Jill Clayton-Smith

List of Publications by Year
in descending order

160	11,851	31949 53	30894 102
papers	citations	h-index	g-index

169	169	169	16789
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Personalised virtual gene panels reduce interpretation workload and maintain diagnostic rates of proband-only clinical exome sequencing for rare disorders. Journal of Medical Genetics, 2022, 59, 393-398.	1.5	14
2	A standard of care for individuals with <scp>PIK3CA</scp>â€related disorders: An international expert consensus statement. Clinical Genetics, 2022, 101, 32-47.	1.0	21
3	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	1.1	14
4	Further delineation of phenotypic spectrum of <scp>SCN2A</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 867-877.	0.7	3
5	Co-designing models for the communication of genomic results for rare diseases: a comparative study in the Czech Republic and the United Kingdom. Journal of Community Genetics, 2022, 13, 313-327.	0.5	3
6	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	1.4	48
7	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	3.7	35
8	Recurrent <scp>KCNT2</scp> missense variants affecting p.Arg190 result in a recognizable phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3083-3091.	0.7	7
9	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
10	The adaptive functioning profile of Pitt-Hopkins syndrome. European Journal of Medical Genetics, 2021, 64, 104279.	0.7	3
11	The diagnostic utility of clinical exome sequencing in 60 patients with hearing loss disorders: A singleâ€institution experience. Clinical Otolaryngology, 2021, 46, 1257-1262.	0.6	3
12	Prevalence of fetal alcohol spectrum disorder in Greater Manchester, UK: An active case ascertainment study. Alcoholism: Clinical and Experimental Research, 2021, 45, 2271-2281.	1.4	21
13	Identification of LAMA1 mutations ends diagnostic odyssey and has prognostic implications for patients with presumed Joubert syndrome. Brain Communications, 2021, 3, fcab163.	1.5	8
14	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607.	1.6	37
15	Congenital cataracts in females caused by BCOR mutations; report of six further families demonstrating clinical variability and diverse genetic mechanisms. European Journal of Medical Genetics, 2020, 63, 103658.	0.7	6
16	Exome sequencing in patients with antiepileptic drug exposure and complex phenotypes. Archives of Disease in Childhood, 2020, 105, 384-389.	1.0	3
17	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	1.1	53
18	Clinical utility of genetic testing in 201 preschool children with inherited eye disorders. Genetics in Medicine, 2020, 22, 745-751.	1.1	42

#	Article	IF	Citations
19	Ligase IV syndrome can present with microcephaly and radial ray anomalies similar to Fanconi anaemia plus fatal kidney malformations. European Journal of Medical Genetics, 2020, 63, 103974.	0.7	5
20	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
21	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	1.5	11
22	Mowat-Wilson syndrome: growth charts. Orphanet Journal of Rare Diseases, 2020, 15, 151.	1.2	12
23	Oral-Facial-Digital Syndrome Type 1: Further Clinical and Molecular Delineation in 2 New Families. Cleft Palate-Craniofacial Journal, 2020, 57, 606-615.	0.5	1
24	A new mutational hotspot in the SKI gene in the context of MFS/TAA molecular diagnosis. Human Genetics, 2020, 139, 461-472.	1.8	8
25	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. Orphanet Journal of Rare Diseases, 2020, 15, 103.	1.2	23
26	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
27	Update of the EMQN/ACGS best practice guidelines for molecular analysis of Prader-Willi and Angelman syndromes. European Journal of Human Genetics, 2019, 27, 1326-1340.	1.4	41
28	Diagnosis and management of individuals with Fetal Valproate Spectrum Disorder; a consensus statement from the European Reference Network for Congenital Malformations and Intellectual Disability. Orphanet Journal of Rare Diseases, 2019, 14, 180.	1.2	33
29	The phenotype of Sotos syndrome in adulthood: A review of 44 individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 502-508.	0.7	31
30	Deep phenotyping of 14 new patients with IQSEC2 variants, including monozygotic twins of discordant phenotype. Clinical Genetics, 2019, 95, 496-506.	1.0	20
31	Diagnosis and management in PittâЄHopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	1.0	63
32	Delivering effective genetic services for patients and families affected by cleft lip and/or palate. European Journal of Human Genetics, 2019, 27, 1018-1025.	1.4	6
33	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
34	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
35	Genotypeâ€“phenotype specificity in Menkeâ€“Hennekam syndrome caused by missense variants in exon 30 or 31 of CREBBP. American Journal of Medical Genetics, Part A, 2019, 179, 1058-1062.	0.7	23
36	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	2.6	45

#	Article	IF	Citations
37	Clinical and genetic variability in children with partial albinism. Scientific Reports, 2019, 9, 16576.	1.6	26
38	Traboulsi syndrome due to ASPH mutation: an under-recognised cause of ectopia lentis. Clinical Dysmorphology, 2019, 28, 184-189.	0.1	10
39	Timing Of Primary Surgery for cleft palate (TOPS): protocol for a randomised trial of palate surgery at 6 months versus 12 months of age. BMJ Open, 2019, 9, e029780.	0.8	37
40	Clinical and genetic heterogeneity in Melkersson-Rosenthal Syndrome. European Journal of Medical Genetics, 2019, 62, 103536.	0.7	8
41	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	2.4	70
42	Fetal antiepileptic drug exposure and learning and memory functioning at 6â€¯years of age: The NEAD prospective observational study. Epilepsy and Behavior, 2019, 92, 154-164.	0.9	30
43	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	1.1	80
44	Intellectual functioning in clinically confirmed fetal valproate syndrome. Neurotoxicology and Teratology, 2019, 71, 16-21.	1.2	18
45	Heterozygous loss-of-function variants of MEIS2 cause a triad of palatal defects, congenital heart defects, and intellectual disability. European Journal of Human Genetics, 2019, 27, 278-290.	1.4	30
46	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
47	Observation of Cleft Palate in an Individual with SOX11 Mutation. Cleft Palate-Craniofacial Journal, 2018, 55, 456-461.	0.5	15
48	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479.	2.6	63
49	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204
50	CNVs affecting cancer predisposing genes (CPGs) detected as incidental findings in routine germline diagnostic chromosomal microarray (CMA) testing. Journal of Medical Genetics, 2018, 55, 89-96.	1.5	7
51	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Journal of Medical Genetics, 2018, 55, 104-113.	1.5	59
52	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	2.6	37
53	Monotherapy treatment of epilepsy in pregnancy: congenital malformation outcomes in the child. The Cochrane Library, 2017, 2017, CD010224.	1.5	135
54	Structural analysis of pathogenic mutations in theDYRK1Agene in patients with developmental disorders. Human Molecular Genetics, 2017, 26, ddw409.	1.4	33

#	Article	IF	Citations
55	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	1.1	46
56	Interrupted/bipartite clavicle as a diagnostic clue in Kabuki syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1115-1118.	0.7	3
57	Genitourinary malformations: an under-recognized feature of ectrodactyly, ectodermal dysplasia and cleft lip/palate syndrome. Clinical Dysmorphology, 2017, 26, 78-82.	0.1	7
58	Phenotypic Heterogeneity in a Congenital Disorder of Glycosylation Caused by Mutations in STT3A. Journal of Child Neurology, 2017, 32, 560-565.	0.7	10
59	Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339.	1.4	43
60	Panel-Based Clinical Genetic Testing in 85 Children with Inherited Retinal Disease. Ophthalmology, 2017, 124, 985-991.	2.5	51
61	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. Genetics in Medicine, 2017, 19, 701-710.	1.1	13
62	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	2.6	119
63	Confirmation that mutations in DDX59 cause an autosomal recessive form of oral-facial-digital syndrome: Further delineation of the DDX59 phenotype in two new families. European Journal of Medical Genetics, 2017, 60, 527-532.	0.7	7
64	Further Clinical Delineation of the MEF2C Haploinsufficiency Syndrome: Report on New Cases and Literature Review of Severe Neurodevelopmental Disorders Presenting with Seizures, Absent Speech, and Involuntary Movements. Journal of Pediatric Genetics, 2017, 06, 129-141.	0.3	38
65	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	2.6	83
66	Protein structure and phenotypic analysis of pathogenic and population missense variants inSTXBP1. Molecular Genetics & Genomic Medicine, 2017, 5, 495-507.	0.6	29
67	Novel PEX11B Mutations Extend the Peroxisome Biogenesis Disorder 14B Phenotypic Spectrum and Underscore Congenital Cataract as an Early Feature. , 2017, 58, 594.		25
68	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	1.1	54
69	In utero exposure to valproate increases the risk of isolated cleft palate. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2016, 101, F207-F211.	1.4	30
70	Mutations in the HECT domain of NEDD4L lead to AKTâ€“mTOR pathway deregulation and cause periventricular nodular heterotopia. Nature Genetics, 2016, 48, 1349-1358.	9.4	101
71	Cognition in school-age children exposed to levetiracetam, topiramate, or sodium valproate. Neurology, 2016, 87, 1943-1953.	1.5	98
72	A Recurrent Mosaic Mutation in SMO , Encoding the Hedgehog Signal Transducer Smoothened, Is the Major Cause of Curry-Jones Syndrome. American Journal of Human Genetics, 2016, 98, 1256-1265.	2.6	70

#	Article	IF	Citations
73	Exploring the genetic basis of 3MC syndrome: Findings in 12 further families. American Journal of Medical Genetics, Part A, 2016, 170, 1216-1224.	0.7	25
74	Detection of a mosaic PIK3CA mutation in dental DNA from a child with megalencephaly capillary malformation syndrome. Clinical Dysmorphology, 2016, 25, 16-18.	0.1	8
75	Next-generation Sequencing in the Diagnosis of Metabolic Disease Marked by Pediatric Cataract. Ophthalmology, 2016, 123, 217-220.	2.5	44
76	Rutherfurd syndrome revisited. Clinical Dysmorphology, 2015, 24, 125-127.	0.1	5
77	Abrogation of HMX1 Function Causes Rare Oculoauricular Syndrome Associated With Congenital Cataract, Anterior Segment Dysgenesis, and Retinal Dystrophy. Investigative Ophthalmology and Visual Science, 2015, 56, 883-891.	3.3	22
78	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. European Journal of Medical Genetics, 2015, 58, 455-465.	0.7	83
79	IQ at 6 years after in utero exposure to antiepileptic drugs. Neurology, 2015, 84, 382-390.	1.5	226
80	Mutations inSIPA1L3cause eye defects through disruption of cell polarity and cytoskeleton organization. Human Molecular Genetics, 2015, 24, 5789-5804.	1.4	32
81	Agnathia-otocephaly complex and asymmetric velopharyngeal insufficiency due to an in-frame duplication in OTX2. Journal of Human Genetics, 2015, 60, 199-202.	1.1	25
82	A recurrent synonymous KAT6B mutation causes Sayâ€Barberâ€Biesecker/Youngâ€Simpson syndrome by inducing aberrant splicing. American Journal of Medical Genetics, Part A, 2015, 167, 3006-3010.	0.7	17
83	Deletion of 19q13 reveals clinical overlap with Dubowitz syndrome. Journal of Human Genetics, 2015, 60, 781-785.	1.1	12
84	Interchromosomal Insertional Translocation at Xq26.3 AltersSOX3Expression in an Individual With XX Male Sex Reversal. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E815-E820.	1.8	46
85	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	1.4	56
86	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	1.5	141
87	Velopharyngeal insufficiency: high detection rate of genetic abnormalities if associated with additional features. Archives of Disease in Childhood, 2014, 99, 52-57.	1.0	6
88	Compound Heterozygosity of Low-Frequency Promoter Deletions and Rare Loss-of-Function Mutations in TXNL4A Causes Burn-McKeown Syndrome. American Journal of Human Genetics, 2014, 95, 698-707.	2.6	55
89	Breastfeeding in Children of Women Taking Antiepileptic Drugs. JAMA Pediatrics, 2014, 168, 729.	3.3	201
90	Ocular coloboma and foetal valproate syndrome. Clinical Dysmorphology, 2014, 23, 74-75.	0.1	7

#	Article	IF	Citations
91	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. Human Molecular Genetics, 2014, 23, 2752-2768.	1.4	140
92	Mutations in CKAP2L, the Human Homolog of the Mouse Radmis Gene, Cause Filippi Syndrome. American Journal of Human Genetics, 2014, 95, 622-632.	2.6	34
93	Personalized Diagnosis and Management of Congenital Cataract by Next-Generation Sequencing. Ophthalmology, 2014, 121, 2124-2137.e2.	2.5	153
94	A novel method for detecting uniparental disomy from trio genotypes identifies a significant excess in children with developmental disorders. Genome Research, 2014, 24, 673-687.	2.4	53
95	Oculo-auriculo-vertebral spectrum: a review of the literature and genetic update. Journal of Medical Genetics, 2014, 51, 635-645.	1.5	140
96	Trisomy 18 mosaicism: report of two cases. World Journal of Pediatrics, 2013, 9, 179-181.	0.8	15
97	Perrault Syndrome Is Caused by Recessive Mutations in CLPP, Encoding a Mitochondrial ATP-Dependent Chambered Protease. American Journal of Human Genetics, 2013, 92, 605-613.	2.6	186
98	Fetal antiepileptic drug exposure: Adaptive and emotional/behavioral functioning at age 6years. Epilepsy and Behavior, 2013, 29, 308-315.	0.9	132
99	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	1.1	178
100	Fetal antiepileptic drug exposure and cognitive outcomes at age 6 years (NEAD study): a prospective observational study. Lancet Neurology, The, 2013, 12, 244-252.	4.9	665
101	ALDH1A3 Mutations Cause Recessive Anophthalmia and Microphthalmia. American Journal of Human Genetics, 2013, 92, 265-270.	2.6	92
102	The prevalence of neurodevelopmental disorders in children prenatally exposed to antiepileptic drugs. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 637-643.	0.9	280
103	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	3.7	85
104	12p13.33 microdeletion including ELKS/ERC1, a new locus associated with childhood apraxia of speech. European Journal of Human Genetics, 2013, 21, 82-88.	1.4	70
105	FETAL EFFECTS OF SELECTIVE SEROTONIN REUPTAKE INHIBITOR TREATMENT DURING PREGNANCY: IMMEDIATE AND LONGER TERM CHILD OUTCOMES. Fetal and Maternal Medicine Review, 2012, 23, 230-275.	0.3	4
106	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. Archives of Disease in Childhood, 2012, 97, 812-817.	1.0	36
107	Meierâ€“Gorlin syndrome genotypeâ€“phenotype studies: 35 individuals with pre-replication complex gene mutations and 10 without molecular diagnosis. European Journal of Human Genetics, 2012, 20, 598-606.	1.4	95
108	FETAL DYSMORPHOLOGY. Fetal and Maternal Medicine Review, 2012, 23, 52-70.	0.3	0

#	Article	IF	Citations
109	Another cause of vaccine encephalopathy: A case of Angelman syndrome. European Journal of Medical Genetics, 2012, 55, 338-341.	0.7	7
110	How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. European Journal of Human Genetics, 2012, 20, 381-388.	1.4	142
111	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. Human Mutation, 2012, 33, 1261-1266.	1.1	47
112	Perrault syndrome: further evidence for genetic heterogeneity. Journal of Neurology, 2012, 259, 974-976.	1.8	27
113	Relationship of child IQ to parental IQ and education in children with fetal antiepileptic drug exposure. Epilepsy and Behavior, 2011, 21, 147-152.	0.9	29
114	Mutations in CEP57 cause mosaic variegated aneuploidy syndrome. Nature Genetics, 2011, 43, 527-529.	9.4	117
115	Fetal antiepileptic drug exposure: Motor, adaptive, and emotional/behavioral functioning at age 3years. Epilepsy and Behavior, 2011, 22, 240-246.	0.9	76
116	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. American Journal of Human Genetics, 2011, 88, 767-777.	2.6	106
117	Whole-Exome-Sequencing Identifies Mutations in Histone Acetyltransferase Gene KAT6B in Individuals with the Say-Barber-Biesecker Variant of Ohdo Syndrome. American Journal of Human Genetics, 2011, 89, 675-681.	2.6	156
118	Foetal antiepileptic drug exposure and verbal versus non-verbal abilities at three years of age. Brain, 2011, 134, 396-404.	3.7	140
119	Cutaneous features in 17q21.31 deletion syndrome. Clinical Dysmorphology, 2011, 20, 15-20.	0.1	20
120	LRP4 Mutations Alter Wnt/Î²-Catenin Signaling and Cause Limb and Kidney Malformations in Cenani-Lenz Syndrome. American Journal of Human Genetics, 2010, 86, 696-706.	2.6	151
121	Familial 3q29 microdeletion syndrome providing further evidence of involvement of the 3q29 region in bipolar disorder. Clinical Dysmorphology, 2010, 19, 128-132.	0.1	34
122	De-novo duplication of 5(q13.3q21.1) in a child with vitreo-retinal dysplasia and learning disability. Clinical Dysmorphology, 2010, 19, 73-75.	0.1	0
123	VSX2 in microphthalmia: a novel splice site mutation producing a severe microphthalmia phenotype. British Journal of Ophthalmology, 2010, 94, 386-388.	2.1	18
124	Large, rare chromosomal deletions associated with severe early-onset obesity. Nature, 2010, 463, 666-670.	13.7	487
125	Cognitive Function at 3 Years of Age after Fetal Exposure to Antiepileptic Drugs. New England Journal of Medicine, 2009, 360, 1597-1605.	13.9	754
126	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

#	Article	IF	Citations
127	Complex Segmental Duplications Mediate a Recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. American Journal of Human Genetics, 2009, 85, 394-400.	2.6	60
128	4.5ÂMb microdeletion in chromosome band 2q33.1 associated with learning disability and cleft palate. European Journal of Medical Genetics, 2009, 52, 454-457.	0.7	33
129	Fibular aplasia in a child exposed to sodium valproate in pregnancy. Clinical Dysmorphology, 2009, 18, 37-39.	0.1	9
130	Identification of 34 novel and 56 knownFOXL2mutations in patients with blepharophimosis syndrome. Human Mutation, 2008, 29, E205-E219.	1.1	42
131	Human Osteoclast-Poor Osteopetrosis with Hypogammaglobulinemia due to TNFRSF11A (RANK) Mutations. American Journal of Human Genetics, 2008, 83, 64-76.	2.6	270
132	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. Nature Genetics, 2008, 40, 1410-1412.	9.4	138
133	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
134	Bardet-Biedl Syndrome: An Atypical Phenotype in Brothers with a ProvenBBS1Mutation. Ophthalmic Genetics, 2008, 29, 128-132.	0.5	18
135	Bilateral camptodactyly and recurrent patellar dislocation: a new sign of 22q11 deletions or an independent dominant disorder?. Clinical Dysmorphology, 2008, 17, 157-159.	0.1	3
136	Complementation in a 45,X/47,XX,+14 patient?. Clinical Dysmorphology, 2008, 17, 291.	0.1	0
137	Cerebro-facio-thoracic dysplasia: expanding the phenotype. Clinical Dysmorphology, 2007, 16, 121-125.	0.1	8
138	Osteocraniostenosis: a further case report documenting the antenatal findings. Clinical Dysmorphology, 2007, 16, 117-120.	0.1	2
139	A new X-linked mental retardation (XLMR) syndrome with late-onset primary testicular failure, short stature and microcephaly maps to Xq25â€“q26. European Journal of Medical Genetics, 2007, 50, 216-223.	0.7	3
140	Influence of the MTHFR genotype on the rate of malformations following exposure to antiepileptic drugs in utero. European Journal of Medical Genetics, 2007, 50, 411-420.	0.7	29
141	Mutations in CUL4B, Which Encodes a Ubiquitin E3 Ligase Subunit, Cause an X-linked Mental Retardation Syndrome Associated with Aggressive Outbursts, Seizures, Relative Macrocephaly, Central Obesity, Hypogonadism, Pes Cavus, and Tremor. American Journal of Human Genetics, 2007, 80, 345-352.	2.6	197
142	Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). American Journal of Human Genetics, 2007, 80, 994-1001.	2.6	261
143	Mapping of Deletion and Translocation Breakpoints in 1q44 Implicates the Serine/Threonine Kinase AKT3 in Postnatal Microcephaly and Agenesis of the Corpus Callosum. American Journal of Human Genetics, 2007, 81, 292-303.	2.6	144
144	Guidelines for molecular karyotyping in constitutional genetic diagnosis. European Journal of Human Genetics, 2007, 15, 1105-1114.	1.4	144

#	Article	IF	Citations
145	Exploring the Role of Patients' Spiritual/Religious Beliefs around Predictive Genetic Testing. Australian Journal of Cancer Nursing, 2007, 9, 252-252.	0.8	2
146	Aplasia cutis congenita and low molecular weight heparin. BJOG: an International Journal of Obstetrics and Gynaecology, 2005, 112, 256-258.	1.1	12
147	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. Nature Genetics, 2005, 37, 221-224.	9.4	201
148	A cross-linker-sensitive myeloid leukemia cell line from a 2-year-old boy with severe Fanconi anemia and biallelicFANCD1/BRCA2 mutations. Genes Chromosomes and Cancer, 2005, 42, 404-415.	1.5	28
149	Discriminating Power of Localized Three-Dimensional Facial Morphology. American Journal of Human Genetics, 2005, 77, 999-1010.	2.6	133
150	Delineation of Cohen Syndrome Following a Large-Scale Genotype-Phenotype Screen. American Journal of Human Genetics, 2004, 75, 122-127.	2.6	99
151	Acromegaloid facial appearance syndrome: a further case report. Clinical Dysmorphology, 2004, 13, 251-253.	0.1	5
152	Cohen Syndrome Is Caused by Mutations in a Novel Gene, COH1, Encoding a Transmembrane Protein with a Presumed Role in Vesicle-Mediated Sorting and Intracellular Protein Transport. American Journal of Human Genetics, 2003, 72, 1359-1369.	2.6	321
153	Mutations in PHF6 are associated with BÃ¶rjesonâ€“ForssmanÂâ€“Lehmann syndrome. Nature Genetics, 2002, 32, 661-665.	9.4	192
154	Outcome of pregnancy in women attending an outpatient epilepsy clinic: adverse features associated with higher doses of sodium valproate. Seizure: the Journal of the British Epilepsy Association, 2002, 11, 512-518.	0.9	91
155	Chromosome 7p disruptions in Silver Russell syndrome: delineating an imprinted candidate gene region.. Human Genetics, 2002, 111, 376-387.	1.8	79
156	Exploring the complex relationship between adolescent sexual offending and sex chromosome abnormality. Psychiatric Genetics, 2001, 11, 5-10.	0.6	9
157	Angelman syndrome: evolution of the phenotype in adolescents and adults. Developmental Medicine and Child Neurology, 2001, 43, 476.	1.1	100
158	A survey ofTWIST for mutations in craniosynostosis reveals a variable length polyglycine tract in asymptomatic individuals. Human Mutation, 2001, 18, 535-541.	1.1	39
159	Angelman syndrome associated with a maternal 15q11â€“13 deletion of less than 200 kb. Human Molecular Genetics, 1994, 3, 1409-1413.	1.4	23
160	Alphaâ€fetoprotein in Angelman Syndrome. Developmental Medicine and Child Neurology, 1991, 33, 182-183.	1.1	1