Tania Attie-Bitach

List of Publications by Year
in descending order

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196 papers	14,805 citations	12303 69 h-index	20900 115 g-index
209 all docs	209 docs citations	209 times ranked	15272 citing authors

#	Article	IF	Citations
1	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
2	The ciliary gene RPGRIP1L is mutated in cerebello-oculo-renal syndrome (Joubert syndrome type B) and Meckel syndrome. Nature Genetics, 2007, 39, 875-881.	9.4	442
3	Mutation of the endothelin-3 gene in the Waardenburg-Hirschsprung disease (Shah-Waardenburg) Tj ETQq1 1 0.784314 rgBT /Overlock	9.4	425
4	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	2.6	352
5	Diversity of RET proto-oncogene mutations in familial and sporadic Hirschsprung disease. Human Molecular Genetics, 1995, 4, 1381-1386.	1.4	342
6	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	9.4	326
7	Planar Cell Polarity Acts Through Septins to Control Collective Cell Movement and Ciliogenesis. Science, 2010, 329, 1337-1340.	6.0	309
8	Segregation at three loci explains familial and population risk in Hirschsprung disease. Nature Genetics, 2002, 31, 89-93.	9.4	269
9	The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome and the wpk rat. Nature Genetics, 2006, 38, 191-196.	9.4	266
10	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	9.4	261
11	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	9.4	255
12	Pleiotropic Effects of CEP290 (NPHP6) Mutations Extend to Meckel Syndrome. American Journal of Human Genetics, 2007, 81, 170-179.	2.6	248
13	The Meckelâ€“Gruber Syndrome proteins MKS1 and meckelin interact and are required for primary cilium formation. Human Molecular Genetics, 2007, 16, 173-186.	1.4	245
14	Diverse phenotypes associated with exon 10 mutations of the RET proto-oncogene. Human Molecular Genetics, 1994, 3, 2163-2168.	1.4	239
15	Mutations in the neuronal Î²-tubulin subunit TUBB3 result in malformation of cortical development and neuronal migration defects. Human Molecular Genetics, 2010, 19, 4462-4473.	1.4	231
16	The Meckel-Gruber Syndrome Gene, MKS3, Is Mutated in Joubert Syndrome. American Journal of Human Genetics, 2007, 80, 186-194.	2.6	217
17	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
18	Mutation of the endothelin-receptor B gene in Waardenburg-Hirschsprung disease. Human Molecular Genetics, 1995, 4, 2407-2409.	1.4	214

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19	Germline mutations of the RET ligand GDNF are not sufficient to cause Hirschsprung disease. Nature Genetics, 1996, 14, 345-347.	9.4	203
20	KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	9.4	203
21	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
22	PAX8,TITF1, andFOXE1Gene Expression Patterns during Human Development: New Insights into Human Thyroid Development and Thyroid Dysgenesis-Associated Malformations. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 455-462.	1.8	195
23	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
24	Truncating Neurotrypsin Mutation in Autosomal Recessive Nonsyndromic Mental Retardation. Science, 2002, 298, 1779-1781.	6.0	176
25	Heterozygous endothelin receptor B (EDNRB) mutations in isolated Hirschsprung disease. Human Molecular Genetics, 1996, 5, 355-357.	1.4	174
26	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
27	Neurological Phenotype in Waardenburg Syndrome Type 4 Correlates with Novel SOX10 Truncating Mutations and Expression in Developing Brain. American Journal of Human Genetics, 2000, 66, 1496-1503.	2.6	172
28	JAGGED1 Gene Expression During Human Embryogenesis Elucidates the Wide Phenotypic Spectrum of Alagille Syndrome. Hepatology, 2000, 32, 574-581.	3.6	161
29	Cleft lip/palate and CDH1/E-cadherin mutations in families with hereditary diffuse gastric cancer. Journal of Medical Genetics, 2005, 43, 138-142.	1.5	161
30	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	9.4	157
31	Cobblestone lissencephaly: neuropathological subtypes and correlations with genes of dystroglycanopathies. Brain, 2012, 135, 469-482.	3.7	151
32	Impaired Mitochondrial Glutamate Transport in Autosomal Recessive Neonatal Myoclonic Epilepsy. American Journal of Human Genetics, 2005, 76, 334-339.	2.6	149
33	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
34	Expression of the Sonic hedgehog (SHH) Gene during Early Human Development and Phenotypic Expression of New Mutations Causing Holoprosencephaly. Human Molecular Genetics, 1999, 8, 1683-1689.	1.4	137
35	Evidence for and against vertical transmission for severe acute respiratory syndrome coronavirus 2. American Journal of Obstetrics and Gynecology, 2020, 223, 91.e1-91.e4.	0.7	137
36	Disruption of a Ciliary B9 Protein Complex Causes Meckel Syndrome. American Journal of Human Genetics, 2011, 89, 94-110.	2.6	136

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37	Pitchfork Regulates Primary Cilia Disassembly and Left-Right Asymmetry. Developmental Cell, 2010, 19, 66-77.	3.1	133
38	Identification of Mutations in TMEM5 and ISPD as a Cause of Severe Cobblestone Lissencephaly. American Journal of Human Genetics, 2012, 91, 1135-1143.	2.6	126
39	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
40	TCTN3 Mutations Cause Mohr-Majewski Syndrome. American Journal of Human Genetics, 2012, 91, 372-378.	2.6	123
41	The oral-facial-digital syndrome gene C2CD3 encodes a positive regulator of centriole elongation. Nature Genetics, 2014, 46, 905-911.	9.4	121
42	Antenatal Presentation of Bardet-Biedl Syndrome May Mimic Meckel Syndrome. American Journal of Human Genetics, 2005, 76, 493-504.	2.6	120
43	TMEM107 recruits ciliopathy proteins to subdomains of the ciliary transition zone and causes JoubertÂsyndrome. Nature Cell Biology, 2016, 18, 122-131.	4.6	118
44	Inappropriate p53 activation during development induces features of CHARGE syndrome. Nature, 2014, 514, 228-232.	13.7	117
45	Mutations of the RET-GDNF Signaling Pathway in Ondine's Curse. American Journal of Human Genetics, 1998, 62, 715-717.	2.6	115
46	Expression of PKD1 and PKD2 Transcripts and Proteins in Human Embryo and during Normal Kidney Development. American Journal of Pathology, 2002, 160, 973-983.	1.9	113
47	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	1.1	113
48	PAX2 mutations in oligomeganephronia. Kidney International, 2001, 59, 457-462.	2.6	106
49	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	15.2	103
50	Renal coloboma syndrome. Ophthalmology, 2001, 108, 1912-1916.	2.5	100
51	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
52	New insights into genotypeâ€“phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	1.4	97
53	A Gene for Meckel Syndrome Maps to Chromosome 11q13. American Journal of Human Genetics, 1998, 63, 1095-1101.	2.6	95
54	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	2.3	95

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55	Spectrum ofMKS1andMKS3mutations in Meckel syndrome: a genotype-phenotype correlation. Human Mutation, 2007, 28, 523-524.	1.1	92
56	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
57	Phenotypic spectrum ofSTRA6mutations: from Matthew-Wood syndrome to non-lethal anophthalmia. Human Mutation, 2009, 30, E673-E681.	1.1	89
58	EFTUD2haploinsufficiency leads to syndromic oesophageal atresia. Journal of Medical Genetics, 2012, 49, 737-746.	1.5	89
59	Severe Prenatal Renal Anomalies Associated with Mutations in HNF1B or PAX2 Genes. Clinical Journal of the American Society of Nephrology: CJASN, 2013, 8, 1179-1187.	2.2	87
60	Long-Chain Fatty Acid Oxidation during Early Human Development. Pediatric Research, 2005, 57, 755-759.	1.1	86
61	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	1.5	85
62	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	6.0	84
63	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
64	CC2D2Amutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. Human Mutation, 2009, 30, 1574-1582.	1.1	80
65	PAX2 mutations in renalâ€“coloboma syndrome: mutational hotspot and germline mosaicism. European Journal of Human Genetics, 2000, 8, 820-826.	1.4	77
66	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	1.1	77
67	Analysis of human samples reveals impaired SHH-dependent cerebellar development in Joubert syndrome/Meckel syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 16951-16956.	3.3	77
68	Novel NEK8 Mutations Cause Severe Syndromic Renal Cystic Dysplasia through YAP Dysregulation. PLoS Genetics, 2016, 12, e1005894.	1.5	77
69	Familial CHARGE syndrome because of CHD7 mutation: clinical intraâ€•and interfamilial variability. Clinical Genetics, 2007, 72, 112-121.	1.0	76
70	Epistatic interactions with a common hypomorphicRET allele in syndromic Hirschsprung disease. Human Mutation, 2007, 28, 790-796.	1.1	75
71	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
72	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	1.8	71

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73	Phenotypic variability of Bardet-Biedl syndrome: focusing on the kidney. Pediatric Nephrology, 2012, 27, 7-15.	0.9	70
74	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	9.4	69
75	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	1.8	66
76	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
77	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	1.4	64
78	DUX4 and DUX4 downstream target genes are expressed in fetal FSHD muscles. Human Molecular Genetics, 2014, 23, 171-181.	1.4	61
79	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
80	Expression of theRET proto-oncogene in human Embryos. , 1998, 80, 481-486.		55
81	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
82	PMX2B , a new candidate gene for Hirschsprung's disease. Clinical Genetics, 2003, 64, 204-209.	1.0	53
83	High throughput SNP and expression analyses of candidate genes for non-syndromic oral clefts. Journal of Medical Genetics, 2005, 43, 598-608.	1.5	53
84	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
85	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	1.1	50
86	Expression of the SMADIP1 gene during early human development. Mechanisms of Development, 2002, 114, 187-191.	1.7	49
87	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
88	Pathophysiology of syndromic combined pituitary hormone deficiency due to a LHX3 defect in light of LHX3 and LHX4 expression during early human development. Gene Expression Patterns, 2004, 5, 279-284.	0.3	46
89	Antenatal spectrum of CHARGE syndrome in 40 fetuses withCHD7mutations. Journal of Medical Genetics, 2012, 49, 698-707.	1.5	45
90	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. Brain, 2019, 142, 35-49.	3.7	44

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91	Expression and mutation analysis of BRUNOL3, a candidate gene for heart and thymus developmental defects associated with partial monosomy 10p. Journal of Molecular Medicine, 2002, 80, 431-442.	1.7	42
92	l-Carnitine is Synthesized in the Human Fetalâ€“Placental Unit: Potential Roles in Placental and Fetal Metabolism. Placenta, 2006, 27, 841-846.	0.7	41
93	Monozygotic twins discordant for 18q21.2qter deletion detected byÂarray CGH in amniotic fluid. European Journal of Medical Genetics, 2013, 56, 502-505.	0.7	40
94	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	2.6	40
95	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	0.7	38
96	Clinical, genetic and neuropathological findings in a series of 138 fetuses with a corpus callosum malformation. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 36-46.	1.6	37
97	Gene expression in pharyngeal arch 1 during human embryonic development. Human Molecular Genetics, 2005, 14, 903-912.	1.4	35
98	The gene responsible for Dyggve-Melchior-Clausen syndrome encodes a novel peripheral membrane protein dynamically associated with the Golgi apparatus. Human Molecular Genetics, 2009, 18, 440-453.	1.4	34
99	Fatty acid oxidation in the human fetus: Implications for fetal and adult disease. Journal of Inherited Metabolic Disease, 2006, 29, 71-75.	1.7	33
100	Should PMM2-deficiency (CDG Ia) be searched in every case of unexplained hydrops fetalis?. Molecular Genetics and Metabolism, 2010, 101, 253-257.	0.5	32
101	Phenotypic spectrum of fetal Smithâ€“Lemliâ€“Opitz syndrome. European Journal of Medical Genetics, 2012, 55, 81-90.	0.7	32
102	OFD1mutations in males: phenotypic spectrum and ciliary basal body docking impairment. Clinical Genetics, 2013, 84, 86-90.	1.0	32
103	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.	1.5	32
104	Basal exon skipping and nonsense-associated altered splicing allows bypassing complete CEP290 loss-of-function in individuals with unusually mild retinal disease. Human Molecular Genetics, 2018, 27, 2689-2702.	1.4	31
105	Genotypeâ€“phenotype correlation in four 15q24 deleted patients identified by arrayâ€CGH. American Journal of Medical Genetics, Part A, 2009, 149A, 2813-2819.	0.7	30
106	PAX2 mutations in fetal renal hypodysplasia. American Journal of Medical Genetics, Part A, 2010, 152A, 830-835.	0.7	30
107	Homozygous truncating mutation of the KBP gene, encoding a KIF1B-binding protein, in a familial case of fetal polymicrogyria. Neurogenetics, 2013, 14, 215-224.	0.7	30
108	Bardetâ€Biedl syndrome: Antenatal presentation of fortyâ€five fetuses with biallelic pathogenic variants in known Bardetâ€Biedl syndrome genes. Clinical Genetics, 2019, 95, 384-397.	1.0	30

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109	NovelKIF7mutations extend the phenotypic spectrum of acrocallosal syndrome. Journal of Medical Genetics, 2012, 49, 713-720.	1.5	28
110	WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells. Brain, 2017, 140, 2597-2609.	3.7	28
111	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
112	ARID1Bmutations are the major genetic cause of corpus callosum anomalies in patients with intellectual disability. Brain, 2016, 139, e64-e64.	3.7	26
113	Regional and cellular specificity of the expression of TPRD, the tetratricopeptide Down syndrome gene, during human embryonic development. Mechanisms of Development, 2000, 93, 189-193.	1.7	25
114	BBS10 mutations are common in 'Meckel'-type cystic kidneys. Journal of Medical Genetics, 2010, 47, 848-852.	1.5	25
115	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. Journal of Medical Genetics, 2022, 59, 559-567.	1.5	25
116	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
117	De-novo mutations of the RET proto-oncogene in Hirschsprung's disease. Lancet, The, 1994, 344, 1769-1770.	6.3	24
118	Identification and Characterization of an Inner Ear-Expressed Human Melanoma Inhibitory Activity (MIA)-like Gene (MIAL) with a Frequent Polymorphism That Abolishes Translation. Genomics, 2001, 71, 40-52.	1.3	22
119	Novel mutations in the ciliopathy-associated gene CPLANE1 (C5orf42) cause OFD syndrome type VI rather than Joubert syndrome. European Journal of Medical Genetics, 2018, 61, 585-595.	0.7	22
120	C618R mutation in exon 10 of the RET proto-oncogene in a kindred with multiple endocrine neoplasia type 2A and Hirschsprung's disease. Journal of Clinical Endocrinology and Metabolism, 1996, 81, 2731-2733.	1.8	20
121	Posterior fossa imaging in 158Âchildren with ataxia. Journal of Neuroradiology, 2010, 37, 220-230.	0.6	20
122	Autosomal recessive <scp>IFT57</scp> hypomorphic mutation cause ciliary transport defect in unclassified oralâ€“facialâ€“digital syndrome with short stature and brachymesophalangia. Clinical Genetics, 2016, 90, 509-517.	1.0	20
123	A 7 bp deletion of the RET proto-oncogene in familial Hirschsprungâ€˜s disease. Human Molecular Genetics, 1994, 3, 1439-1440.	1.4	19
124	De novo trisomy 20p of paternal origin. American Journal of Medical Genetics, Part A, 2007, 143A, 1100-1103.	0.7	19
125	A clinical and histopathological study of malformations observed in fetuses infected by the Zika virus. Brain Pathology, 2019, 29, 114-125.	2.1	19
126	Refining the clinicopathological pattern of cerebral proliferative glomeruloid vasculopathy (Fowler) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	0.7	18

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127	Mutations of TSEN and CASK genes are prevalent in pontocerebellar hypoplasias type 2 and 4. Brain, 2012, 135, e199-e199.	3.7	18
128	Clinical and Functional Characterization of the Recurrent TUBA1A p.(Arg2His) Mutation. Brain Sciences, 2018, 8, 145.	1.1	18
129	Fetal megacystisâ€microcolon: Genetic mutational spectrum and identification of <scp>PDCL3</scp> as a novel candidate gene. Clinical Genetics, 2020, 98, 261-273.	1.0	18
130	Bi-allelic pathogenic variations in DNAJB11 cause Ivemark II syndrome, a renal-hepatic-pancreatic dysplasia. Kidney International, 2021, 99, 405-409.	2.6	18
131	Acyl-CoA dehydrogenase 9 (ACAD 9) is the long-chain acyl-CoA dehydrogenase in human embryonic and fetal brain. Biochemical and Biophysical Research Communications, 2006, 346, 33-37.	1.0	17
132	Giant diencephalic harmartoma and related anomalies: A newly recognized entity distinct from the Pallisterâ€“Hall syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1108-1115.	0.7	17
133	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. European Journal of Medical Genetics, 2013, 56, 301-308.	0.7	17
134	A French Approach to Test Fetuses with Ultrasound Abnormalities Using a Customized Microarray as First-Tier Genetic Test. Cytogenetic and Genome Research, 2015, 147, 103-110.	0.6	17
135	First fetal case of the 8q24.3 contiguous genes syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 239-242.	0.7	17
136	TAR syndrome: Clinical and molecular characterization of a cohort of 26 patients and description of novel noncoding variants of RBM8A. Human Mutation, 2020, 41, 1220-1225.	1.1	17
137	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
138	Novel de novo ZBTB20 mutations in three cases with Primrose syndrome and constant corpus callosum anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 1091-1098.	0.7	16
139	Loss of function IFT27 variants associated with an unclassified lethal fetal ciliopathy with renal agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 1610-1613.	0.7	16
140	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
141	12q21 Microdeletion in a fetus with Meckel syndrome involving CEP290/MKS4. European Journal of Medical Genetics, 2013, 56, 580-583.	0.7	15
142	SOX3 duplication: A genetic cause to investigate in fetuses with neural tube defects. Prenatal Diagnosis, 2019, 39, 1026-1034.	1.1	15
143	Altered GLI3 and FGF8 signaling underlies acrocallosal syndrome phenotypes inKif7depleted mice. Human Molecular Genetics, 2019, 28, 877-887.	1.4	15
144	Severe and progressive neuronal loss in myelomeningocele begins before 16 weeks of pregnancy. American Journal of Obstetrics and Gynecology, 2020, 223, 256.e1-256.e9.	0.7	15

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145	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guionâ€Almeida type. Human Mutation, 2020, 41, 1372-1382.	1.1	15
146	Diagnostic strategy in segmentation defect of the vertebrae: a retrospective study of 73 patients. Journal of Medical Genetics, 2018, 55, 422.2-429.	1.5	14
147	PAK3 mutations responsible for severe intellectual disability and callosal agenesis inhibit cell migration. Neurobiology of Disease, 2020, 136, 104709.	2.1	14
148	Molecular characterisation of a prenatally diagnosed 5q15q21.3 deletion and review of the literature. Prenatal Diagnosis, 2006, 26, 231-238.	1.1	13
149	Genotype phenotype correlation of 30 patients with Smith-Magenis syndrome (SMS) using comparative genome hybridisation array: cleft palate in SMS is associated with larger deletions. Journal of Medical Genetics, 2007, 44, 537-540.	1.5	13
150	A novel polymorphism in the coding sequence of the human RET proto-oncogene. Human Genetics, 1994, 94, 579-80.	1.8	12
151	Prenatal diagnosis andÂmolecular characterization ofÂanÂinterstitial 1q24.2q25.2Âdeletion. European Journal of Medical Genetics, 2006, 49, 487-493.	0.7	12
152	Matthew-Wood syndrome: Report of two new cases supporting autosomal recessive inheritance and exclusion ofFGF10 andFGFR2. American Journal of Medical Genetics, Part A, 2007, 143A, 219-228.	0.7	12
153	Screening of MITF and SOX10 Regulatory Regions in Waardenburg Syndrome Type 2. PLoS ONE, 2012, 7, e41927.	1.1	12
154	Targeted nextâ€generation sequencing in a large series of fetuses with severe renal diseases. Human Mutation, 2022, 43, 347-361.	1.1	12
155	Prenatal diagnosis of carnitine palmitoyltransferase 2 deficiency in chorionic villi: a novel approach. Prenatal Diagnosis, 2003, 23, 884-887.	1.1	11
156	Whole exome sequencing diagnoses the first fetal case of <scp>B</scp>ainbridgeâ€<scp>R</scp>opers syndrome presenting as pontocerebellar hypoplasia type 1. Birth Defects Research, 2018, 110, 538-542.	0.8	10
157	Molecular screening of theZFHX1B gene in prenatally diagnosed isolated agenesis of thecorpus callosum. Prenatal Diagnosis, 2004, 24, 298-301.	1.1	9
158	A practical approach to the examination of the malformed fetal brain: impact on genetic counselling. Pathology, 2008, 40, 180-187.	0.3	9
159	First evidence of <scp>SOX2</scp> mutations in Peters' anomaly: Lessons from molecular screening of 95 patients. Clinical Genetics, 2022, 101, 494-506.	1.0	9
160	A homozygous AHI1 gene mutation (p.Thr304AsnfsX6) in a consanguineous Moroccan family with Joubert syndrome: a case report. Journal of Medical Case Reports, 2015, 9, 254.	0.4	8
161	Whole-exome sequence analysis highlights the role of unmasked recessive mutations in copy number variants with incomplete penetrance. European Journal of Human Genetics, 2018, 26, 912-918.	1.4	8
162	A case of mild CHARGE syndrome associated with a splice site mutation in CHD7. European Journal of Medical Genetics, 2016, 59, 195-197.	0.7	7

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163	A neuropathological study of novel RTTN gene mutations causing a familial microcephaly with simplified gyral pattern. Birth Defects Research, 2018, 110, 598-602.	0.8	7
164	CHARGE syndrome: a recurrent hotspot of mutations in CHD7 IVS25 analyzed by bioinformatic tools and minigene assays. European Journal of Human Genetics, 2018, 26, 287-292.	1.4	7
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