Jill Clayton-Smith

List of Publications by Year
in descending order

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160	11,851	31976	30922
papers	citations	53 h-index	102 g-index

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

#	Article	IF	Citations
1	Cognitive Function at 3 Years of Age after Fetal Exposure to Antiepileptic Drugs. New England Journal of Medicine, 2009, 360, 1597-1605.	27.0	754
2	Fetal antiepileptic drug exposure and cognitive outcomes at age 6 years (NEAD study): a prospective observational study. Lancet Neurology, The, 2013, 12, 244-252.	10.2	665
3	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
4	Large, rare chromosomal deletions associated with severe early-onset obesity. Nature, 2010, 463, 666-670.	27.8	487
5	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.	6.2	321
6	The prevalence of neurodevelopmental disorders in children prenatally exposed to antiepileptic drugs. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 637-643.	1.9	280
7	Human Osteoclast-Poor Osteopetrosis with Hypogammaglobulinemia due to TNFRSF11A (RANK) Mutations. American Journal of Human Genetics, 2008, 83, 64-76.	6.2	270
8	Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). American Journal of Human Genetics, 2007, 80, 994-1001.	6.2	261
9	IQ at 6 years after in utero exposure to antiepileptic drugs. Neurology, 2015, 84, 382-390.	1.1	226
10	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
11	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. Nature Genetics, 2005, 37, 221-224.	21.4	201
12	Breastfeeding in Children of Women Taking Antiepileptic Drugs. JAMA Pediatrics, 2014, 168, 729.	6.2	201
13	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.	6.2	197
14	Mutations in PHF6 are associated with BÃ¶rjesonâ€“ForssmanÂâ€“Lehmann syndrome. Nature Genetics, 2002, 32, 661-665.	21.4	192
15	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.	6.2	186
16	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
17	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.	6.2	156
18	Personalized Diagnosis and Management of Congenital Cataract by Next-Generation Sequencing. Ophthalmology, 2014, 121, 2124-2137.e2.	5.2	153

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19	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.	6.2	151
20	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
21	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.	6.2	144
22	Guidelines for molecular karyotyping in constitutional genetic diagnosis. European Journal of Human Genetics, 2007, 15, 1105-1114.	2.8	144
23	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.	2.8	142
24	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.	3.2	141
25	Foetal antiepileptic drug exposure and verbal versus non-verbal abilities at three years of age. Brain, 2011, 134, 396-404.	7.6	140
26	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.	2.9	140
27	Oculo-auriculo-vertebral spectrum: a review of the literature and genetic update. Journal of Medical Genetics, 2014, 51, 635-645.	3.2	140
28	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. Nature Genetics, 2008, 40, 1410-1412.	21.4	138
29	Monotherapy treatment of epilepsy in pregnancy: congenital malformation outcomes in the child. The Cochrane Library, 2017, 2017, CD010224.	2.8	135
30	Discriminating Power of Localized Three-Dimensional Facial Morphology. American Journal of Human Genetics, 2005, 77, 999-1010.	6.2	133
31	Fetal antiepileptic drug exposure: Adaptive and emotional/behavioral functioning at age 6years. Epilepsy and Behavior, 2013, 29, 308-315.	1.7	132
32	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	6.2	119
33	Mutations in CEP57 cause mosaic variegated aneuploidy syndrome. Nature Genetics, 2011, 43, 527-529.	21.4	117
34	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
35	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.	6.2	106
36	Mutations in the HECT domain of NEDD4L lead to AKTâ€“mTOR pathway deregulation and cause periventricular nodular heterotopia. Nature Genetics, 2016, 48, 1349-1358.	21.4	101

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37	Angelman syndrome: evolution of the phenotype in adolescents and adults. Developmental Medicine and Child Neurology, 2001, 43, 476.	2.1	100
38	Delineation of Cohen Syndrome Following a Large-Scale Genotype-Phenotype Screen. American Journal of Human Genetics, 2004, 75, 122-127.	6.2	99
39	Cognition in school-age children exposed to levetiracetam, topiramate, or sodium valproate. Neurology, 2016, 87, 1943-1953.	1.1	98
40	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.	2.8	95
41	ALDH1A3 Mutations Cause Recessive Anophthalmia and Microphthalmia. American Journal of Human Genetics, 2013, 92, 265-270.	6.2	92
42	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.	2.0	91
43	Xq28 duplication presenting with intestinal and bladder dysfunction and a distinctive facial appearance. European Journal of Human Genetics, 2009, 17, 434-443.	2.8	87
44	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
45	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. European Journal of Medical Genetics, 2015, 58, 455-465.	1.3	83
46	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	6.2	83
47	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
48	Chromosome 7p disruptions in Silver Russell syndrome: delineating an imprinted candidate gene region.. Human Genetics, 2002, 111, 376-387.	3.8	79
49	Fetal antiepileptic drug exposure: Motor, adaptive, and emotional/behavioral functioning at age 3years. Epilepsy and Behavior, 2011, 22, 240-246.	1.7	76
50	12p13.33 microdeletion including ELKS/ERC1, a new locus associated with childhood apraxia of speech. European Journal of Human Genetics, 2013, 21, 82-88.	2.8	70
51	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.	6.2	70
52	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
53	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479.	6.2	63
54	Diagnosis and management in Pittâ€Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	2.0	63

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55	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.	6.2	60
56	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Journal of Medical Genetics, 2018, 55, 104-113.	3.2	59
57	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
58	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
59	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.	6.2	55
60	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	2.5	54
61	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.	5.5	53
62	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	2.4	53
63	Panel-Based Clinical Genetic Testing in 85 Children with Inherited Retinal Disease. Ophthalmology, 2017, 124, 985-991.	5.2	51
64	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
65	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. Human Mutation, 2012, 33, 1261-1266.	2.5	47
66	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.	3.6	46
67	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
68	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
69	Next-generation Sequencing in the Diagnosis of Metabolic Disease Marked by Pediatric Cataract. Ophthalmology, 2016, 123, 217-220.	5.2	44
70	Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339.	2.5	43
71	Identification of 34 novel and 56 knownFOXL2mutations in patients with blepharophimosis syndrome. Human Mutation, 2008, 29, E205-E219.	2.5	42
72	Clinical utility of genetic testing in 201 preschool children with inherited eye disorders. Genetics in Medicine, 2020, 22, 745-751.	2.4	42

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73	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.	2.8	41
74	A survey ofTWIST for mutations in craniosynostosis reveals a variable length polyglycine tract in asymptomatic individuals. Human Mutation, 2001, 18, 535-541.	2.5	39
75	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.	0.7	38
76	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.	6.2	37
77	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.	1.9	37
78	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607.	3.3	37
79	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. Archives of Disease in Childhood, 2012, 97, 812-817.	1.9	36
80	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
81	Familial 3q29 microdeletion syndrome providing further evidence of involvement of the 3q29 region in bipolar disorder. Clinical Dysmorphology, 2010, 19, 128-132.	0.3	34
82	Mutations in CKAP2L, the Human Homolog of the Mouse Radmis Gene, Cause Filippi Syndrome. American Journal of Human Genetics, 2014, 95, 622-632.	6.2	34
83	4.5ÂMb microdeletion in chromosome band 2q33.1 associated with learning disability and cleft palate. European Journal of Medical Genetics, 2009, 52, 454-457.	1.3	33
84	Structural analysis of pathogenic mutations in theDYRK1Agene in patients with developmental disorders. Human Molecular Genetics, 2017, 26, ddw409.	2.9	33
85	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.	2.7	33
86	Mutations inSIPA1L3cause eye defects through disruption of cell polarity and cytoskeleton organization. Human Molecular Genetics, 2015, 24, 5789-5804.	2.9	32
87	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.	1.6	31
88	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.	2.8	30
89	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.	1.7	30
90	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.	2.8	30

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91	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.	1.3	29
92	Relationship of child IQ to parental IQ and education in children with fetal antiepileptic drug exposure. Epilepsy and Behavior, 2011, 21, 147-152.	1.7	29
93	Protein structure and phenotypic analysis of pathogenic and population missense variants inSTXBP1. Molecular Genetics & Genomic Medicine, 2017, 5, 495-507.	1.2	29
94	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.	2.8	28
95	Perrault syndrome: further evidence for genetic heterogeneity. Journal of Neurology, 2012, 259, 974-976.	3.6	27
96	Clinical and genetic variability in children with partial albinism. Scientific Reports, 2019, 9, 16576.	3.3	26
97	Agnathia-otocephaly complex and asymmetric velopharyngeal insufficiency due to an in-frame duplication in OTX2. Journal of Human Genetics, 2015, 60, 199-202.	2.3	25
98	Exploring the genetic basis of 3MC syndrome: Findings in 12 further families. American Journal of Medical Genetics, Part A, 2016, 170, 1216-1224.	1.2	25
99	Novel PEX11B Mutations Extend the Peroxisome Biogenesis Disorder 14B Phenotypic Spectrum and Underscore Congenital Cataract as an Early Feature. , 2017, 58, 594.		25
100	Angelman syndrome associated with a maternal 15q11â€“13 deletion of less than 200 kb. Human Molecular Genetics, 1994, 3, 1409-1413.	2.9	23
101	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.	1.2	23
102	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.	2.7	23
103	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
104	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
105	A standard of care for individuals with <scp>PIK3CA</scp>â€related disorders: An international expert consensus statement. Clinical Genetics, 2022, 101, 32-47.	2.0	21
106	Prevalence of fetal alcohol spectrum disorder in Greater Manchester, UK: An active case ascertainment study. Alcoholism: Clinical and Experimental Research, 2021, 45, 2271-2281.	2.4	21
107	Cutaneous features in 17q21.31 deletion syndrome. Clinical Dysmorphology, 2011, 20, 15-20.	0.3	20
108	Deep phenotyping of 14 new patients with IQSEC2 variants, including monozygotic twins of discordant phenotype. Clinical Genetics, 2019, 95, 496-506.	2.0	20

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109	Bardet-Biedl Syndrome: An Atypical Phenotype in Brothers with a ProvenBBS1Mutation. Ophthalmic Genetics, 2008, 29, 128-132.	1.2	18
110	VSX2 in microphthalmia: a novel splice site mutation producing a severe microphthalmia phenotype. British Journal of Ophthalmology, 2010, 94, 386-388.	3.9	18
111	Intellectual functioning in clinically confirmed fetal valproate syndrome. Neurotoxicology and Teratology, 2019, 71, 16-21.	2.4	18
112	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.	1.2	17
113	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.	6.2	17
114	Trisomy 18 mosaicism: report of two cases. World Journal of Pediatrics, 2013, 9, 179-181.	1.8	15
115	Observation of Cleft Palate in an Individual with SOX11 Mutation. Cleft Palate-Craniofacial Journal, 2018, 55, 456-461.	0.9	15
116	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
117	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.	3.2	14
118	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.	2.4	14
119	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. Genetics in Medicine, 2017, 19, 701-710.	2.4	13
120	Aplasia cutis congenita and low molecular weight heparin. BJOG: an International Journal of Obstetrics and Gynaecology, 2005, 112, 256-258.	2.3	12
121	Deletion of 19q13 reveals clinical overlap with Dubowitz syndrome. Journal of Human Genetics, 2015, 60, 781-785.	2.3	12
122	Mowat-Wilson syndrome: growth charts. Orphanet Journal of Rare Diseases, 2020, 15, 151.	2.7	12
123	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.	3.2	11
124	Phenotypic Heterogeneity in a Congenital Disorder of Glycosylation Caused by Mutations in STT3A. Journal of Child Neurology, 2017, 32, 560-565.	1.4	10
125	Traboulsi syndrome due to ASPH mutation: an under-recognised cause of ectopia lentis. Clinical Dysmorphology, 2019, 28, 184-189.	0.3	10
126	Exploring the complex relationship between adolescent sexual offending and sex chromosome abnormality. Psychiatric Genetics, 2001, 11, 5-10.	1.1	9

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127	Fibular aplasia in a child exposed to sodium valproate in pregnancy. Clinical Dysmorphology, 2009, 18, 37-39.	0.3	9
128	Cerebro-facio-thoracic dysplasia: expanding the phenotype. Clinical Dysmorphology, 2007, 16, 121-125.	0.3	8
129	Detection of a mosaic PIK3CA mutation in dental DNA from a child with megalencephaly capillary malformation syndrome. Clinical Dysmorphology, 2016, 25, 16-18.	0.3	8
130	Clinical and genetic heterogeneity in Melkersson-Rosenthal Syndrome. European Journal of Medical Genetics, 2019, 62, 103536.	1.3	8
131	A new mutational hotspot in the SKI gene in the context of MFS/TAA molecular diagnosis. Human Genetics, 2020, 139, 461-472.	3.8	8
132	Identification of LAMA1 mutations ends diagnostic odyssey and has prognostic implications for patients with presumed Joubert syndrome. Brain Communications, 2021, 3, fcab163.	3.3	8
133	Another cause of vaccine encephalopathy: A case of Angelman syndrome. European Journal of Medical Genetics, 2012, 55, 338-341.	1.3	7
134	Ocular coloboma and foetal valproate syndrome. Clinical Dysmorphology, 2014, 23, 74-75.	0.3	7
135	Genitourinary malformations: an under-recognized feature of ectrodactyly, ectodermal dysplasia and cleft lip/palate syndrome. Clinical Dysmorphology, 2017, 26, 78-82.	0.3	7
136	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.	1.3	7
137	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.	3.2	7
138	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.	1.2	7
139	Velopharyngeal insufficiency: high detection rate of genetic abnormalities if associated with additional features. Archives of Disease in Childhood, 2014, 99, 52-57.	1.9	6
140	Delivering effective genetic services for patients and families affected by cleft lip and/or palate. European Journal of Human Genetics, 2019, 27, 1018-1025.	2.8	6
141	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.	1.3	6
142	Rutherfurd syndrome revisited. Clinical Dysmorphology, 2015, 24, 125-127.	0.3	5
143	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.	1.3	5
144	Acromegaloid facial appearance syndrome: a further case report. Clinical Dysmorphology, 2004, 13, 251-253.	0.3	5

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145	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
146	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.	1.3	3
147	Bilateral camptodactyly and recurrent patellar dislocation: a new sign of 22q11 deletions or an independent dominant disorder?. Clinical Dysmorphology, 2008, 17, 157-159.	0.3	3
148	Interrupted/bipartite clavicle as a diagnostic clue in Kabuki syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1115-1118.	1.2	3
149	Exome sequencing in patients with antiepileptic drug exposure and complex phenotypes. Archives of Disease in Childhood, 2020, 105, 384-389.	1.9	3
150	The adaptive functioning profile of Pitt-Hopkins syndrome. European Journal of Medical Genetics, 2021, 64, 104279.	1.3	3
151	The diagnostic utility of clinical exome sequencing in 60 patients with hearing loss disorders: A singleâ€institution experience. Clinical Otolaryngology, 2021, 46, 1257-1262.	1.2	3
152	Further delineation of phenotypic spectrum of <scp>SCN2A</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 867-877.	1.2	3
153	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.	1.2	3
154	Osteocraniostenosis: a further case report documenting the antenatal findings. Clinical Dysmorphology, 2007, 16, 117-120.	0.3	2
155	Exploring the Role of Patients' Spiritual/Religious Beliefs around Predictive Genetic Testing. Australian Journal of Cancer Nursing, 2007, 9, 252-252.	1.6	2
156	Alphaâ€fetoprotein in Angelman Syndrome. Developmental Medicine and Child Neurology, 1991, 33, 182-183.	2.1	1
157	Oral-Facial-Digital Syndrome Type 1: Further Clinical and Molecular Delineation in 2 New Families. Cleft Palate-Craniofacial Journal, 2020, 57, 606-615.	0.9	1
158	Complementation in a 45,X/47,XX,+14 patient?. Clinical Dysmorphology, 2008, 17, 291.	0.3	0
159	De-novo duplication of 5(q13.3q21.1) in a child with vitreo-retinal dysplasia and learning disability. Clinical Dysmorphology, 2010, 19, 73-75.	0.3	0
160	FETAL DYSMORPHOLOGY. Fetal and Maternal Medicine Review, 2012, 23, 52-70.	0.3	0