Jeanne Amiel

List of Publications by Year
in descending order

200	18,718	14124	15253
papers	citations	h-index	g-index

205	205	205	22175
all docs	docs citations	times ranked	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	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Human Mutation, 2022, 43, 582-594.	1.1	6
3	Discovery of a genetic module essential for assigning leftâ€“right asymmetry in humans and ancestral vertebrates. Nature Genetics, 2022, 54, 62-72.	9.4	16
4	Suleiman-El-Hattab syndrome: a histone modification disorder caused by TASP1 deficiency. Human Molecular Genetics, 2022, 31, 3083-3094.	1.4	3
5	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. Human Molecular Genetics, 2021, 29, 3662-3678.	1.4	14
6	Reply: MN1 gene loss-of-function mutation causes cleft palate in a pedigree. Brain, 2021, 144, e19-e19.	3.7	3
7	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. Nature Communications, 2021, 12, 833.	5.8	41
8	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	3.9	24
9	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	5.8	28
10	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
11	PRICKLE2 revisitedâ€”further evidence implicating PRICKLE2 in neurodevelopmental disorders. European Journal of Human Genetics, 2021, 29, 1235-1244.	1.4	5
12	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
13	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	1.1	8
14	Genetics of craniofacial malformations. Seminars in Fetal and Neonatal Medicine, 2021, 26, 101290.	1.1	5
15	Quality of life and phonatory and morphological outcomes in cognitively unimpaired adolescents with Pierre Robin sequence: a cross-sectional study of 72 patients. Orphanet Journal of Rare Diseases, 2021, 16, 442.	1.2	6
16	Further delineation of the phenotypic spectrum associated with hemizygous lossâ€ofâ€function variants in NONO. American Journal of Medical Genetics, Part A, 2020, 182, 652-658.	0.7	17
17	Generation of an iPSC line (IMAGINi022-A) from a patient carrying a SOX10 missense mutation and presenting with deafness, depigmentation and progressive neurological impairment. Stem Cell Research, 2020, 48, 101936.	0.3	2
18	Overlapping phenotypes between SHORT and Noonan syndromes in patients with PTPN11 pathogenic variants. Clinical Genetics, 2020, 98, 10-18.	1.0	9

#	Article	IF	Citations
19	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guionâ€Almeida type. Human Mutation, 2020, 41, 1372-1382.	1.1	15
20	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
21	SMCHD1 is involved inde novomethylation of theDUX4-encoding D4Z4 macrosatellite. Nucleic Acids Research, 2019, 47, 2822-2839.	6.5	39
22	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
23	De Novo Mutations Affecting the Catalytic CÎ± Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 104, 139-156.	2.6	39
24	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	2.6	30
25	Further delineation of theMECP2duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. Journal of Medical Genetics, 2018, 55, 359-371.	1.5	45
26	Mandibulofacial dysostosis Guion-Almeida type caused by novel EFTUD2 splice site variants in two Asian children. Clinical Dysmorphology, 2018, 27, 31-35.	0.1	21
27	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La RÃ©union Island, in patients with Fryns syndrome. European Journal of Human Genetics, 2018, 26, 340-349.	1.4	27
28	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
29	NONO Detects the Nuclear HIV Capsid to Promote cGAS-Mediated Innate Immune Activation. Cell, 2018, 175, 488-501.e22.	13.5	154
30	Heterozygous Mutations in TBX1 as a Cause of Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4023-4032.	1.8	15
31	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255.	9.4	88
32	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	1.1	90
33	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
34	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	3.8	72
35	Kaposi sarcoma, oral malformations, mitral dysplasia, and scoliosis associated with 7q34â€q36.3 heterozygous terminal deletion. American Journal of Medical Genetics, Part A, 2017, 173, 1858-1865.	0.7	4
36	Targeted molecular investigation in patients within the clinical spectrum of Auriculocondylar syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 938-945.	0.7	11

#	Article	IF	Citations
37	De novo mutations in CBL causing early-onset paediatric moyamoya angiopathy. Journal of Medical Genetics, 2017, 54, 550-557.	1.5	33
38	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. Genetics in Medicine, 2017, 19, 701-710.	1.1	13
39	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	2.6	63
40	Efficiency of prenatal diagnosis in Pierre Robin sequence. Prenatal Diagnosis, 2017, 37, 1169-1175.	1.1	18
41	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. American Journal of Human Genetics, 2017, 101, 995-1005.	2.6	23
42	Respiratory and gastrointestinal dysfunctions associated with auriculoâ€condylar syndrome and a homozygous PLCB4 lossâ€ofâ€function mutation. American Journal of Medical Genetics, Part A, 2016, 170, 1471-1478.	0.7	12
43	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. Human Molecular Genetics, 2016, 25, ddw333.	1.4	38
44	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	2.6	39
45	Large national series of patients with Xq28 duplication involving MECP2: Delineation of brain MRI abnormalities in 30 affected patients. American Journal of Medical Genetics, Part A, 2016, 170, 116-129.	0.7	19
46	Oncologic Phenotype of Peripheral Neuroblastic Tumors Associated With PHOX2B Nonâ€Polyalanine Repeat Expansion Mutations. Pediatric Blood and Cancer, 2016, 63, 71-77.	0.8	14
47	The expanding spectrum of COL2A1 gene variants IN 136 patients with a skeletal dysplasia phenotype. European Journal of Human Genetics, 2016, 24, 992-1000.	1.4	39
48	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
49	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
50	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
51	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
52	Contiguous mutation syndrome in the era of highâ€throughput sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 215-220.	0.6	9
53	Mutations in NONO lead to syndromic intellectual disability and inhibitory synaptic defects. Nature Neuroscience, 2015, 18, 1731-1736.	7.1	65
54	Clinical and molecular delineation of Tetrasomy 9p syndrome: Report of 12 new cases and literature review. American Journal of Medical Genetics, Part A, 2015, 167, 1252-1261.	0.7	20

#	Article	IF	Citations
55	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531.	2.6	47
56	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
57	New insights into genotypeâ€“phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	1.4	97
58	Late-onset congenital central hypoventilation syndrome and a rare PHOX2B gene mutation. Sleep and Breathing, 2015, 19, 55-60.	0.9	20
59	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
60	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	1.1	50
61	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
62	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	1.1	113
63	Mutations in the tricarboxylic acid cycle enzyme, aconitase 2, cause either isolated or syndromic optic neuropathy with encephalopathy and cerebellar atrophy. Journal of Medical Genetics, 2014, 51, 834-838.	1.5	80
64	Rubinstein-Taybi syndrome predisposing to non-WNT, non-SHH, group 3 medulloblastoma. Pediatric Blood and Cancer, 2014, 61, 383-386.	0.8	33
65	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. European Journal of Medical Genetics, 2014, 57, 587-595.	0.7	18
66	An unusual cause of fetal hypomobility:congenital central hypoventilation syndrome associated with hirschsprung disease. European Journal of Pediatrics, 2014, 173, 1607-1609.	1.3	3
67	Identification of Novel Craniofacial Regulatory Domains Located far Upstream ofSOX9and Disrupted in Pierre Robin Sequence. Human Mutation, 2014, 35, 1011-1020.	1.1	69
68	Blepharophimosis, short humeri, developmental delay and hirschsprung disease: Expanding the phenotypic spectrum of MED12 mutations. American Journal of Medical Genetics, Part A, 2014, 164, 1821-1825.	0.7	19
69	PIK3R1 Mutations Cause Syndromic Insulin Resistance with Lipoatrophy. American Journal of Human Genetics, 2013, 93, 141-149.	2.6	162
70	Mutations in KCTD1 Cause Scalp-Ear-Nipple Syndrome. American Journal of Human Genetics, 2013, 92, 621-626.	2.6	65
71	Contribution of rare and common variants determine complex diseasesâ€”Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	0.9	119
72	Autonomic dysfunction of glucose homoeostasis in congenital central hypoventilation syndrome. Acta Paediatrica, International Journal of Paediatrics, 2013, 102, e178-80.	0.7	15

#	Article	IF	Citations
73	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
74	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
75	Clinical and Molecular Spectrum of Renal Malformations in Kabuki Syndrome. Journal of Pediatrics, 2013, 163, 742-746.	0.9	27
76	Phenotypic Spectrum of Simpsonâ€“<scp>G</scp>olabiâ€“<scp>B</scp>ehmel Syndrome in a Series of 42 Cases With a Mutation in <scp>GPC</scp>3 and Review of the Literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 92-105.	0.7	78
77	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
78	Finger creases lend a hand in Kabuki syndrome. European Journal of Medical Genetics, 2013, 56, 556-560.	0.7	11
79	NF-ÎºB signalling requirement for brain myelin formation is shown by genotype/MRI phenotype correlations in patients with Xq28 duplications. European Journal of Human Genetics, 2013, 21, 195-199.	1.4	21
80	FOX gene cluster defects in alveolar capillary dysplasia associated with congenital heart disease. Cardiology in the Young, 2013, 23, 697-704.	0.4	12
81	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
82	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. Journal of Medical Genetics, 2013, 50, 174-186.	1.5	44
83	Congenital Heart Defects in Patients with Deletions Upstream ofSOX9. Human Mutation, 2013, 34, 1628-1631.	1.1	33
84	Contiguous gene deletion of TBX5 and TBX3 leads to a varible phenotype with combined features of holtâ€oram and ulnarâ€mammary syndromes. American Journal of Medical Genetics, Part A, 2013, 161, 1797-1802.	0.7	16
85	Contributions of PHOX2B in the Pathogenesis of Hirschsprung Disease. PLoS ONE, 2013, 8, e54043.	1.1	30
86	Chromosome 21 Scan in Down Syndrome Reveals DSCAM as a Predisposing Locus in Hirschsprung Disease. PLoS ONE, 2013, 8, e62519.	1.1	22
87	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
88	EFTUD2haploinsufficiency leads to syndromic oesophageal atresia. Journal of Medical Genetics, 2012, 49, 737-746.	1.5	89
89	IntragenicCAMTA1rearrangements cause non-progressive congenital ataxia with or without intellectual disability. Journal of Medical Genetics, 2012, 49, 400-408.	1.5	39
90	Mutations of TSEN and CASK genes are prevalent in pontocerebellar hypoplasias type 2 and 4. Brain, 2012, 135, e199-e199.	3.7	18

#	Article	IF	Citations
91	MicroRNAs in Genetic Disease: Rethinking the Dosage. Current Gene Therapy, 2012, 12, 292-300.	0.9	10
92	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	9.4	194
93	Two independent de novo mutations as a cause for neurofibromatosis type 1 and Noonan syndrome in a single family. American Journal of Medical Genetics, Part A, 2012, 158A, 2290-2291.	0.7	4
94	Constitutional NRAS mutations are rare among patients with Noonan syndrome or juvenile myelomonocytic leukemia. American Journal of Medical Genetics, Part A, 2012, 158A, 2407-2411.	0.7	20
95	miRNA, Development and Disease. Advances in Genetics, 2012, 80, 1-36.	0.8	60
96	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
97	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. American Journal of Human Genetics, 2012, 91, 950-957.	2.6	95
98	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
99	Novel comprehensive diagnostic strategy in Pitt-Hopkins syndrome: Clinical score and further delineation of the TCF4 mutational spectrum. Human Mutation, 2012, 33, 64-72.	1.1	102
100	Autonomic neurocristopathy-associated mutations in PHOX2B dysregulate Sox10 expression. Journal of Clinical Investigation, 2012, 122, 3145-3158.	3.9	89
101	Cis-Regulatory Disruption at the SOX9 Locus as a Cause of Pierre Robin Sequence. , 2012, , 123-136.		0
102	RET and GDNF mutations are rare in fetuses with renal agenesis or other severe kidney development defects. Journal of Medical Genetics, 2011, 48, 497-504.	1.5	60
103	Genetic Factors in Isolated and Syndromic Esophageal Atresia. Journal of Pediatric Gastroenterology and Nutrition, 2011, 52, S6-8.	0.9	22
104	Dissection of the MYCN locus in Feingold syndrome and isolated oesophageal atresia. European Journal of Human Genetics, 2011, 19, 602-606.	1.4	24
105	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
106	Germline gain-of-function mutations of ALK disrupt central nervous system development. Human Mutation, 2011, 32, 272-276.	1.1	38
107	Pregnancy in women heterozygous for MCT8 mutations: risk of maternal hypothyroxinemia and fetal care. European Journal of Endocrinology, 2011, 164, 309-314.	1.9	19
108	Germline deletion of the miR-17â¼92 cluster causes skeletal and growth defects in humans. Nature Genetics, 2011, 43, 1026-1030.	9.4	275

#	Article	IF	Citations
109	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
110	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
111	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
112	IDH2 Mutations in Patients with <scp>d</scp> -2-Hydroxyglutaric Aciduria. Science, 2010, 330, 336-336.	6.0	177
113	Late-onset central hypoventilation presenting as extubation failure. Israel Medical Association Journal, 2010, 12, 249-50.	0.1	7
114	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
115	PHOX2B in respiratory control: Lessons from congenital central hypoventilation syndrome and its mouse models. Respiratory Physiology and Neurobiology, 2009, 168, 125-132.	0.7	63
116	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
117	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	1.1	126
118	Interaction between a chromosome 10RETenhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. Human Mutation, 2009, 30, 771-775.	1.1	57
119	CC2D2Amutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. Human Mutation, 2009, 30, 1574-1582.	1.1	80
120	Cholinergic switch associated with morphological differentiation in neuroblastoma. Journal of Pathology, 2009, 219, 463-472.	2.1	26
121	Hypertelorismâ€microtiaâ€clefting syndrome (HMC syndrome): prenatal diagnosis in two siblings. Prenatal Diagnosis, 2009, 29, 1064-1065.	1.1	0
122	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
123	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	9.4	364
124	Sporadic case of unusual facies, cerebral vascular anomalies and developmental delay. Clinical Dysmorphology, 2009, 18, 110-111.	0.1	2
125	Homozygous mutation of the PHOX2B gene in congenital central hypoventilation syndrome (Ondine's) Tj ETQq1 1 0.784314 rgBT /O	1.1	26
126	Somatic and germline activating mutations of the ALK kinase receptor in neuroblastoma. Nature, 2008, 455, 967-970.	13.7	787

#	Article	IF	Citations
127	A human mutation in Phox2b causes lack of CO ₂ chemosensitivity, fatal central apnea, and specific loss of parafacial neurons. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 1067-1072.	3.3	271
128	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
129	Delineation of Late Onset Hypoventilation Associated with Hypothalamic Dysfunction Syndrome. Pediatric Research, 2008, 64, 689-694.	1.1	63
130	Hirschsprung disease, associated syndromes and genetics: a review. Journal of Medical Genetics, 2007, 45, 1-14.	1.5	848
131	Methylation-associated PHOX2B gene silencing is a rare event in human neuroblastoma. European Journal of Cancer, 2007, 43, 2366-2372.	1.3	20
132	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
133	Deletions at the SOX10 Gene Locus Cause Waardenburg Syndrome Types 2 and 4. American Journal of Human Genetics, 2007, 81, 1169-1185.	2.6	216
134	Polyalanine expansions might not result from unequal crossing-over. Human Mutation, 2007, 28, 1043-1044.	1.1	19
135	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
136	An overview of isolated and syndromic oesophageal atresia. Clinical Genetics, 2007, 71, 392-399.	1.0	70
137	Polymorphic length of FOXE1 alanine stretch: evidence for genetic susceptibility to thyroid dysgenesis. Human Genetics, 2007, 122, 467-476.	1.8	61
138	Congenital pontocerebellar atrophy and telencephalic defects in three siblings: a new subtype. Acta Neuropathologica, 2007, 114, 387-399.	3.9	7
139	Loss-of-Function Mutations in Euchromatin Histone Methyl Transferase 1 (EHMT1) Cause the 9q34 Subtelomeric Deletion Syndrome. American Journal of Human Genetics, 2006, 79, 370-377.	2.6	343
140	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
141	Molecular Bases of Human Neurocristopathies. , 2006, 589, 213-234.		79
142	The French Congenital Central Hypoventilation Syndrome Registry. Chest, 2005, 127, 72-79.	0.4	199
143	Autonomic Function in Children With Congenital Central Hypoventilation Syndrome and Their Families. Chest, 2005, 128, 2478-2484.	0.4	42
144	Failure to detect an 8p22â€“8p23.1 duplication in patients with Kabuki (Niikawaâ€“Kuroki) syndrome. European Journal of Human Genetics, 2005, 13, 690-693.	1.4	28

#	Article	IF	Citations
145	Adams-Oliver syndrome and hepatoportal sclerosis: Occasional association or common mechanism?. American Journal of Medical Genetics, Part A, 2005, 135A, 186-189.	0.7	52
146	Molecular consequences of PHOX2B missense, frameshift and alanine expansion mutations leading to autonomic dysfunction. Human Molecular Genetics, 2005, 14, 3697-3708.	1.4	135
147	CHARGE Syndrome Includes Hypogonadotropic Hypogonadism and Abnormal Olfactory Bulb Development. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 5621-5626.	1.8	142
148	PHOX2B Genotype Allows for Prediction of Tumor Risk in Congenital Central Hypoventilation Syndrome. American Journal of Human Genetics, 2005, 76, 421-426.	2.6	222
149	Germline mutations of the paired-like homeobox 2B (PHOX2B) gene in neuroblastoma. Cancer Letters, 2005, 228, 51-58.	3.2	63
150	Phenotypic spectrum of CHARGE syndrome in fetuses with CHD7 truncating mutations correlates with expression during human development. Journal of Medical Genetics, 2005, 43, 211-317.	1.5	199
151	Genetics and Early Disturbances of Breathing Control: The Genetics of Childhood Disease and Development: A Series of Review Articles. Pediatric Research, 2004, 55, 729-733.	1.1	64
152	Polyalanine expansions in human. Human Molecular Genetics, 2004, 13, R235-R243.	1.4	131
153	Molecular screening of theZFHX1B gene in prenatally diagnosed isolated agenesis of thecorpus callosum. Prenatal Diagnosis, 2004, 24, 298-301.	1.1	9
154	PHOX2B gene mutation in a patient with late-onset central hypoventilation. Pediatric Pulmonology, 2004, 38, 349-351.	1.0	50
155	Germline Mutations of the Pairedâ€“Like Homeobox 2B (PHOX2B) Gene in Neuroblastoma. American Journal of Human Genetics, 2004, 74, 761-764.	2.6	288
156	Lamin A Truncation in Hutchinson-Gilford Progeria. Science, 2003, 300, 2055-2055.	6.0	1,247
157	Exclusion ofRNX as a major gene in congenital central hypoventilation syndrome (CCHS, Ondine's) Tj ETQq1 1 0.784314 rgBT /Overlo	2.4	16
158	Polyalanine expansion and frameshift mutations of the paired-like homeobox gene PHOX2B in congenital central hypoventilation syndrome. Nature Genetics, 2003, 33, 459-461.	9.4	771
159	Antenatal manifestations of mitochondrial respiratory chain deficiency. Journal of Pediatrics, 2003, 143, 208-212.	0.9	129
160	Noradrenergic neuronal development is impaired by mutation of the proneural HASH-1 gene in congenital central hypoventilation syndrome (Ondine's curse). Human Molecular Genetics, 2003, 12, 3173-3180.	1.4	72
161	Spectrum of NSD1 mutations in Sotos and Weaver syndromes. Journal of Medical Genetics, 2003, 40, 436-440.	1.5	116
162	A genomic rearrangement resulting in a tandem duplication is associated with split hand-split foot malformation 3 (SHFM3) at 10q24. Human Molecular Genetics, 2003, 12, 1959-1971.	1.4	88

#	Article	IF	Citations
163	Expression of the SMADIP1 gene during early human development. Mechanisms of Development, 2002, 114, 187-191.	1.7	49
164	Facial appearance in persistent hyperinsulinemic hypoglycemia. American Journal of Medical Genetics Part A, 2002, 111, 130-133.	2.4	29
165	Overgrowth and trisomy 15q26.1-qter including the IGF1 receptor gene: report of two families and review of the literature. European Journal of Human Genetics, 2002, 10, 699-706.	1.4	44
166	Segregation at three loci explains familial and population risk in Hirschsprung disease. Nature Genetics, 2002, 31, 89-93.	9.4	269
167	Pierre Robin Sequence: A series of 117 consecutive cases. Journal of Pediatrics, 2001, 139, 588-590.	0.9	136
168	Large-Scale Deletions and SMADIP1 Truncating Mutations in Syndromic Hirschsprung Disease with Involvement of Midline Structures. American Journal of Human Genetics, 2001, 69, 1370-1377.	2.6	105
169	Renal coloboma syndrome. Ophthalmology, 2001, 108, 1912-1916.	2.5	100
170	Functional Characterization of Three Mutations of the Endothelin B Receptor Gene in Patients With Hirschsprungâ€™s Disease: Evidence for Selective Loss of Gi Coupling. Molecular Medicine, 2001, 7, 115-124.	1.9	53
171	Absent lacrimal ducts, distichiasis, dysmorphic features, and brachydactyly: a case report. Clinical Dysmorphology, 2001, 10, 253-255.	0.1	4
172	Hypertelorism-Microtia-Clefting Syndrome (Bixler syndrome): report of two unrelated cases. Clinical Dysmorphology, 2001, 10, 15-18.	0.1	7
173	PAX2 mutations in oligomeganephronia. Kidney International, 2001, 59, 457-462.	2.6	106
174	Prenatal diagnosis of respiratory chain deficiency by direct mutation screening. Prenatal Diagnosis, 2001, 21, 602-604.	1.1	15
175	Temporal bone anomaly proposed as a major criteria for diagnosis of CHARGE syndrome. American Journal of Medical Genetics Part A, 2001, 99, 124-127.	2.4	109
176	Five novel frameshift mutations in exon 3 and 4 of the MECP2 gene identified in Rett patients: Consequences for the molecular diagnosis strategy. Human Mutation, 2001, 18, 251-252.	1.1	2
177	A novel automated strategy for screening cryptic telomeric rearrangements in children with idiopathic mental retardation. European Journal of Human Genetics, 2001, 9, 319-327.	1.4	55
178	TP63 gene mutation in ADULT syndrome. European Journal of Human Genetics, 2001, 9, 642-645.	1.4	82
179	MECP2 mutation in non-fatal, non-progressive encephalopathy in a male. Journal of Medical Genetics, 2001, 38, 171-174.	1.5	87
180	Hirschsprung disease, associated syndromes, and genetics: a review. Journal of Medical Genetics, 2001, 38, 729-739.	1.5	394

#	Article	IF	Citations
181	Expression of thePAX2 gene in human embryos and exclusion in the CHARGE syndrome. American Journal of Medical Genetics Part A, 2000, 93, 85-88.	2.4	55
182	Determination of enzyme activities for prenatal diagnosis of respiratory chain deficiency. Prenatal Diagnosis, 2000, 20, 732-737.	1.1	39
183	PAX2 mutations in renalâ€“coloboma syndrome: mutational hotspot and germline mosaicism. European Journal of Human Genetics, 2000, 8, 820-826.	1.4	77
184	Vestibular anomalies in CHARGE syndrome: investigations on and consequences for postural development. European Journal of Pediatrics, 2000, 159, 569-574.	1.3	75
185	Mutations of the SCO1 Gene in Mitochondrial Cytochrome c Oxidase Deficiency with Neonatal-Onset Hepatic Failure and Encephalopathy. American Journal of Human Genetics, 2000, 67, 1104-1109.	2.6	157
186	Mutations of theSCO1Gene in Mitochondrial CytochromecOxidase Deficiency with Neonatalâ€Onset Hepatic Failure and Encephalopathy. American Journal of Human Genetics, 2000, 67, 1104-1109.	2.6	322
187	Facial anomalies inD-2-hydroxyglutaric aciduria. American Journal of Medical Genetics Part A, 1999, 86, 124-129.	2.4	17
188	Recurrent Williams-Beuren Syndrome in a Sibship Suggestive of Maternal Germ-Line Mosaicism. American Journal of Human Genetics, 1999, 64, 1475-1478.	2.6	27
189	Ebstein anomaly associated with rearrangements of chromosomal region 11q. , 1998, 80, 157-159.		27
190	Expression of theRET proto-oncogene in human Embryos. , 1998, 80, 481-486.		55
191	SOX10 mutations in patients with Waardenburg-Hirschsprung disease. Nature Genetics, 1998, 18, 171-173.	9.4	733
192	Mutations of the RET-GDNF Signaling Pathway in Ondine's Curse. American Journal of Human Genetics, 1998, 62, 715-717.	2.6	115
193	Mutation of the MITF gene in albinism-deafness syndrome (Tietz syndrome). Clinical Dysmorphology, 1998, 7, 17???20.	0.1	86
194	Mutation of the RET ligand, neurturin, supports multigenic inheritance in Hirschsprung disease [published erratum appears in Hum Mol Genet 1998 Oct;7(11):1831]. Human Molecular Genetics, 1998, 7, 1449-1452.	1.4	145
195	Endothelin-3 Gene Mutations in Isolated and Syndromic Hirschsprung Disease. European Journal of Human Genetics, 1997, 5, 247-251.	1.4	67
196	Mutation of the endothelin-3 gene in the Waardenburg-Hirschsprung disease (Shah-Waardenburg) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	9.4	425
197	Germline mutations of the RET ligand GDNF are not sufficient to cause Hirschsprung disease. Nature Genetics, 1996, 14, 345-347.	9.4	203
198	Heterozygous endothelin receptor B (EDNRB) mutations in isolated Hirschsprung disease. Human Molecular Genetics, 1996, 5, 355-357.	1.4	174

#	Article	IF	Citations
199	Mutation of the endothelin-receptor B gene in Waardenburg-Hirschsprung disease. Human Molecular Genetics, 1995, 4, 2407-2409.	1.4	214
200	Diversity of RET proto-oncogene mutations in familial and sporadic Hirschsprung disease. Human Molecular Genetics, 1995, 4, 1381-1386.	1.4	342