John M Graham

List of Publications by Year
in descending order

170 papers	10,775 citations	41344 49 h-index	38395 95 g-index
177 all docs	177 docs citations	177 times ranked	11863 citing authors

#	Article	IF	Citations
1	Coloboma, congenital heart disease, and choanal atresia with multiple anomalies: CHARGE association. Journal of Pediatrics, 1981, 99, 223-227.	1.8	780
2	Incidence of fetal alcohol syndrome and prevalence of alcohol-related neurodevelopmental disorder. Teratology, 1997, 56, 317-326.	1.6	706
3	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. Nature Genetics, 2012, 44, 934-940.	21.4	621
4	CHARGE Association: An Update and Review for the Primary Pediatrician. Clinical Pediatrics, 1998, 37, 159-173.	0.8	394
5	Proteus syndrome: Diagnostic criteria, differential diagnosis, and patient evaluation. American Journal of Medical Genetics Part A, 1999, 84, 389-395.	2.4	374
6	Spectrum of CHD7 Mutations in 110 Individuals with CHARGE Syndrome and Genotype-Phenotype Correlation. American Journal of Human Genetics, 2006, 78, 303-314.	6.2	352
7	Evaluation of mental retardation: Recommendations of a consensus conference. , 1997, 72, 468-477.		344
8	Clinical delineation and natural history of the PIK3CAâ€related overgrowth spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 1713-1733.	1.2	249
9	Molar tooth sign of the midbrain-hindbrain junction: Occurrence in multiple distinct syndromes. American Journal of Medical Genetics Part A, 2004, 125A, 125-134.	2.4	213
10	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. Nature Genetics, 2005, 37, 221-224.	21.4	201
11	An epidemiological analysis of CHARGE syndrome: Preliminary results from a Canadian study. American Journal of Medical Genetics, Part A, 2005, 133A, 309-317.	1.2	200
12	Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. Brain, 2011, 134, 143-156.	7.6	200
13	Megalencephalyâ€capillary malformation (MCAP) and megalencephalyâ€polydactylyâ€polymicrogyriaâ€hydrocephalus (MPPH) syndromes: Two closely related disorders of brain overgrowth and abnormal brain and body morphogenesis. American Journal of Medical Genetics, Part A, 2012, 158A, 269-291.	1.2	188
14	A recurrent mutation in MED12 leading to R961W causes Opitz-Kaveggia syndrome. Nature Genetics, 2007, 39, 451-453.	21.4	179
15	Teratogen update: Gestational effects of maternal hyperthermia due to febrile illnesses and resultant patterns of defects in humans. , 1998, 58, 209-221.		177
16	Management of deformational plagiocephaly: Repositioning versus orthotic therapy. Journal of Pediatrics, 2005, 146, 258-262.	1.8	173
17	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	6.2	171
18	Loss-of-Function Mutations in RAB18 Cause Warburg Micro Syndrome. American Journal of Human Genetics, 2011, 88, 499-507.	6.2	158

#	Article	IF	Citations
19	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
20	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
21	Cerebro-Oculo-Facio-Skeletal Syndrome with a Nucleotide Excisionâ€“Repair Defect and a Mutated XPD Gene, with Prenatal Diagnosis in a Triplet Pregnancy. American Journal of Human Genetics, 2001, 69, 291-300.	6.2	120
22	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	2.5	114
23	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. Human Mutation, 2010, 31, 1142-1154.	2.5	111
24	Neuroimaging findings in macrocephalyâ€“capillary malformation: A longitudinal study of 17 patients. American Journal of Medical Genetics, Part A, 2007, 143A, 2981-3008.	1.2	103
25	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
26	Manitoba Aboriginal Kindred with Original Cerebro-Oculo-Facio-Skeletal Syndrome Has a Mutation in the Cockayne Syndrome Group B (CSB) Gene. American Journal of Human Genetics, 2000, 66, 1221-1228.	6.2	99
27	Central nervous system malformations in the CHARGE association. American Journal of Medical Genetics Part A, 1990, 37, 304-310.	2.4	95
28	Kilnefelter Syndrome and Its Variants: An Update and Review for the Primary Pediatrician. Clinical Pediatrics, 2001, 40, 639-651.	0.8	95
29	Deformational brachycephaly in supine-sleeping infants. Journal of Pediatrics, 2005, 146, 253-257.	1.8	92
30	Longitudinal observations on 15 children with Wiedemannâ€Beckwith syndrome. American Journal of Medical Genetics Part A, 1995, 56, 366-373.	2.4	89
31	Clinical features and management issues in Mowatâ€“Wilson syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2730-2741.	1.2	89
32	Further delineation of Kabuki syndrome in 48 wellâ€defined new individuals. American Journal of Medical Genetics, Part A, 2005, 132A, 265-272.	1.2	84
33	The mutational spectrum of brachydactyly type C. American Journal of Medical Genetics Part A, 2002, 112, 291-296.	2.4	82
34	GRIN1 mutation associated with intellectual disability alters NMDA receptor trafficking and function. Journal of Human Genetics, 2017, 62, 589-597.	2.3	81
35	Fetal constraint as a potential risk factor for craniosynostosis. American Journal of Medical Genetics, Part A, 2010, 152A, 394-400.	1.2	79
36	Williams-Beuren Syndrome: An Update and Review for the Primary Physician. Clinical Pediatrics, 1999, 38, 189-208.	0.8	78

#	Article	IF	Citations
37	Social function in multiple X and Y chromosome disorders: XXY, XYY, XXYY, XXXY. Developmental Disabilities Research Reviews, 2009, 15, 328-332.	2.9	77
38	Behavioral phenotype of sex chromosome aneuploidies: 48,XXYY, 48,XXXY, and 49,XXXXY. American Journal of Medical Genetics, Part A, 2007, 143A, 1198-1203.	1.2	72
39	Deletions of 20p12 in Alagille syndrome: Frequency and molecular characterization. , 1997, 70, 80-86.		71
40	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
41	Elements of morphology: Standard terminology for the periorbital region. American Journal of Medical Genetics, Part A, 2009, 149A, 29-39.	1.2	70
42	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
43	The morphogenesis of wormian bones: A study of craniosynostosis and purposeful cranial deformation. American Journal of Medical Genetics, Part A, 2007, 143A, 3243-3251.	1.2	68
44	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	6.2	66
45	MED12 related disorders. American Journal of Medical Genetics, Part A, 2013, 161, 2734-2740.	1.2	64
46	Agnathia, situs inversus, and associated malformations. Teratology, 1981, 23, 85-93.	1.6	61
47	Clinical and molecular studies in full trisomy 22: Further delineation of the phenotype and review of the literature. American Journal of Medical Genetics Part A, 1995, 56, 359-365.	2.4	61
48	Syndrome of coronal craniosynostosis with brachydactyly and carpal/tarsal coalition due to Pro250Arg mutation in FGFR3 gene. , 1998, 77, 322-329.		61
49	Preaxial ray reduction defects as part of valproic acid embryofetopathy. Prenatal Diagnosis, 1993, 13, 909-918.	2.3	60
50	A recognizable syndrome within CHARGE association: Hall-Hittner syndrome. American Journal of Medical Genetics Part A, 2001, 99, 120-123.	2.4	59
51	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
52	Behavioral features of CHARGE syndrome (Hall-Hittner syndrome) comparison with Down syndrome, Prader-Willi syndrome, and Williams syndrome. American Journal of Medical Genetics, Part A, 2005, 133A, 240-247.	1.2	57
53	MICRO syndrome: An entity distinct from COFS syndrome. American Journal of Medical Genetics Part A, 2004, 128A, 235-245.	2.4	54
54	Congenital diaphragmatic hernia in the Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1993, 47, 1018-1021.	2.4	53

#	Article	IF	Citations
55	Clinical management of patients with ASXL1 mutations and Bohringâ€“Opitz syndrome, emphasizing the need for Wilms tumor surveillance. American Journal of Medical Genetics, Part A, 2015, 167, 2122-2131.	1.2	52
56	Multigene deletions on chromosome 20q13.13-q13.2 includingSALL4 result in an expanded phenotype of Okihiro syndrome plus developmental delay. Human Mutation, 2007, 28, 830-830.	2.5	50
57	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
58	Subtle radiographic findings of achondroplasia in patients with Crouzon syndrome with acanthosis nigricans due to an Ala391Glu substitution in FGFR3. American Journal of Medical Genetics Part A, 2001, 98, 75-91.	2.4	47
59	Review of genetic and environmental factors leading to hypospadias. European Journal of Medical Genetics, 2014, 57, 453-463.	1.3	47
60	Compromise of the spinal canal in Proteus syndrome. American Journal of Medical Genetics Part A, 1993, 47, 656-659.	2.4	46
61	Broad phenotypic spectrum caused by an identical heterozygous CDMP-1 mutation in three unrelated families. , 2002, 117A, 136-142.		45
62	Distinctive phenotype in 9 patients with deletion of chromosome 1q24â€q25. American Journal of Medical Genetics, Part A, 2011, 155, 1336-1351.	1.2	42
63	Clinical review of genetic epileptic encephalopathies. European Journal of Medical Genetics, 2012, 55, 281-298.	1.3	42
64	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
65	Triploidy: Pregnancy complications and clinical findings in seven cases. Prenatal Diagnosis, 1989, 9, 409-419.	2.3	41
66	Prenatal diagnosis and dysmorphic findings in mosaic trisomy 16. Prenatal Diagnosis, 1994, 14, 257-266.	2.3	36
67	A new X-linked syndrome with agenesis of the corpus callosum, mental retardation, coloboma, micrognathia, and a mutation in theAlpha 4 gene at Xq13. American Journal of Medical Genetics Part A, 2003, 123A, 37-44.	2.4	35
68	Syndromic Immunodeficiencies: Genetic Syndromes Associated with Immune Abnormalities. Critical Reviews in Clinical Laboratory Sciences, 2003, 40, 587-642.	6.1	35
69	FG syndrome, an X-linked multiple congenital anomaly syndrome: The clinical phenotype and an algorithm for diagnostic testing. Genetics in Medicine, 2009, 11, 769-775.	2.4	35
70	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
71	Trisomy 8 mosaicism in chorionic villus sampling: Case report and counselling issues. Prenatal Diagnosis, 1994, 14, 451-454.	2.3	33
72	FG syndrome: Report of three new families with linkage to xq12-q22.1. American Journal of Medical Genetics Part A, 1998, 80, 145-156.	2.4	33

#	Article	IF	Citations
73	Radiographic findings and Gs-alpha bioactivity studies and mutation screening in acrodysostosis indicate a different etiology from pseudohypoparathyroidism. Pediatric Radiology, 2001, 31, 2-9.	2.0	33
74	KCNK9 imprinting syndromeâ€"further delineation of a possible treatable disorder. American Journal of Medical Genetics, Part A, 2016, 170, 2632-2637.	1.2	32
75	Bosma arhinia microphthalmia syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 189-193.	1.2	30
76	Severe cleidocranial dysplasia and hypophosphatasia in a child with microdeletion of the Câ€terminal region of RUNX2. American Journal of Medical Genetics, Part A, 2010, 152A, 169-174.	1.2	30
77	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	7.6	29
78	Update on the gestational effects of maternal hyperthermia. Birth Defects Research, 2020, 112, 943-952.	1.5	29
79	Immunodeficiency as a component of recognizable syndromes. , 1996, 66, 378-398.		28
80	Clinical and behavioral characteristics in FG syndrome. , 1999, 85, 470-475.		28
81	Familial lissencephaly with cleft palate and severe cerebellar hypoplasia. American Journal of Medical Genetics Part A, 1999, 87, 440-445.	2.4	28
82	Genomic duplication of PTPN11 is an uncommon cause of Noonan syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 2122-2128.	1.2	28
83	Fetal akinesia and associated abnormalities on prenatal MRI. Prenatal Diagnosis, 2011, 31, 484-490.	2.3	28
84	Marshall J. Edwards: Discoverer of maternal hyperthermia as a human teratogen. Birth Defects Research Part A: Clinical and Molecular Teratology, 2005, 73, 857-864.	1.6	27
85	Molecular and neuroimaging findings in pontocerebellar hypoplasia type 2 (PCH2): Is prenatal diagnosis possible?. American Journal of Medical Genetics, Part A, 2010, 152A, 2268-2276.	1.2	26
86	Heat- and Alcohol-Induced Neural Tube Defects: Interactions with Folate in a Golden Hamster Model. Pediatric Research, 1985, 19, 247-251.	2.3	25
87	van den Endeâ€"Gupta syndrome of blepharophimosis, arachnodactyly, and congenital contractures: Clinical delineation and recurrence in brothers. American Journal of Medical Genetics Part A, 2003, 118A, 267-273.	2.4	25
88	CHARGE syndrome from birth to adulthood: An individual reported on from 0 to 33 years. American Journal of Medical Genetics, Part A, 2005, 133A, 344-349.	1.2	25
89	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.1	24
90	Toward a genetic etiology of CHARGE syndrome: I. A systematic scan for submicroscopic deletions. American Journal of Medical Genetics Part A, 2003, 118A, 260-266.	2.4	23

#	Article	IF	Citations
91	MR imaging of the fetal musculoskeletal system. Prenatal Diagnosis, 2012, 32, 205-213.	2.3	23
92	Pregnancy and Birth Outcomes among Women with Idiopathic Thrombocytopenic Purpura. Journal of Pregnancy, 2016, 2016, 1-8.	2.4	23
93	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	2.4	23
94	Behavior of 10 patients with FG syndrome (Opitzâ€“Kaveggia syndrome) and the p.R961W mutation in the MED12 gene. American Journal of Medical Genetics, Part A, 2008, 146A, 3011-3017.	1.2	22
95	Expanding our knowledge of conditions associated with the ASXL gene family. Genome Medicine, 2013, 5, 16.	8.2	22
96	Novel de novo SPOCK1 mutation in a proband with developmental delay, microcephaly and agenesis of corpus callosum. European Journal of Medical Genetics, 2014, 57, 181-184.	1.3	22
97	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	2.4	22
98	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
99	Inside the 8p23.1 duplication syndrome; eight microduplications of likely or uncertain clinical significance. American Journal of Medical Genetics, Part A, 2015, 167, 2052-2064.	1.2	21
100	The NuRD complex and macrocephaly associated neurodevelopmental disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 548-556.	1.6	21
101	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
102	Blaschkolinear skin pigmentary variation due to trisomy 7 mosaicism. American Journal of Medical Genetics Part A, 2000, 95, 281-284.	2.4	20
103	Preaxial hallucal polydactyly as a marker for diabetic embryopathy. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 13-19.	1.6	20
104	Exome sequencing identifies novel mutations in C5orf42 in patients with Joubert syndrome with oralâ€“facialâ€“digital anomalies. Human Genome Variation, 2015, 2, 15045.	0.7	20
105	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. Genetics in Medicine, 2019, 21, 2059-2069.	2.4	20
106	Fetal akinesia/hypokinesia sequence: Prenatal diagnosis and intra-familial variability. Prenatal Diagnosis, 1993, 13, 1011-1019.	2.3	19
107	Behavioral features in young adults with FG syndrome (Opitzâ€“Kaveggia syndrome). American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 477-485.	1.6	19
108	New genetic testing in prenatal diagnosis. Seminars in Fetal and Neonatal Medicine, 2014, 19, 214-219.	2.3	19

#	Article	IF	Citations
109	Mild Brachmann-de Lange syndrome. Phenotypic and developmental characteristics of mildly affected individuals. American Journal of Medical Genetics Part A, 1993, 47, 969-976.	2.4	18
110	P1148A in fibrillin-1 is not a mutation anymore. Nature Genetics, 1997, 15, 12-12.	21.4	17
111	A previously unreported mutation in a Currarino syndrome kindred. American Journal of Medical Genetics, Part A, 2006, 140A, 1923-1930.	1.2	17
112	High-Resolution genomic arrays identify CNVs that phenocopy the chromosome 22q11.2 deletion syndrome. Human Mutation, 2011, 32, 91-97.	2.5	17
113	19q13.32 microdeletion syndrome: Three new cases. European Journal of Medical Genetics, 2014, 57, 654-658.	1.3	17
114	Novel pathogenic <scp>COX</scp>20 variants causing dysarthria, ataxia, and sensory neuropathy. Annals of Clinical and Translational Neurology, 2019, 6, 154-160.	3.7	17
115	Contribution of clinical teratologists and geneticists to the evaluation of the etiology of congenital malformations alleged to be caused by environmental agents: Ionizing radiation, electromagnetic fields, microwaves, radionuclides, and ultrasound. , 1999, 59, 307-313.		16
116	Diaphanospondylodysostosis: Six new cases and exclusion of the candidate genes, PAX1 and MEOX1. American Journal of Medical Genetics, Part A, 2007, 143A, 2292-2302.	1.2	16
117	A study of 534 fetal pathology cases from prenatal diagnosis referrals analyzed from 1989 through 2000. American Journal of Medical Genetics, Part A, 2007, 143A, 3107-3120.	1.2	16
118	2q23.1 microdeletion of the MBD5 gene in a female with seizures, developmental delay and distinct dysmorphic features. European Journal of Medical Genetics, 2012, 55, 59-62.	1.3	16
119	De novo exon 1 missense mutations of SKI and Shprintzenâ€Goldberg syndrome: Two new cases and a clinical review. American Journal of Medical Genetics, Part A, 2014, 164, 676-684.	1.2	16
120	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
121	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143.	1.4	16
122	Congenital Muscular Torticollis and Positional Plagiocephaly. Pediatrics in Review, 2014, 35, 79-87.	0.4	16
123	Tummy Time is Important. Clinical Pediatrics, 2006, 45, 119-121.	0.8	15
124	Clubfeet and associated abnormalities on fetal magnetic resonance imaging. Prenatal Diagnosis, 2012, 32, 822-828.	2.3	15
125	Exome sequencing identifies a mutation in OFD1 in a male with Joubert syndrome, orofaciodigital spectrum anomalies and complex polydactyly. Human Genome Variation, 2016, 3, 15069.	0.7	15
126	Approach to overgrowth syndromes in the genome era. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 483-490.	1.6	15

#	Article	IF	Citations
127	GLUT1 deficiency syndrome as a cause of encephalopathy that includes cognitive disability, treatment-resistant infantile epilepsy and a complex movement disorder. European Journal of Medical Genetics, 2012, 55, 332-334.	1.3	14
128	Proximal variants in <scp>CCND2</scp> associated with microcephaly, short stature, and developmental delay: A case series and review of inverse brain growth phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 2719-2738.	1.2	14
129	Terminal microdeletions of 13q34 chromosome region in patients with intellectual disability: Delineation of an emerging new microdeletion syndrome. Molecular Genetics and Metabolism, 2016, 118, 60-63.	1.1	13
130	Growth, development, and phenotypic spectrum of individuals with deletions of 2q33.1 involving SATB2. Clinical Genetics, 2021, 99, 547-557.	2.0	13
131	Johnson-McMillin syndrome, a neuroectodermal syndrome with conductive hearing loss and microtia: Report of a new case. American Journal of Medical Genetics Part A, 2003, 120A, 400-405.	2.4	12
132	De novo microdeletion of Xp11.3 exclusively encompassing the monoamine oxidase A and B genes in a male infant with episodic hypotonia: A genomics approach to personalized medicine. European Journal of Medical Genetics, 2012, 55, 349-353.	1.3	12
133	When moments matter: Finding answers with rapid exome sequencing. Molecular Genetics & Genomic Medicine, 2020, 8, e1027.	1.2	12
134	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
135	Male genital abnormalities in intrauterine growth restriction. Prenatal Diagnosis, 2012, 32, 427-431.	2.3	11
136	Mutation in the sixth immunoglobulin domain of L1CAM is associated with migrational brain anomalies. Neurology: Genetics, 2015, 1, e34.	1.9	10
137	Spectrum of dolichospondylic dysplasia: Two new patients with distinctive findings. American Journal of Medical Genetics Part A, 2002, 113, 351-361.	2.4	9
138	Early Infantile Epileptic Encephalopathy with a de novo variant in ZEB2 identified by exome sequencing. European Journal of Medical Genetics, 2016, 59, 70-74.	1.3	8
139	Insight Into the Ontogeny of GnRH Neurons From Patients Born Without a Nose. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1538-1551.	3.6	7
140	Cumming Syndrome: report of two additional cases. Pediatric Radiology, 1998, 28, 798-801.	2.0	6
141	De novo copy number variants and parental age: Is there an association?. European Journal of Medical Genetics, 2020, 63, 103829.	1.3	6
142	Gain and loss of TASK3 channel function and its regulation by novel variation cause KCNK9 imprinting syndrome. Genome Medicine, 2022, 14, .	8.2	6
143	Morphogenesis: Re: Clinical, natural history, and imaging information on patients included in reports. American Journal of Medical Genetics Part A, 2003, 119A, 93-93.	2.4	5
144	A syndrome of facial dysmorphism, cubital pterygium, short distal phalanges, swan neck deformity of fingers, and scoliosis. American Journal of Medical Genetics, Part A, 2014, 164, 1035-1040.	1.2	5

#	Article	IF	Citations
145	Non-Cystic Fibrosisâ^'Related Meconium Ileus: GUCY2C-Associated Disease Discovered through Rapid Neonatal Whole-Exome Sequencing. Journal of Pediatrics, 2019, 211, 207-210.	1.8	5
146	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1543-1546.	1.2	5
147	Further delineation of van den Endeâ€Gupta syndrome: Genetic heterogeneity and overlap with congenital heart defects and skeletal malformations syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2136-2149.	1.2	5
148	Variants in PHF8 cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology. Human Genetics and Genomics Advances, 2022, 3, 100102.	1.7	5
149	Diagnosis and Management of Extensive Vertex Birth Molding. Clinical Pediatrics, 2006, 45, 672-678.	0.8	4
150	Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. Molecular Genetics & Genomic Medicine, 2021, 9, e1809.	1.2	4
151	Genetic syndromes associated with immunodeficiency. Immunology and Allergy Clinics of North America, 2002, 22, 261-280.	1.9	3
152	Genetic Syndromes with Evidence of Immune Deficiency. , 2014, , 281-324.		3
153	Genetics of common malformations. European Journal of Medical Genetics, 2014, 57, 353-354.	1.3	3
154	Diagnosis and Treatment of Prostate Cancer: What Americans Can Learn From International Oncologists. American Society of Clinical Oncology Educational Book / ASCO American Society of Clinical Oncology Meeting, 2017, 37, 344-357.	3.8	3
155	A tribute to Bryan D. Hall: Festschrift 2003. American Journal of Medical Genetics Part A, 2003, 123A, 1-4.	2.4	2
156	Growth retardation, intellectual disability, facial anomalies, cataract, thoracic hypoplasia, and skeletal abnormalities: A novel phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2941-2945.	1.2	2
157	The intellectual disabilities evaluation and advice system (IDEAS): Outcome of the first 55 cases. American Journal of Medical Genetics, Part A, 2014, 164, 1102-1117.	1.2	2
158	A case of severe TBCE â€negative hypoparathyroidismâ€retardationâ€dysmorphism syndrome: Case report and literature review. American Journal of Medical Genetics, Part A, 2018, 176, 1768-1772.	1.2	2
159	Reflections on a career in dysmorphology, teratology, and clinical genetics. American Journal of Medical Genetics, Part A, 2021, 185, 2620-2621.	1.2	2
160	Incidence of fetal alcohol syndrome and prevalence of alcoholâ€related neurodevelopmental disorder. Teratology, 1997, 56, 317-326.	1.6	2
161	Incidence of fetal alcohol syndrome and prevalence of alcohol-related neurodevelopmental disorder. , 1997, 56, 317.		1
162	Congenital Anomalies of the Skull. , 2012, , 247-262.		1

#	Article	IF	Citations
163	Twelfth Robert J. Gorlin conference on Dysmorphology. American Journal of Medical Genetics Part A, 2003, 122A, 281-282.	2.4	0
164	Reply to letter to the editor by Lowry et al.: An epidemiological analysis of CHARGE syndrome: Preliminary results from a Canadian study [Issekutz et al., 2005]. American Journal of Medical Genetics, Part A, 2005, 139A, 170-171.	1.2	0
165	Preface. European Journal of Medical Genetics, 2012, 55, 279-280.	1.3	0
166	Abnormal Body Size and Proportion. , 2013, , 1-25.		0
167	Editorial. Seminars in Fetal and Neonatal Medicine, 2014, 19, 137-138.	2.3	0
168	Abnormal Body Size and Proportion. , 2019, , 81-143.		0
169	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
170	Facial Palsy. , 2007, , 85-88.		0