.	0.7	6
61	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	1.1	113
62	3q27.3 microdeletional syndrome: a recognisable clinical entity associating dysmorphic features, marfanoid habitus, intellectual disability and psychosis with mood disorder. Journal of Medical Genetics, 2014, 51, 21-27.	1.5	12
63	Genetic characterization of congenital tufting enteropathy: epcam associated phenotype and involvement of SPINT2 in the syndromic form. Human Genetics, 2014, 133, 299-310.	1.8	83
64	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	1.8	71
65	Enhancer mutations and phenotype modularity. Nature Genetics, 2014, 46, 3-4.	9.4	24
66	Identification of Novel Craniofacial Regulatory Domains Located far Upstream ofSOX9and Disrupted in Pierre Robin Sequence. Human Mutation, 2014, 35, 1011-1020.	1.1	69
67	CNS involvement in OFD1 syndrome: a clinical, molecular, and neuroimaging study. Orphanet Journal of Rare Diseases, 2014, 9, 74.	1.2	34
68	Contribution of rare and common variants determine complex diseasesâ€”Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	0.9	119
69	Pathways systematically associated to Hirschsprungâ€™s disease. Orphanet Journal of Rare Diseases, 2013, 8, 187.	1.2	17
70	Further characterization of atypical features in auriculocondylar syndrome caused by recessive PLCB4 mutations. American Journal of Medical Genetics, Part A, 2013, 161, 2339-2346.	0.7	22
71	Developmental outcome in Pierre Robin sequence: A longitudinal and prospective study of a consecutive series of severe phenotypes. American Journal of Medical Genetics, Part A, 2013, 161, 312-319.	0.7	34
72	Mutations in Endothelin 1 Cause Recessive Auriculocondylar Syndrome and Dominant Isolated Question-Mark Ears. American Journal of Human Genetics, 2013, 93, 1118-1125.	2.6	59

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73	Clinical and Molecular Spectrum of Renal Malformations in Kabuki Syndrome. Journal of Pediatrics, 2013, 163, 742-746.	0.9	27
74	ALDH1A3 Mutations Cause Recessive Anophthalmia and Microphthalmia. American Journal of Human Genetics, 2013, 92, 265-270.	2.6	92
75	Understanding the basis of auriculocondylar syndrome: Insights from human, mouse and zebrafish genetic studies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 306-317.	0.7	48
76	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	1.4	190
77	Finger creases lend a hand in Kabuki syndrome. European Journal of Medical Genetics, 2013, 56, 556-560.	0.7	11
78	Sporadic NF1 mutation associated with a de-novo 20q11.3 deletion explains the association of unusual facies, Moyamoya vasculopathy, and developmental delay, reported by Bertoli et al. in 2009. Clinical Dysmorphology, 2013, 22, 42-43.	0.1	8
79	ZEB2 zinc-finger missense mutations lead to hypomorphic alleles and a mild Mowatâ€“Wilson syndrome. Human Molecular Genetics, 2013, 22, 2652-2661.	1.4	51
80	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. Journal of Medical Genetics, 2013, 50, 174-186.	1.5	44
81	Congenital Heart Defects in Patients with Deletions Upstream ofSOX9. Human Mutation, 2013, 34, 1628-1631.	1.1	33
82	Genetic Variations Creating MicroRNA Target Sites in the FXN 3â€²-UTR Affect Frataxin Expression in Friedreich Ataxia. PLoS ONE, 2013, 8, e54791.	1.1	24
83	Chromosome 21 Scan in Down Syndrome Reveals DSCAM as a Predisposing Locus in Hirschsprung Disease. PLoS ONE, 2013, 8, e62519.	1.1	22
84	Nasal speech in patients with 12q15 microdeletions. European Journal of Human Genetics, 2012, 20, 367-367.	1.4	0
85	Male and female differential reproductive rate could explain parental transmission asymmetry of mutation origin in Hirschsprung disease. European Journal of Human Genetics, 2012, 20, 917-920.	1.4	8
86	Antenatal spectrum of CHARGE syndrome in 40 fetuses withCHD7mutations. Journal of Medical Genetics, 2012, 49, 698-707.	1.5	45
87	Mutation in a primate-conserved retrotransposon reveals a noncoding RNA as a mediator of infantile encephalopathy. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 4980-4985.	3.3	58
88	EFTUD2haploinsufficiency leads to syndromic oesophageal atresia. Journal of Medical Genetics, 2012, 49, 737-746.	1.5	89
89	Mutations of TSEN and CASK genes are prevalent in pontocerebellar hypoplasias type 2 and 4. Brain, 2012, 135, e199-e199.	3.7	18
90	CLMP Is Required for Intestinal Development, and Loss-of-Function Mutations Cause Congenital Short-Bowel Syndrome. Gastroenterology, 2012, 142, 453-462.e3.	0.6	49

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91	TCTN3 Mutations Cause Mohr-Majewski Syndrome. American Journal of Human Genetics, 2012, 91, 372-378.	2.6	123
92	A Human Homeotic Transformation Resulting from Mutations in PLCB4 and GNAI3 Causes Auriculocondylar Syndrome. American Journal of Human Genetics, 2012, 91, 397.	2.6	1
93	ISL1 Directly Regulates FGF10 Transcription during Human Cardiac Outflow Formation. PLoS ONE, 2012, 7, e30677.	1.1	46
94	Exome Sequencing Identifies PDE4D Mutations as Another Cause of Acrodysostosis. American Journal of Human Genetics, 2012, 90, 740-745.	2.6	115
95	A Human Homeotic Transformation Resulting from Mutations in PLCB4 and GNAI3 Causes Auriculocondylar Syndrome. American Journal of Human Genetics, 2012, 90, 907-914.	2.6	75
96	Cis-Regulatory Disruption at the SOX9 Locus as a Cause of Pierre Robin Sequence. , 2012, , 123-136.		0
97	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. Journal of Medical Genetics, 2011, 48, 417-421.	1.5	45
98	A founder effect at the EPCAM locus in Congenital Tufting Enteropathy in the Arabic Gulf. European Journal of Medical Genetics, 2011, 54, 319-322.	0.7	37
99	Nuclear Outsourcing of RNA Interference Components to Human Mitochondria. PLoS ONE, 2011, 6, e20746.	1.1	249
100	Genetic Factors in Isolated and Syndromic Esophageal Atresia. Journal of Pediatric Gastroenterology and Nutrition, 2011, 52, S6-8.	0.9	22
101	KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	9.4	203
102	Dissection of the MYCN locus in Feingold syndrome and isolated oesophageal atresia. European Journal of Human Genetics, 2011, 19, 602-606.	1.4	24
103	Nasal speech and hypothyroidism are common hallmarks of 12q15 microdeletions. European Journal of Human Genetics, 2011, 19, 1032-1037.	1.4	11
104	Mortality Associated with Neurofibromatosis 1: A Cohort Study of 1895 Patients in 1980-2006 in France. Orphanet Journal of Rare Diseases, 2011, 6, 18.	1.2	96
105	Polynesian ecology determines seasonality of biliary atresia. Hepatology, 2011, 54, 1893-1894.	3.6	12
106	Only four genes (EDA1, EDAR, EDARADD, and WNT10A) account for 90% of hypohidrotic/anhidrotic ectodermal dysplasia cases. Human Mutation, 2011, 32, 70-72.	1.1	240
107	Germline gain-of-function mutations of ALK disrupt central nervous system development. Human Mutation, 2011, 32, 272-276.	1.1	38
108	REEP1 mutations in SPG31: Frequency, mutational spectrum, and potential association with mitochondrial morpho-functional dysfunction. Human Mutation, 2011, 32, 1118-1127.	1.1	83

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109	Germline deletion of the miR-17â^¼92 cluster causes skeletal and growth defects in humans. Nature Genetics, 2011, 43, 1026-1030.	9.4	275
110	Disruption of a long distance regulatory region upstream of SOX9 in isolated disorders of sex development. Journal of Medical Genetics, 2011, 48, 825-830.	1.5	162
111	Differential Contributions of Rare and Common, Coding and Noncoding Ret Mutations to Multifactorial Hirschsprung Disease Liability. American Journal of Human Genetics, 2010, 87, 60-74.	2.6	230
112	Mutation update for theCSB/ERCC6andCSA/ERCC8genes involved in Cockayne syndrome. Human Mutation, 2010, 31, 113-126.	1.1	193
113	High-throughput sequencing of a 4.1â€‰Mb linkage interval reveals FLVCR2 deletions and mutations in lethal cerebral vasculopathy. Human Mutation, 2010, 31, 1134-1141.	1.1	27
114	Paternal uniparental isodisomy of chromosome 6 causing a complex syndrome including complete IFNâ€Î³ receptor 1 deficiency. American Journal of Medical Genetics, Part A, 2010, 152A, 622-629.	0.7	22
115	Additional clinical and molecular analyses of TFAP2A in patients with the branchioâ€oculoâ€facial syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 994-999.	0.7	22
116	Additional clinical and molecular analyses of TFAP2A in patients with the Branchioâ€Oculoâ€Facial syndrome: Previously reported patient. American Journal of Medical Genetics, Part A, 2010, 152A, 2143-2143.	0.7	3
117	Disruption of longâ€distance highly conserved noncoding elements in neurocristopathies. Annals of the New York Academy of Sciences, 2010, 1214, 34-46.	1.8	15
118	microRNAs in diseases: from candidate to modifier genes. Clinical Genetics, 2010, 77, 306-313.	1.0	87
119	Functional Effects of PTPN11 (SHP2) Mutations Causing LEOPARD Syndrome on Epidermal Growth Factor-Induced Phosphoinositide 3-Kinase/AKT/Glycogen Synthase Kinase 3Î² Signaling. Molecular and Cellular Biology, 2010, 30, 2498-2507.	1.1	85
120	Molecular analysis of pericentrin gene (PCNT) in a series of 24 Seckel/microcephalic osteodysplastic primordial dwarfism type II (MOPD II) families. Journal of Medical Genetics, 2010, 47, 797-802.	1.5	100
121	Epistasis between RET and BBS mutations modulates enteric innervation and causes syndromic Hirschsprung disease. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 13921-13926.	3.3	51
122	Comparison of Clinical Presentations and Outcomes Between Patients With TGFBR2 and FBN1 Mutations in Marfan Syndrome and Related Disorders. Circulation, 2009, 120, 2541-2549.	1.6	203
123	Long-range regulation at the SOX9 locus in development and disease. Journal of Medical Genetics, 2009, 46, 649-656.	1.5	148
124	Evaluation of methods for amplification of picogram amounts of total RNA for whole genome expression profiling. BMC Genomics, 2009, 10, 246.	1.2	54
125	In Vitrostudies of non poly alanine PHOX2B mutations argue against a loss-of-function mechanism for congenital central hypoventilation. Human Mutation, 2009, 30, E421-E431.	1.1	34
126	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	1.1	126

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127	Interaction between a chromosome 10RETenhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. Human Mutation, 2009, 30, 771-775.	1.1	57
128	Cholinergic switch associated with morphological differentiation in neuroblastoma. Journal of Pathology, 2009, 219, 463-472.	2.1	26
129	Xq28 duplication presenting with intestinal and bladder dysfunction and a distinctive facial appearance. European Journal of Human Genetics, 2009, 17, 434-443.	1.4	87
130	Genotypeâ€“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, 2009, 17, 454-466.	1.4	240
131	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	9.4	364
132	Sporadic case of unusual facies, cerebral vascular anomalies and developmental delay. Clinical Dysmorphology, 2009, 18, 110-111.	0.1	2
133	The C20orf133 gene is disrupted in a patient with Kabuki syndrome. BMJ Case Reports, 2009, 2009, bcr0620091994-bcr0620091994.	0.2	4
134	Deletion of Pten in the mouse enteric nervous system induces ganglioneuromatosis and mimics intestinal pseudoobstruction. Journal of Clinical Investigation, 2009, 119, 3586-3596.	3.9	52
135	Homozygous mutation of the PHOX2B gene in congenital central hypoventilation syndrome (Ondine's) Tj ETQq1 1 0.784314 rgBT /O	1.1	26
136	Identification of 23TGFBR2and 6TGFBR1gene mutations and genotype-phenotype investigations in 457 patients with Marfan syndrome type I and II, Loeys-Dietz syndrome and related disorders. Human Mutation, 2008, 29, E284-E295.	1.1	86
137	New case of interstitial deletion 12(q15â€q21.2) in a girl with facial dysmorphism and mental retardation. American Journal of Medical Genetics, Part A, 2008, 146A, 93-96.	0.7	16
138	Somatic and germline activating mutations of the ALK kinase receptor in neuroblastoma. Nature, 2008, 455, 967-970.	13.7	787
139	miR-122, a paradigm for the role of microRNAs in the liver. Journal of Hepatology, 2008, 48, 648-656.	1.8	330
140	Stabilization of RNA during laser capture microdissection by performing experiments under argon atmosphere or using ethanol as a solvent in staining solutions. Rna, 2008, 14, 2698-2704.	1.6	68
141	PHOX2B Germline and Somatic Mutations in Late-Onset Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2008, 177, 906-911.	2.5	85
142	Human neural crest cells display molecular and phenotypic hallmarks of stem cells. Human Molecular Genetics, 2008, 17, 3411-3425.	1.4	87
143	Mutations in STAT3 and IL12RB1 impair the development of human IL-17â€“producing T cells. Journal of Experimental Medicine, 2008, 205, 1543-1550.	4.2	406
144	Delineation of Late Onset Hypoventilation Associated with Hypothalamic Dysfunction Syndrome. Pediatric Research, 2008, 64, 689-694.	1.1	63

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145	Hirschsprung disease, associated syndromes and genetics: a review. Journal of Medical Genetics, 2007, 45, 1-14.	1.5	848
146	The C20orf133 gene is disrupted in a patient with Kabuki syndrome. Journal of Medical Genetics, 2007, 44, 562-569.	1.5	56
147	Early Grade Repetition and Inattention Associated With Neurofibromatosis Type 1. Journal of Attention Disorders, 2007, 11, 101-105.	1.5	22
148	Methylation-associated PHOX2B gene silencing is a rare event in human neuroblastoma. European Journal of Cancer, 2007, 43, 2366-2372.	1.3	20
149	The Meckel-Gruber Syndrome Gene, MKS3, Is Mutated in Joubert Syndrome. American Journal of Human Genetics, 2007, 80, 186-194.	2.6	217
150	Mutations in TCF4, Encoding a Class I Basic Helix-Loop-Helix Transcription Factor, Are Responsible for Pitt-Hopkins Syndrome, a Severe Epileptic Encephalopathy Associated with Autonomic Dysfunction. American Journal of Human Genetics, 2007, 80, 988-993.	2.6	264
151	Matthew-Wood Syndrome Is Caused by Truncating Mutations in the Retinol-Binding Protein Receptor Gene STRA6. American Journal of Human Genetics, 2007, 80, 1179-1187.	2.6	174
152	Pleiotropic Effects of CEP290 (NPHP6) Mutations Extend to Meckel Syndrome. American Journal of Human Genetics, 2007, 81, 170-179.	2.6	248
153	Rokitansky Syndrome: Clinical Experience and Results of Sigmoid Vaginoplasty in 23 Young Girls. Journal of Urology, 2007, 177, 1107-1111.	0.2	86
154	Recurrent inverted duplication of 2p with terminal deletion in a patient with the classical phenotype of trisomy 2p23â€pter. American Journal of Medical Genetics, Part A, 2007, 143A, 2417-2422.	0.7	20
155	Polyalanine expansions might not result from unequal crossing-over. Human Mutation, 2007, 28, 1043-1044.	1.1	19
156	Spectrum ofMKS1andMKS3mutations in Meckel syndrome: a genotype-phenotype correlation. Human Mutation, 2007, 28, 523-524.	1.1	92
157	Homozygous silencing of T-box transcription factor EOMES leads to microcephaly with polymicrogyria and corpus callosum agenesis. Nature Genetics, 2007, 39, 454-456.	9.4	181
158	Allele dosage-dependent penetrance of RET proto-oncogene in an Israeli-Arab inbred family segregating Hirschsprung disease. European Journal of Human Genetics, 2007, 15, 242-245.	1.4	13
159	An overview of isolated and syndromic oesophageal atresia. Clinical Genetics, 2007, 71, 392-399.	1.0	70
160	Reply to Salviati et al.. American Journal of Human Genetics, 2006, 79, 596-597.	2.6	11
161	Contiguous Gene Deletion within Chromosome Arm 10q Is Associated with Juvenile Polyposis of Infancy, Reflecting Cooperation between the BMPR1A and PTEN Tumor-Suppressor Genes. American Journal of Human Genetics, 2006, 78, 1066-1074.	2.6	127
162	Array-based comparative genomic hybridisation identifies high frequency of cryptic chromosomal rearrangements in patients with syndromic autism spectrum disorders. Journal of Medical Genetics, 2006, 43, 843-849.	1.5	267

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