Tania Attie-Bitach

List of Publications by Year
in descending order

196 papers	14,805 citations	12330 69 h-index	20961 115 g-index
209 all docs	209 docs citations	209 times ranked	15272 citing authors

#	Article	IF	Citations
1	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. Journal of Medical Genetics, 2022, 59, 559-567.	3.2	25
2	Targeted nextâ€generation sequencing in a large series of fetuses with severe renal diseases. Human Mutation, 2022, 43, 347-361.	2.5	12
3	First evidence of <scp>SOX2</scp> mutations in Peters' anomaly: Lessons from molecular screening of 95 patients. Clinical Genetics, 2022, 101, 494-506.	2.0	9
4	Extending the prenatal Noonan's phenotype by review of ultrasound and autopsy data. Prenatal Diagnosis, 2022, 42, 574-582.	2.3	4
5	Discovery of a genetic module essential for assigning leftâ€“right asymmetry in humans and ancestral vertebrates. Nature Genetics, 2022, 54, 62-72.	21.4	16
6	Biallelic <scp>THOC6</scp> pathogenic variants: Prenatal phenotype and review of the literature. Birth Defects Research, 2022, 114, 499-504.	1.5	5
7	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
8	Bi-allelic pathogenic variations in DNAJB11 cause Ivemark II syndrome, a renal-hepatic-pancreatic dysplasia. Kidney International, 2021, 99, 405-409.	5.2	18
9	Novel <scp>CDK10</scp> variants with multicystic dysplastic kidney, left ventricular nonâ€compaction, and a solitary median maxillary central incisor. Clinical Genetics, 2021, 100, 348-349.	2.0	2
10	Human neuropathology confirms projection neuron and interneuron defects and delayed oligodendrocyte production and maturation in FOXG1 syndrome. European Journal of Medical Genetics, 2021, 64, 104282.	1.3	4
11	The first two <scp>nonâ€Finnish HYLS1</scp> variants: Expanding the phenotypic spectrum of hydrolethalus syndrome. Clinical Genetics, 2021, 100, 462-467.	2.0	0
12	Homozygous GLI3 variants observed in three unrelated patients presenting with syndromic polydactyly. American Journal of Medical Genetics, Part A, 2021, 185, 3831-3837.	1.2	0
13	Prenatalâ€onset of congenital neuronal ceroid lipofuscinosis with a novel CTSD mutation. Birth Defects Research, 2021, 113, 1324-1332.	1.5	1
14	TALPID3/KIAA0586 Regulates Multiple Aspects of Neuromuscular Patterning During Gastrointestinal Development in Animal Models and Human. Frontiers in Molecular Neuroscience, 2021, 14, 757646.	2.9	3
15	PAK3 mutations responsible for severe intellectual disability and callosal agenesis inhibit cell migration. Neurobiology of Disease, 2020, 136, 104709.	4.4	14
16	Hydrothorax in fetal cases of Opitz G/<scp>BBB</scp> diagnosis: Extending the phenotype?. Clinical Genetics, 2020, 98, 620-621.	2.0	1
17	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.	1.3	137
18	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.	2.7	4

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19	Fetal megacystisâ€microcolon: Genetic mutational spectrum and identification of <scp>PDCL3</scp> as a novel candidate gene. Clinical Genetics, 2020, 98, 261-273.	2.0	18
20	Prenatal diagnosis of cerebroâ€oculoâ€facioâ€skeletal syndrome: Report of three fetuses and review of the literature. American Journal of Medical Genetics, Part A, 2020, 182, 1236-1242.	1.2	4
21	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.	2.5	17
22	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.	1.3	15
23	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guionâ€Almeida type. Human Mutation, 2020, 41, 1372-1382.	2.5	15
24	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.	6.2	25
25	Functional Vision Analysis in Patients With CHARGE Syndrome. Journal of Pediatric Ophthalmology and Strabismus, 2020, 57, 120-128.	0.7	6
26	A clinical and histopathological study of malformations observed in fetuses infected by the Zika virus. Brain Pathology, 2019, 29, 114-125.	4.1	19
27	SOX3 duplication: A genetic cause to investigate in fetuses with neural tube defects. Prenatal Diagnosis, 2019, 39, 1026-1034.	2.3	15
28	Significant contribution of intragenic deletions to ARID1B mutation spectrum. Genetics in Medicine, 2019, 21, 2654-2655.	2.4	3
29	Fetal phenotype of Rubinsteinâ€Taybi syndrome caused by CREBBP mutations. Clinical Genetics, 2019, 95, 420-426.	2.0	7
30	Mutations in IFT80 cause SRPS Type IV. Report of two families and review. American Journal of Medical Genetics, Part A, 2019, 179, 639-644.	1.2	6
31	Altered GLI3 and FGF8 signaling underlies acrocallosal syndrome phenotypes inKif7depleted mice. Human Molecular Genetics, 2019, 28, 877-887.	2.9	15
32	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.	2.0	30
33	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. Brain, 2019, 142, 35-49.	7.6	44
34	Diagnostic strategy in segmentation defect of the vertebrae: a retrospective study of 73 patients. Journal of Medical Genetics, 2018, 55, 422.2-429.	3.2	14
35	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.	2.8	8
36	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.	1.2	16

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37	A neuropathological study of novel RTTN gene mutations causing a familial microcephaly with simplified gyral pattern. Birth Defects Research, 2018, 110, 598-602.	1.5	7
38	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.	2.8	7
39	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.	1.5	10
40	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.	1.2	16
41	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.	1.3	22
42	In utero ultrasound diagnosis of corpus callosum agenesis leading to the identification of orofaciodigital type 1 syndrome in female fetuses. Birth Defects Research, 2018, 110, 382-389.	1.5	7
43	Corpus Callosum Abnormalities and Short Femurs in Beckwithâ€“Wiedemann Syndrome: A Report of Two Fetal Cases. Fetal and Pediatric Pathology, 2018, 37, 411-417.	0.7	2
44	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	6.2	40
45	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.	2.9	31
46	Clinical and Functional Characterization of the Recurrent TUBA1A p.(Arg2His) Mutation. Brain Sciences, 2018, 8, 145.	2.3	18
47	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	2.4	90
48	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	21.4	69
49	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	3.8	66
50	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	3.2	85
51	WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells. Brain, 2017, 140, 2597-2609.	7.6	28
52	Fetal Cerebral Ventricular Dilatation: Etiopathogenic Study of 130 Observations. Birth Defects Research, 2017, 109, 1586-1595.	1.5	2
53	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.	1.6	65
54	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

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55	First fetal case of the 8q24.3 contiguous genes syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 239-242.	1.2	17
56	ARID1Bmutations are the major genetic cause of corpus callosum anomalies in patients with intellectual disability. Brain, 2016, 139, e64-e64.	7.6	26
57	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.	2.0	20
58	A case of mild CHARGE syndrome associated with a splice site mutation in CHD7. European Journal of Medical Genetics, 2016, 59, 195-197.	1.3	7
59	TMEM107 recruits ciliopathy proteins to subdomains of the ciliary transition zone and causes JoubertÂsyndrome. Nature Cell Biology, 2016, 18, 122-131.	10.3	118
60	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	2.4	125
61	Novel NEK8 Mutations Cause Severe Syndromic Renal Cystic Dysplasia through YAP Dysregulation. PLoS Genetics, 2016, 12, e1005894.	3.5	77
62	Mutations of IFT81, encoding an IFT-B core protein, as a rare cause of a ciliopathy. Cilia, 2015, 4, .	1.8	0
63	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.	0.8	8
64	A study of new NEK8 mutations in patients with severe renal cystic hypodysplasia and ciliopathy-associated defects. Cilia, 2015, 4, .	1.8	0
65	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.	1.1	17
66	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.	6.2	82
67	A novel KIF7 mutation in two affected siblings with acrocallosal syndrome. Clinical Dysmorphology, 2015, 24, 61-64.	0.3	6
68	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	5.2	95
69	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.	3.2	32
70	New insights into genotypeâ€“phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	2.8	97
71	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.	2.8	48
72	Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. Human Molecular Genetics, 2014, 23, 2279-2289.	2.9	98

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73	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	2.5	50
74	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	2.5	113
75	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	3.8	71
76	The oral-facial-digital syndrome gene C2CD3 encodes a positive regulator of centriole elongation. Nature Genetics, 2014, 46, 905-911.	21.4	121
77	DUX4 and DUX4 downstream target genes are expressed in fetal FSHD muscles. Human Molecular Genetics, 2014, 23, 171-181.	2.9	61
78	Inappropriate p53 activation during development induces features of CHARGE syndrome. Nature, 2014, 514, 228-232.	27.8	117
79	Monozygotic twins discordant for 18q21.2qter deletion detected byÂarray CGH in amniotic fluid. European Journal of Medical Genetics, 2013, 56, 502-505.	1.3	40
80	Homozygous truncating mutation of the KBP gene, encoding a KIF1B-binding protein, in a familial case of fetal polymicrogyria. Neurogenetics, 2013, 14, 215-224.	1.4	30
81	OFD1mutations in males: phenotypic spectrum and ciliary basal body docking impairment. Clinical Genetics, 2013, 84, 86-90.	2.0	32
82	12q21 Microdeletion in a fetus with Meckel syndrome involving CEP290/MKS4. European Journal of Medical Genetics, 2013, 56, 580-583.	1.3	15
83	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. European Journal of Medical Genetics, 2013, 56, 301-308.	1.3	17
84	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.	4.5	87
85	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
86	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.	1.2	16
87	Antenatal spectrum of CHARGE syndrome in 40 fetuses withCHD7mutations. Journal of Medical Genetics, 2012, 49, 698-707.	3.2	45
88	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84
89	Cobblestone lissencephaly: neuropathological subtypes and correlations with genes of dystroglycanopathies. Brain, 2012, 135, 469-482.	7.6	151
90	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.	7.1	77

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91	EFTUD2haploinsufficiency leads to syndromic oesophageal atresia. Journal of Medical Genetics, 2012, 49, 737-746.	3.2	89
92	Mutations of TSEN and CASK genes are prevalent in pontocerebellar hypoplasias type 2 and 4. Brain, 2012, 135, e199-e199.	7.6	18
93	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	21.4	157
94	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	30.7	103
95	Phenotypic spectrum of fetal Smithâ€“Lemliâ€“Opitz syndrome. European Journal of Medical Genetics, 2012, 55, 81-90.	1.3	32
96	TCTN3 Mutations Cause Mohr-Majewski Syndrome. American Journal of Human Genetics, 2012, 91, 372-378.	6.2	123
97	Identification of Mutations in TMEM5 and ISPD as a Cause of Severe Cobblestone Lissencephaly. American Journal of Human Genetics, 2012, 91, 1135-1143.	6.2	126
98	NovelKIF7mutations extend the phenotypic spectrum of acrocallosal syndrome. Journal of Medical Genetics, 2012, 49, 713-720.	3.2	28
99	Phenotypic variability of Bardet-Biedl syndrome: focusing on the kidney. Pediatric Nephrology, 2012, 27, 7-15.	1.7	70
100	Screening of MITF and SOX10 Regulatory Regions in Waardenburg Syndrome Type 2. PLoS ONE, 2012, 7, e41927.	2.5	12
101	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.	3.2	60
102	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326
103	KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	21.4	203
104	Disruption of a Ciliary B9 Protein Complex Causes Meckel Syndrome. American Journal of Human Genetics, 2011, 89, 94-110.	6.2	136
105	Disruption of a Ciliary B9 Protein Complex Causes Meckel Syndrome. American Journal of Human Genetics, 2011, 89, 589.	6.2	2
106	Cerebral dysgenesis does not exclude OFD I syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 455-457.	1.2	4
107	Macrostomia, thin upper vermilion border, long philtrum, broad halluces, and intellectual disability in two sibs. American Journal of Medical Genetics, Part A, 2011, 155, 2465-2468.	1.2	2
108	GLI3 is rarely implicated in OFD syndromes with midline abnormalities. Human Mutation, 2011, 32, 1332-1333.	2.5	5

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109	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
110	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.	2.5	27
111	PAX2 mutations in fetal renal hypodysplasia. American Journal of Medical Genetics, Part A, 2010, 152A, 830-835.	1.2	30
112	New ocular phenotype associated with a mutation in the PAX2 gene. Eye, 2010, 24, 1293-1294.	2.1	2
113	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
114	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.	2.9	231
115	BBS10 mutations are common in 'Meckel'-type cystic kidneys. Journal of Medical Genetics, 2010, 47, 848-852.	3.2	25
116	Planar Cell Polarity Acts Through Septins to Control Collective Cell Movement and Ciliogenesis. Science, 2010, 329, 1337-1340.	12.6	309
117	Pitchfork Regulates Primary Cilia Disassembly and Left-Right Asymmetry. Developmental Cell, 2010, 19, 66-77.	7.0	133
118	Should PMM2-deficiency (CDG Ia) be searched in every case of unexplained hydrops fetalis?. Molecular Genetics and Metabolism, 2010, 101, 253-257.	1.1	32
119	Posterior fossa imaging in 158Âchildren with ataxia. Journal of Neuroradiology, 2010, 37, 220-230.	1.1	20
120	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.	7.1	51
121	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.	2.9	34
122	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, 1714-1716.	2.9	0
123	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.	1.2	17
124	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
125	Genotypeâ€“phenotype correlation in four 15q24 deleted patients identified by arrayâ€CGH. American Journal of Medical Genetics, Part A, 2009, 149A, 2813-2819.	1.2	30
126	Phenotypic spectrum ofSTRA6mutations: from Matthew-Wood syndrome to non-lethal anophthalmia. Human Mutation, 2009, 30, E673-E681.	2.5	89

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127	CC2D2Amutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. Human Mutation, 2009, 30, 1574-1582.	2.5	80
128	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	21.4	255
129	A 24-Mb deletion in 14q in a girl with corpus callosum hypoplasia. European Journal of Medical Genetics, 2009, 52, 256-259.	1.3	1
130	Refining the clinicopathological pattern of cerebral proliferative glomeruloid vasculopathy (Fowler) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.3	18
131	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	6.2	352
132	A practical approach to the examination of the malformed fetal brain: impact on genetic counselling. Pathology, 2008, 40, 180-187.	0.6	9
133	The Meckelâ€“Gruber Syndrome proteins MKS1 and meckelin interact and are required for primary cilium formation. Human Molecular Genetics, 2007, 16, 173-186.	2.9	245
134	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.	3.2	13
135	The Meckel-Gruber Syndrome Gene, MKS3, Is Mutated in Joubert Syndrome. American Journal of Human Genetics, 2007, 80, 186-194.	6.2	217
136	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.	6.2	174
137	Pleiotropic Effects of CEP290 (NPHP6) Mutations Extend to Meckel Syndrome. American Journal of Human Genetics, 2007, 81, 170-179.	6.2	248
138	Deletions at the SOX10 Gene Locus Cause Waardenburg Syndrome Types 2 and 4. American Journal of Human Genetics, 2007, 81, 1169-1185.	6.2	216
139	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.	1.2	12
140	De novo trisomy 20p of paternal origin. American Journal of Medical Genetics, Part A, 2007, 143A, 1100-1103.	1.2	19
141	Epistatic interactions with a common hypomorphicRET allele in syndromic Hirschsprung disease. Human Mutation, 2007, 28, 790-796.	2.5	75
142	Spectrum ofMKS1andMKS3mutations in Meckel syndrome: a genotype-phenotype correlation. Human Mutation, 2007, 28, 523-524.	2.5	92
143	Homozygous silencing of T-box transcription factor EOMES leads to microcephaly with polymicrogyria and corpus callosum agenesis. Nature Genetics, 2007, 39, 454-456.	21.4	181
144	The ciliary gene RPGRIP1L is mutated in cerebello-oculo-renal syndrome (Joubert syndrome type B) and Meckel syndrome. Nature Genetics, 2007, 39, 875-881.	21.4	442

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145	Familial CHARGE syndrome because of CHD7 mutation: clinical intraâ€•and interfamilial variability. Clinical Genetics, 2007, 72, 112-121.	2.0	76
146	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.	2.1	17
147	Prenatal diagnosis andÂmolecular characterization ofÂanÂinterstitial 1q24.2q25.2Âdeletion. European Journal of Medical Genetics, 2006, 49, 487-493.	1.3	12
148	Molecular characterisation of a prenatally diagnosed 5q15q21.3 deletion and review of the literature. Prenatal Diagnosis, 2006, 26, 231-238.	2.3	13
149	Cytogenetic and histological features of a human embryo with homogeneous chromosome 8 trisomy. Prenatal Diagnosis, 2006, 26, 1201-1205.	2.3	5
150	The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome and the wpk rat. Nature Genetics, 2006, 38, 191-196.	21.4	266
151	Fatty acid oxidation in the human fetus: Implications for fetal and adult disease. Journal of Inherited Metabolic Disease, 2006, 29, 71-75.	3.6	33
152	l-Carnitine is Synthesized in the Human Fetalâ€“Placental Unit: Potential Roles in Placental and Fetal Metabolism. Placenta, 2006, 27, 841-846.	1.5	41
153	Unusual phenotype with progressive vertebral fusion in a girl with an apparently balanced t(10;20)(p11;p13) translocation. , 2005, 134A, 39-44.		4
154	Cleft lip/palate and CDH1/E-cadherin mutations in families with hereditary diffuse gastric cancer. Journal of Medical Genetics, 2005, 43, 138-142.	3.2	161
155	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.	3.6	195
156	Gene expression in pharyngeal arch 1 during human embryonic development. Human Molecular Genetics, 2005, 14, 903-912.	2.9	35
157	Long-Chain Fatty Acid Oxidation during Early Human Development. Pediatric Research, 2005, 57, 755-759.	2.3	86
158	High throughput SNP and expression analyses of candidate genes for non-syndromic oral clefts. Journal of Medical Genetics, 2005, 43, 598-608.	3.2	53
159	Impaired Mitochondrial Glutamate Transport in Autosomal Recessive Neonatal Myoclonic Epilepsy. American Journal of Human Genetics, 2005, 76, 334-339.	6.2	149
160	Antenatal Presentation of Bardet-Biedl Syndrome May Mimic Meckel Syndrome. American Journal of Human Genetics, 2005, 76, 493-504.	6.2	120
161	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.	3.2	199
162	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.	0.8	46

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163	Molecular screening of theZFHX1B gene in prenatally diagnosed isolated agenesis of thecorpus callosum. Prenatal Diagnosis, 2004, 24, 298-301.	2.3	9
164	PMX2B , a new candidate gene for Hirschsprung's disease. Clinical Genetics, 2003, 64, 204-209.	2.0	53
165	Prenatal diagnosis of carnitine palmitoyltransferase 2 deficiency in chorionic villi: a novel approach. Prenatal Diagnosis, 2003, 23, 884-887.	2.3	11
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