Jeanne Amiel

List of Publications by Year
in descending order

200	18,718	12322	13365
papers	citations	69 h-index	130 g-index

205	205	205	20378
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Lamin A Truncation in Hutchinson-Gilford Progeria. Science, 2003, 300, 2055-2055.	6.0	1,247
2	Hirschsprung disease, associated syndromes and genetics: a review. Journal of Medical Genetics, 2007, 45, 1-14.	1.5	848
3	Somatic and germline activating mutations of the ALK kinase receptor in neuroblastoma. Nature, 2008, 455, 967-970.	13.7	787
4	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
5	SOX10 mutations in patients with Waardenburg-Hirschsprung disease. Nature Genetics, 1998, 18, 171-173.	9.4	733
6	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
7	Hirschsprung disease, associated syndromes, and genetics: a review. Journal of Medical Genetics, 2001, 38, 729-739.	1.5	394
8	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	9.4	364
9	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
10	Diversity of RET proto-oncogene mutations in familial and sporadic Hirschsprung disease. Human Molecular Genetics, 1995, 4, 1381-1386.	1.4	342
11	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
12	Germline Mutations of the Pairedâ€“Like Homeobox 2B (PHOX2B) Gene in Neuroblastoma. American Journal of Human Genetics, 2004, 74, 761-764.	2.6	288
13	Germline deletion of the miR-17â¼92 cluster causes skeletal and growth defects in humans. Nature Genetics, 2011, 43, 1026-1030.	9.4	275
14	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
15	Segregation at three loci explains familial and population risk in Hirschsprung disease. Nature Genetics, 2002, 31, 89-93.	9.4	269
16	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
17	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
18	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

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19	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
20	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
21	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
22	Mutation of the endothelin-receptor B gene in Waardenburg-Hirschsprung disease. Human Molecular Genetics, 1995, 4, 2407-2409.	1.4	214
23	Germline mutations of the RET ligand GDNF are not sufficient to cause Hirschsprung disease. Nature Genetics, 1996, 14, 345-347.	9.4	203
24	The French Congenital Central Hypoventilation Syndrome Registry. Chest, 2005, 127, 72-79.	0.4	199
25	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
26	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	9.4	194
27	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
28	IDH2 Mutations in Patients with <scp>d</scp> -2-Hydroxyglutaric Aciduria. Science, 2010, 330, 336-336.	6.0	177
29	Heterozygous endothelin receptor B (EDNRB) mutations in isolated Hirschsprung disease. Human Molecular Genetics, 1996, 5, 355-357.	1.4	174
30	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
31	PIK3R1 Mutations Cause Syndromic Insulin Resistance with Lipoatrophy. American Journal of Human Genetics, 2013, 93, 141-149.	2.6	162
32	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
33	NONO Detects the Nuclear HIV Capsid to Promote cGAS-Mediated Innate Immune Activation. Cell, 2018, 175, 488-501.e22.	13.5	154
34	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
35	CHARGE Syndrome Includes Hypogonadotropic Hypogonadism and Abnormal Olfactory Bulb Development. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 5621-5626.	1.8	142
36	Pierre Robin Sequence: A series of 117 consecutive cases. Journal of Pediatrics, 2001, 139, 588-590.	0.9	136

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37	Molecular consequences of PHOX2B missense, frameshift and alanine expansion mutations leading to autonomic dysfunction. Human Molecular Genetics, 2005, 14, 3697-3708.	1.4	135
38	Polyalanine expansions in human. Human Molecular Genetics, 2004, 13, R235-R243.	1.4	131
39	Antenatal manifestations of mitochondrial respiratory chain deficiency. Journal of Pediatrics, 2003, 143, 208-212.	0.9	129
40	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	1.1	126
41	Contribution of rare and common variants determine complex diseasesâ€"Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	0.9	119
42	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
43	Spectrum of NSD1 mutations in Sotos and Weaver syndromes. Journal of Medical Genetics, 2003, 40, 436-440.	1.5	116
44	Mutations of the RET-GDNF Signaling Pathway in Ondine's Curse. American Journal of Human Genetics, 1998, 62, 715-717.	2.6	115
45	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	1.1	113
46	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
47	PAX2 mutations in oligomeganephronia. Kidney International, 2001, 59, 457-462.	2.6	106
48	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
49	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
50	Renal coloboma syndrome. Ophthalmology, 2001, 108, 1912-1916.	2.5	100
51	New insights into genotypeâ€"phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	1.4	97
52	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
53	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
54	EFTUD2haploinsufficiency leads to syndromic oesophageal atresia. Journal of Medical Genetics, 2012, 49, 737-746.	1.5	89

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55	Autonomic neurocristopathy-associated mutations in PHOX2B dysregulate Sox10 expression. Journal of Clinical Investigation, 2012, 122, 3145-3158.	3.9	89
56	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
57	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255.	9.4	88
58	MECP2 mutation in non-fatal, non-progressive encephalopathy in a male. Journal of Medical Genetics, 2001, 38, 171-174.	1.5	87
59	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
60	Mutation of the MITF gene in albinism-deafness syndrome (Tietz syndrome). Clinical Dysmorphology, 1998, 7, 17???20.	0.1	86
61	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
62	TP63 gene mutation in ADULT syndrome. European Journal of Human Genetics, 2001, 9, 642-645.	1.4	82
63	CC2D2Amutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. Human Mutation, 2009, 30, 1574-1582.	1.1	80
64	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
65	Molecular Bases of Human Neurocristopathies. , 2006, 589, 213-234.		79
66	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
67	PAX2 mutations in renalâ€“coloboma syndrome: mutational hotspot and germline mosaicism. European Journal of Human Genetics, 2000, 8, 820-826.	1.4	77
68	Vestibular anomalies in CHARGE syndrome: investigations on and consequences for postural development. European Journal of Pediatrics, 2000, 159, 569-574.	1.3	75
69	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
70	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
71	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	3.8	72
72	An overview of isolated and syndromic oesophageal atresia. Clinical Genetics, 2007, 71, 392-399.	1.0	70

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73	Identification of Novel Craniofacial Regulatory Domains Located far Upstream ofSOX9and Disrupted in Pierre Robin Sequence. Human Mutation, 2014, 35, 1011-1020.	1.1	69
74	Endothelin-3 Gene Mutations in Isolated and Syndromic Hirschsprung Disease. European Journal of Human Genetics, 1997, 5, 247-251.	1.4	67
75	Mutations in KCTD1 Cause Scalp-Ear-Nipple Syndrome. American Journal of Human Genetics, 2013, 92, 621-626.	2.6	65
76	Mutations in NONO lead to syndromic intellectual disability and inhibitory synaptic defects. Nature Neuroscience, 2015, 18, 1731-1736.	7.1	65
77	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
78	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
79	Germline mutations of the paired-like homeobox 2B (PHOX2B) gene in neuroblastoma. Cancer Letters, 2005, 228, 51-58.	3.2	63
80	Delineation of Late Onset Hypoventilation Associated with Hypothalamic Dysfunction Syndrome. Pediatric Research, 2008, 64, 689-694.	1.1	63
81	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
82	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
83	Polymorphic length of FOXE1 alanine stretch: evidence for genetic susceptibility to thyroid dysgenesis. Human Genetics, 2007, 122, 467-476.	1.8	61
84	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
85	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
86	miRNA, Development and Disease. Advances in Genetics, 2012, 80, 1-36.	0.8	60
87	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
88	Interaction between a chromosome 10RETenhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. Human Mutation, 2009, 30, 771-775.	1.1	57
89	Expression of theRET proto-oncogene in human Embryos. , 1998, 80, 481-486.		55
90	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

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91	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
92	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
93	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
94	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
95	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
96	PHOX2B gene mutation in a patient with late-onset central hypoventilation. Pediatric Pulmonology, 2004, 38, 349-351.	1.0	50
97	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	1.1	50
98	Expression of the SMADIP1 gene during early human development. Mechanisms of Development, 2002, 114, 187-191.	1.7	49
99	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
100	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531.	2.6	47
101	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
102	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
103	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. Journal of Medical Genetics, 2013, 50, 174-186.	1.5	44
104	Autonomic Function in Children With Congenital Central Hypoventilation Syndrome and Their Families. Chest, 2005, 128, 2478-2484.	0.4	42
105	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. Nature Communications, 2021, 12, 833.	5.8	41
106	Determination of enzyme activities for prenatal diagnosis of respiratory chain deficiency. Prenatal Diagnosis, 2000, 20, 732-737.	1.1	39
107	IntragenicCAMTA1rearrangements cause non-progressive congenital ataxia with or without intellectual disability. Journal of Medical Genetics, 2012, 49, 400-408.	1.5	39
108	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	2.6	39

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109	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
110	SMCHD1 is involved inde novomethylation of theDUX4-encoding D4Z4 macrosatellite. Nucleic Acids Research, 2019, 47, 2822-2839.	6.5	39
111	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
112	Germline gain-of-function mutations of ALK disrupt central nervous system development. Human Mutation, 2011, 32, 272-276.	1.1	38
113	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. Human Molecular Genetics, 2016, 25, ddw333.	1.4	38
114	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
115	Congenital Heart Defects in Patients with Deletions Upstream ofSOX9. Human Mutation, 2013, 34, 1628-1631.	1.1	33
116	Rubinstein-Taybi syndrome predisposing to non-WNT, non-SHH, group 3 medulloblastoma. Pediatric Blood and Cancer, 2014, 61, 383-386.	0.8	33
117	De novo mutations in CBL causing early-onset paediatric moyamoya angiopathy. Journal of Medical Genetics, 2017, 54, 550-557.	1.5	33
118	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
119	Contributions of PHOX2B in the Pathogenesis of Hirschsprung Disease. PLoS ONE, 2013, 8, e54043.	1.1	30
120	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
121	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	2.6	30
122	Facial appearance in persistent hyperinsulinemic hypoglycemia. American Journal of Medical Genetics Part A, 2002, 111, 130-133.	2.4	29
123	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
124	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	5.8	28
125	Ebstein anomaly associated with rearrangements of chromosomal region 11q. , 1998, 80, 157-159.		27
126	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

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127	Clinical and Molecular Spectrum of Renal Malformations in Kabuki Syndrome. Journal of Pediatrics, 2013, 163, 742-746.	0.9	27
128	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
129	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
130	Homozygous mutation of the PHOX2B gene in congenital central hypoventilation syndrome (Ondine's) Tj ETQq0 0 0 rgBT /Overlock 10	1.1	26
131	Cholinergic switch associated with morphological differentiation in neuroblastoma. Journal of Pathology, 2009, 219, 463-472.	2.1	26
132	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
133	Dissection of the MYCN locus in Feingold syndrome and isolated oesophageal atresia. European Journal of Human Genetics, 2011, 19, 602-606.	1.4	24
134	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	3.9	24
135	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
136	Genetic Factors in Isolated and Syndromic Esophageal Atresia. Journal of Pediatric Gastroenterology and Nutrition, 2011, 52, S6-8.	0.9	22
137	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
138	Chromosome 21 Scan in Down Syndrome Reveals DSCAM as a Predisposing Locus in Hirschsprung Disease. PLoS ONE, 2013, 8, e62519.	1.1	22
139	NF-Î°B 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
140	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
141	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
142	Methylation-associated PHOX2B gene silencing is a rare event in human neuroblastoma. European Journal of Cancer, 2007, 43, 2366-2372.	1.3	20
143	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
144	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

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145	Late-onset congenital central hypoventilation syndrome and a rare PHOX2B gene mutation. Sleep and Breathing, 2015, 19, 55-60.	0.9	20
146	Polyalanine expansions might not result from unequal crossing-over. Human Mutation, 2007, 28, 1043-1044.	1.1	19
147	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
148	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
149	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
150	Mutations of TSEN and CASK genes are prevalent in pontocerebellar hypoplasias type 2 and 4. Brain, 2012, 135, e199-e199.	3.7	18
151	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
152	Efficiency of prenatal diagnosis in Pierre Robin sequence. Prenatal Diagnosis, 2017, 37, 1169-1175.	1.1	18
153	Facial anomalies inD-2-hydroxyglutaric aciduria. American Journal of Medical Genetics Part A, 1999, 86, 124-129.	2.4	17
154	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
155	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
156	Exclusion ofRNX as a major gene in congenital central hypoventilation syndrome (CCHS, Ondine's) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	2.4	16
157	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
158	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
159	Prenatal diagnosis of respiratory chain deficiency by direct mutation screening. Prenatal Diagnosis, 2001, 21, 602-604.	1.1	15
160	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
161	Autonomic dysfunction of glucose homoeostasis in congenital central hypoventilation syndrome. Acta Paediatrica, International Journal of Paediatrics, 2013, 102, e178-80.	0.7	15
162	Heterozygous Mutations in TBX1 as a Cause of Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4023-4032.	1.8	15

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163	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guionâ€Almeida type. Human Mutation, 2020, 41, 1372-1382.	1.1	15
164	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
165	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. Human Molecular Genetics, 2021, 29, 3662-3678.	1.4	14
166	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
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