

Jill Clayton-Smith

List of Publications by Year in descending order

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Version: 2024-02-01

160
papers

11,851
citations

36691

53
h-index

35168

102
g-index

169
all docs

169
docs citations

169
times ranked

18232
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. <i>Journal of Medical Genetics</i> , 2022, 59, 393-398.	1.5	14
2	A standard of care for individuals with <sc><i>PIK3CA</i></sc>-related disorders: An international expert consensus statement. <i>Clinical Genetics</i> , 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. <i>Genetics in Medicine</i> , 2022, 24, 1261-1273.	1.1	14
4	Further delineation of phenotypic spectrum of <sc><i>SCN2A</i></sc>-related disorder. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Journal of Community Genetics</i> , 2022, 13, 313-327.	0.5	3
6	Recommendations for whole genome sequencing in diagnostics for rare diseases. <i>European Journal of Human Genetics</i> , 2022, 30, 1017-1021.	1.4	48
7	<i>ATP1A2</i> and <i>ATP1A3</i> associated early profound epileptic encephalopathy and polymicrogyria. <i>Brain</i> , 2021, 144, 1435-1450.	3.7	35
8	Recurrent <sc><i>KCNT2</i></sc> missense variants affecting p.Arg190 result in a recognizable phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3083-3091.	0.7	7
9	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021, 108, 1138-1150.	2.6	17
10	The adaptive functioning profile of Pitt-Hopkins syndrome. <i>European Journal of Medical Genetics</i> , 2021, 64, 104279.	0.7	3
11	The diagnostic utility of clinical exome sequencing in 60 patients with hearing loss disorders: A single-institution experience. <i>Clinical Otolaryngology</i> , 2021, 46, 1257-1262.	0.6	3
12	Prevalence of fetal alcohol spectrum disorder in Greater Manchester, UK: An active case ascertainment study. <i>Alcoholism: Clinical and Experimental Research</i> , 2021, 45, 2271-2281.	1.4	21
13	Identification of <i>LAMA1</i> mutations ends diagnostic odyssey and has prognostic implications for patients with presumed Joubert syndrome. <i>Brain Communications</i> , 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. <i>Scientific Reports</i> , 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. <i>European Journal of Medical Genetics</i> , 2020, 63, 103658.	0.7	6
16	Exome sequencing in patients with antiepileptic drug exposure and complex phenotypes. <i>Archives of Disease in Childhood</i> , 2020, 105, 384-389.	1.0	3
17	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotype-phenotype correlations, and molecular basis. <i>Genetics in Medicine</i> , 2020, 22, 389-397.	1.1	53
18	Clinical utility of genetic testing in 201 preschool children with inherited eye disorders. <i>Genetics in Medicine</i> , 2020, 22, 745-751.	1.1	42

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

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37	Clinical and genetic variability in children with partial albinism. <i>Scientific Reports</i> , 2019, 9, 16576.	1.6	26
38	Traboulsi syndrome due to ASPH mutation: an under-recognised cause of ectopia lentis. <i>Clinical Dysmorphology</i> , 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. <i>BMJ Open</i> , 2019, 9, e029780.	0.8	37
40	Clinical and genetic heterogeneity in Melkersson-Rosenthal Syndrome. <i>European Journal of Medical Genetics</i> , 2019, 62, 103536.	0.7	8
41	Pathogenicity and selective constraint on variation near splice sites. <i>Genome Research</i> , 2019, 29, 159-170.	2.4	70
42	Fetal antiepileptic drug exposure and learning and memory functioning at 6 years of age: The NEAD prospective observational study. <i>Epilepsy and Behavior</i> , 2019, 92, 154-164.	0.9	30
43	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307.	1.1	80
44	Intellectual functioning in clinically confirmed fetal valproate syndrome. <i>Neurotoxicology and Teratology</i> , 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. <i>European Journal of Human Genetics</i> , 2019, 27, 278-290.	1.4	30
46	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. <i>Biological Psychiatry</i> , 2019, 85, 287-297.	0.7	108
47	Observation of Cleft Palate in an Individual with <i>SOX11</i> Mutation. <i>Cleft Palate-Craniofacial Journal</i> , 2018, 55, 456-461.	0.5	15
48	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 468-479.	2.6	63
49	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. <i>American Journal of Human Genetics</i> , 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. <i>Journal of Medical Genetics</i> , 2018, 55, 89-96.	1.5	7
51	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. <i>Journal of Medical Genetics</i> , 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. <i>American Journal of Human Genetics</i> , 2018, 102, 1195-1203.	2.6	37
53	Monotherapy treatment of epilepsy in pregnancy: congenital malformation outcomes in the child. <i>The Cochrane Library</i> , 2017, 2017, CD010224.	1.5	135
54	Structural analysis of pathogenic mutations in the <i>DYRK1A</i> gene in patients with developmental disorders. <i>Human Molecular Genetics</i> , 2017, 26, ddw409.	1.4	33

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55	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. <i>Genetics in Medicine</i> , 2017, 19, 900-908.	1.1	46
56	Interrupted/bipartite clavicle as a diagnostic clue in Kabuki syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1115-1118.	0.7	3
57	Genitourinary malformations: an under-recognized feature of ectrodactyly, ectodermal dysplasia and cleft lip/palate syndrome. <i>Clinical Dysmorphology</i> , 2017, 26, 78-82.	0.1	7
58	Phenotypic Heterogeneity in a Congenital Disorder of Glycosylation Caused by Mutations in STT3A. <i>Journal of Child Neurology</i> , 2017, 32, 560-565.	0.7	10
59	Association of Steroid 5 α -Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. <i>JAMA Ophthalmology</i> , 2017, 135, 339.	1.4	43
60	Panel-Based Clinical Genetic Testing in 85 Children with Inherited Retinal Disease. <i>Ophthalmology</i> , 2017, 124, 985-991.	2.5	51
61	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. <i>Genetics in Medicine</i> , 2017, 19, 701-710.	1.1	13
62	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. <i>American Journal of Human Genetics</i> , 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. <i>European Journal of Medical Genetics</i> , 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. <i>Journal of Pediatric Genetics</i> , 2017, 06, 129-141.	0.3	38
65	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 1021-1033.	2.6	83
66	Protein structure and phenotypic analysis of pathogenic and population missense variants in <i>STXBP1</i> . <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 495-507.	0.6	29
67	Novel <i>PEX11B</i> 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 \hat{P} -PAX6-Negative \hat{P} ™ Individuals with Aniridia or Gillespie Syndrome. <i>PLoS ONE</i> , 2016, 11, e0153757.	1.1	54
69	In utero exposure to valproate increases the risk of isolated cleft palate. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , 2016, 101, F207-F211.	1.4	30
70	Mutations in the HECT domain of NEDD4L lead to AKT \hat{P} mTOR pathway deregulation and cause periventricular nodular heterotopia. <i>Nature Genetics</i> , 2016, 48, 1349-1358.	9.4	101
71	Cognition in school-age children exposed to levetiracetam, topiramate, or sodium valproate. <i>Neurology</i> , 2016, 87, 1943-1953.	1.5	98
72	A Recurrent Mosaic Mutation in SMO , Encoding the Hedgehog Signal Transducer Smoothed, Is the Major Cause of Curry-Jones Syndrome. <i>American Journal of Human Genetics</i> , 2016, 98, 1256-1265.	2.6	70

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73	Exploring the genetic basis of 3MC syndrome: Findings in 12 further families. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Clinical Dysmorphology</i> , 2016, 25, 16-18.	0.1	8
75	Next-generation Sequencing in the Diagnosis of Metabolic Disease Marked by Pediatric Cataract. <i>Ophthalmology</i> , 2016, 123, 217-220.	2.5	44
76	Rutherford syndrome revisited. <i>Clinical Dysmorphology</i> , 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. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 883-891.	3.3	22
78	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. <i>European Journal of Medical Genetics</i> , 2015, 58, 455-465.	0.7	83
79	IQ at 6 years after in utero exposure to antiepileptic drugs. <i>Neurology</i> , 2015, 84, 382-390.	1.5	226
80	Mutations in <i>SIPA1L3</i> cause eye defects through disruption of cell polarity and cytoskeleton organization. <i>Human Molecular Genetics</i> , 2015, 24, 5789-5804.	1.4	32
81	Agnathia-otocephaly complex and asymmetric velopharyngeal insufficiency due to an in-frame duplication in OTX2. <i>Journal of Human Genetics</i> , 2015, 60, 199-202.	1.1	25
82	A recurrent synonymous <i>KAT6B</i> mutation causes Say-Barber-Biesecker/Young-Simpson syndrome by inducing aberrant splicing. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3006-3010.	0.7	17
83	Deletion of 19q13 reveals clinical overlap with Dubowitz syndrome. <i>Journal of Human Genetics</i> , 2015, 60, 781-785.	1.1	12
84	Interchromosomal Insertional Translocation at Xq26.3 Alters <i>SOX3</i> Expression in an Individual With XX Male Sex Reversal. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E815-E820.	1.8	46
85	Further delineation of the <i>KAT6B</i> molecular and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 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. <i>Journal of Medical Genetics</i> , 2014, 51, 659-668.	1.5	141
87	Velopharyngeal insufficiency: high detection rate of genetic abnormalities if associated with additional features. <i>Archives of Disease in Childhood</i> , 2014, 99, 52-57.	1.0	6
88	Compound Heterozygosity of Low-Frequency Promoter Deletions and Rare Loss-of-Function Mutations in <i>TXNL4A</i> Causes Burn-McKeown Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 698-707.	2.6	55
89	Breastfeeding in Children of Women Taking Antiepileptic Drugs. <i>JAMA Pediatrics</i> , 2014, 168, 729.	3.3	201
90	Ocular coloboma and foetal valproate syndrome. <i>Clinical Dysmorphology</i> , 2014, 23, 74-75.	0.1	7

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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. <i>Human Molecular Genetics</i> , 2014, 23, 2752-2768.	1.4	140
92	Mutations in CKAP2L, the Human Homolog of the Mouse Radmis Gene, Cause Filippi Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 622-632.	2.6	34
93	Personalized Diagnosis and Management of Congenital Cataract by Next-Generation Sequencing. <i>Ophthalmology</i> , 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. <i>Genome Research</i> , 2014, 24, 673-687.	2.4	53
95	Oculo-auriculo-vertebral spectrum: a review of the literature and genetic update. <i>Journal of Medical Genetics</i> , 2014, 51, 635-645.	1.5	140
96	Trisomy 18 mosaicism: report of two cases. <i>World Journal of Pediatrics</i> , 2013, 9, 179-181.	0.8	15
97	Perrault Syndrome Is Caused by Recessive Mutations in CLPP, Encoding a Mitochondrial ATP-Dependent Chambered Protease. <i>American Journal of Human Genetics</i> , 2013, 92, 605-613.	2.6	186
98	Fetal antiepileptic drug exposure: Adaptive and emotional/behavioral functioning at age 6 years. <i>Epilepsy and Behavior</i> , 2013, 29, 308-315.	0.9	132
99	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. <i>Human Mutation</i> , 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. <i>Lancet Neurology</i> , The, 2013, 12, 244-252.	4.9	665
101	ALDH1A3 Mutations Cause Recessive Anophthalmia and Microphthalmia. <i>American Journal of Human Genetics</i> , 2013, 92, 265-270.	2.6	92
102	The prevalence of neurodevelopmental disorders in children prenatally exposed to antiepileptic drugs. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 637-643.	0.9	280
103	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. <i>Brain</i> , 2013, 136, 3378-3394.	3.7	85
104	12p13.33 microdeletion including ELKS/ERC1, a new locus associated with childhood apraxia of speech. <i>European Journal of Human Genetics</i> , 2013, 21, 82-88.	1.4	70
105	FETAL EFFECTS OF SELECTIVE SEROTONIN REUPTAKE INHIBITOR TREATMENT DURING PREGNANCY: IMMEDIATE AND LONGER TERM CHILD OUTCOMES. <i>Fetal and Maternal Medicine Review</i> , 2012, 23, 230-275.	0.3	4
106	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. <i>Archives of Disease in Childhood</i> , 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. <i>European Journal of Human Genetics</i> , 2012, 20, 598-606.	1.4	95
108	FETAL DYSMORPHOLOGY. <i>Fetal and Maternal Medicine Review</i> , 2012, 23, 52-70.	0.3	0

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109	Another cause of vaccine encephalopathy: A case of Angelman syndrome. <i>European Journal of Medical Genetics</i> , 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. <i>European Journal of Human Genetics</i> , 2012, 20, 381-388.	1.4	142
111	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. <i>Human Mutation</i> , 2012, 33, 1261-1266.	1.1	47
112	Perrault syndrome: further evidence for genetic heterogeneity. <i>Journal of Neurology</i> , 2012, 259, 974-976.	1.8	27
113	Relationship of child IQ to parental IQ and education in children with fetal antiepileptic drug exposure. <i>Epilepsy and Behavior</i> , 2011, 21, 147-152.	0.9	29
114	Mutations in CEP57 cause mosaic variegated aneuploidy syndrome. <i>Nature Genetics</i> , 2011, 43, 527-529.	9.4	117
115	Fetal antiepileptic drug exposure: Motor, adaptive, and emotional/behavioral functioning at age 3years. <i>Epilepsy and Behavior</i> , 2011, 22, 240-246.	0.9	76
116	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. <i>American Journal of Human Genetics</i> , 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. <i>American Journal of Human Genetics</i> , 2011, 89, 675-681.	2.6	156
118	Foetal antiepileptic drug exposure and verbal versus non-verbal abilities at three years of age. <i>Brain</i> , 2011, 134, 396-404.	3.7	140
119	Cutaneous features in 17q21.31 deletion syndrome. <i>Clinical Dysmorphology</i> , 2011, 20, 15-20.	0.1	20
120	LRP4 Mutations Alter Wnt/ β -Catenin Signaling and Cause Limb and Kidney Malformations in Cenani-Lenz Syndrome. <i>American Journal of Human Genetics</i> , 2010, 86, 696-706.	2.6	151
121	Familial 3q29 microdeletion syndrome providing further evidence of involvement of the 3q29 region in bipolar disorder. <i>Clinical Dysmorphology</i> , 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. <i>Clinical Dysmorphology</i> , 2010, 19, 73-75.	0.1	0
123	VSX2 in microphthalmia: a novel splice site mutation producing a severe microphthalmia phenotype. <i>British Journal of Ophthalmology</i> , 2010, 94, 386-388.	2.1	18
124	Large, rare chromosomal deletions associated with severe early-onset obesity. <i>Nature</i> , 2010, 463, 666-670.	13.7	487
125	Cognitive Function at 3 Years of Age after Fetal Exposure to Antiepileptic Drugs. <i>New England Journal of Medicine</i> , 2009, 360, 1597-1605.	13.9	754
126	Xq28 duplication presenting with intestinal and bladder dysfunction and a distinctive facial appearance. <i>European Journal of Human Genetics</i> , 2009, 17, 434-443.	1.4	87

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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. <i>American Journal of Human Genetics</i> , 2009, 85, 394-400.	2.6	60
128	4.5-Mb microdeletion in chromosome band 2q33.1 associated with learning disability and cleft palate. <i>European Journal of Medical Genetics</i> , 2009, 52, 454-457.	0.7	33
129	Fibular aplasia in a child exposed to sodium valproate in pregnancy. <i>Clinical Dysmorphology</i> , 2009, 18, 37-39.	0.1	9
130	Identification of 34 novel and 56 known <i>FOXL2</i> mutations in patients with blepharophimosis syndrome. <i>Human Mutation</i> , 2008, 29, E205-E219.	1.1	42
131	Human Osteoclast-Poor Osteopetrosis with Hypogammaglobulinemia due to <i>TNFRSF11A</i> (RANK) Mutations. <i>American Journal of Human Genetics</i> , 2008, 83, 64-76.	2.6	270
132	Geroderma osteodysplastica is caused by mutations in <i>SCYL1BP1</i> , a Rab-6 interacting golgin. <i>Nature Genetics</i> , 2008, 40, 1410-1412.	9.4	138
133	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. <i>New England Journal of Medicine</i> , 2008, 359, 1685-1699.	13.9	663
134	Bardet-Biedl Syndrome: An Atypical Phenotype in Brothers with a Proven <i>BBS1</i> Mutation. <i>Ophthalmic Genetics</i> , 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?. <i>Clinical Dysmorphology</i> , 2008, 17, 157-159.	0.1	3
136	Complementation in a 45,X/47,XX,+14 patient?. <i>Clinical Dysmorphology</i> , 2008, 17, 291.	0.1	0
137	Cerebro-facio-thoracic dysplasia: expanding the phenotype. <i>Clinical Dysmorphology</i> , 2007, 16, 121-125.	0.1	8
138	Osteocraniostenosis: a further case report documenting the antenatal findings. <i>Clinical Dysmorphology</i> , 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 Xq25-q26. <i>European Journal of Medical Genetics</i> , 2007, 50, 216-223.	0.7	3
140	Influence of the <i>MTHFR</i> genotype on the rate of malformations following exposure to antiepileptic drugs in utero. <i>European Journal of Medical Genetics</i> , 2007, 50, 411-420.	0.7	29
141	Mutations in <i>CUL4B</i> , 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. <i>American Journal of Human Genetics</i> , 2007, 80, 345-352.	2.6	197
142	Haploinsufficiency of <i>TCF4</i> Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). <i>American Journal of Human Genetics</i> , 2007, 80, 994-1001.	2.6	261
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