John M Graham

List of Publications by Year
in descending order

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Version: 2024-02-01

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	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
2	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
3	Gain and loss of TASK3 channel function and its regulation by novel variation cause KCNK9 imprinting syndrome. Genome Medicine, 2022, 14, .	8.2	6
4	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
5	Growth, development, and phenotypic spectrum of individuals with deletions of 2q33.1 involving SATB2. Clinical Genetics, 2021, 99, 547-557.	2.0	13
6	Reflections on a career in dysmorphology, teratology, and clinical genetics. American Journal of Medical Genetics, Part A, 2021, 185, 2620-2621.	1.2	2
7	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
8	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
9	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
10	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
11	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
12	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â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in 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
13	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
14	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
15	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
16	When moments matter: Finding answers with rapid exome sequencing. Molecular Genetics & Genomic Medicine, 2020, 8, e1027.	1.2	12
17	De novo copy number variants and parental age: Is there an association?. European Journal of Medical Genetics, 2020, 63, 103829.	1.3	6
18	Update on the gestational effects of maternal hyperthermia. Birth Defects Research, 2020, 112, 943-952.	1.5	29

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19	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
20	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	2.4	22
21	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
22	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	7.6	29
23	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
24	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
25	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
26	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143.	1.4	16
27	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. Genetics in Medicine, 2019, 21, 2059-2069.	2.4	20
28	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
29	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
30	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
31	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
32	Abnormal Body Size and Proportion. , 2019, , 81-143.		0
33	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
34	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.1	24
35	A case of severe TBCEâ€negative hypoparathyroidismâ€retardationâ€dysmorphism syndrome: Case report and literature review. American Journal of Medical Genetics, Part A, 2018, 176, 1768-1772.	1.2	2
36	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

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37	GRIN1 mutation associated with intellectual disability alters NMDA receptor trafficking and function. Journal of Human Genetics, 2017, 62, 589-597.	2.3	81
38	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
39	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
40	Pregnancy and Birth Outcomes among Women with Idiopathic Thrombocytopenic Purpura. Journal of Pregnancy, 2016, 2016, 1-8.	2.4	23
41	KCNK9 imprinting syndromeâ€"further delineation of a possible treatable disorder. American Journal of Medical Genetics, Part A, 2016, 170, 2632-2637.	1.2	32
42	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
43	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
44	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
45	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
46	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
47	Mutation in the sixth immunoglobulin domain of L1CAM is associated with migrational brain anomalies. Neurology: Genetics, 2015, 1, e34.	1.9	10
48	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
49	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
50	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
51	Genetic Syndromes with Evidence of Immune Deficiency. , 2014, , 281-324.		3
52	19q13.32 microdeletion syndrome: Three new cases. European Journal of Medical Genetics, 2014, 57, 654-658.	1.3	17
53	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
54	New genetic testing in prenatal diagnosis. Seminars in Fetal and Neonatal Medicine, 2014, 19, 214-219.	2.3	19

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55	Review of genetic and environmental factors leading to hypospadias. European Journal of Medical Genetics, 2014, 57, 453-463.	1.3	47
56	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
57	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
58	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
59	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
60	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
61	Editorial. Seminars in Fetal and Neonatal Medicine, 2014, 19, 137-138.	2.3	0
62	Genetics of common malformations. European Journal of Medical Genetics, 2014, 57, 353-354.	1.3	3
63	Congenital Muscular Torticollis and Positional Plagiocephaly. Pediatrics in Review, 2014, 35, 79-87.	0.4	16
64	Abnormal Body Size and Proportion. , 2013, , 1-25.		0
65	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	2.5	114
66	Expanding our knowledge of conditions associated with the ASXL gene family. Genome Medicine, 2013, 5, 16.	8.2	22
67	MED12 related disorders. American Journal of Medical Genetics, Part A, 2013, 161, 2734-2740.	1.2	64
68	MR imaging of the fetal musculoskeletal system. Prenatal Diagnosis, 2012, 32, 205-213.	2.3	23
69	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
70	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
71	Clinical review of genetic epileptic encephalopathies. European Journal of Medical Genetics, 2012, 55, 281-298.	1.3	42
72	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

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73	Preface. European Journal of Medical Genetics, 2012, 55, 279-280.	1.3	0
74	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
75	Male genital abnormalities in intrauterine growth restriction. Prenatal Diagnosis, 2012, 32, 427-431.	2.3	11
76	Clubfeet and associated abnormalities on fetal magnetic resonance imaging. Prenatal Diagnosis, 2012, 32, 822-828.	2.3	15
77	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
78	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
79	Congenital Anomalies of the Skull. , 2012, , 247-262.		1
80	Loss-of-Function Mutations in RAB18 Cause Warburg Micro Syndrome. American Journal of Human Genetics, 2011, 88, 499-507.	6.2	158
81	Fetal akinesia and associated abnormalities on prenatal MRI. Prenatal Diagnosis, 2011, 31, 484-490.	2.3	28
82	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
83	High-Resolution genomic arrays identify CNVs that phenocopy the chromosome 22q11.2 deletion syndrome. Human Mutation, 2011, 32, 91-97.	2.5	17
84	Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. Brain, 2011, 134, 143-156.	7.6	200
85	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. Human Mutation, 2010, 31, 1142-1154.	2.5	111
86	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
87	Fetal constraint as a potential risk factor for craniosynostosis. American Journal of Medical Genetics, Part A, 2010, 152A, 394-400.	1.2	79
88	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
89	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
90	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

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91	Elements of morphology: Standard terminology for the periorbital region. American Journal of Medical Genetics, Part A, 2009, 149A, 29-39.	1.2	70
92	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
93	Social function in multiple X and Y chromosome disorders: XXY, XYY, XXYY, XXXY. Developmental Disabilities Research Reviews, 2009, 15, 328-332.	2.9	77
94	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
95	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
96	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
97	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
98	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
99	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
100	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
101	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
102	A recurrent mutation in MED12 leading to R961W causes Opitz-Kaveggia syndrome. Nature Genetics, 2007, 39, 451-453.	21.4	179
103	Facial Palsy. , 2007, , 85-88.		0
104	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
105	Bosma arhinia microphthalmia syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 189-193.	1.2	30
106	A previously unreported mutation in a Currarino syndrome kindred. American Journal of Medical Genetics, Part A, 2006, 140A, 1923-1930.	1.2	17
107	Clinical features and management issues in Mowatâ€“Wilson syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2730-2741.	1.2	89
108	Tummy Time is Important. Clinical Pediatrics, 2006, 45, 119-121.	0.8	15

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109	Diagnosis and Management of Extensive Vertex Birth Molding. Clinical Pediatrics, 2006, 45, 672-678.	0.8	4
110	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. Nature Genetics, 2005, 37, 221-224.	21.4	201
111	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
112	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
113	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
114	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
115	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
116	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
117	Management of deformational plagiocephaly: Repositioning versus orthotic therapy. Journal of Pediatrics, 2005, 146, 258-262.	1.8	173
118	Deformational brachycephaly in supine-sleeping infants. Journal of Pediatrics, 2005, 146, 253-257.	1.8	92
119	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
120	MICRO syndrome: An entity distinct from COFS syndrome. American Journal of Medical Genetics Part A, 2004, 128A, 235-245.	2.4	54
121	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
122	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
123	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
124	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
125	Twelfth Robert J. Gorlin conference on Dysmorphology. American Journal of Medical Genetics Part A, 2003, 122A, 281-282.	2.4	0
126	A tribute to Bryan D. Hall: Festschrift 2003. American Journal of Medical Genetics Part A, 2003, 123A, 1-4.	2.4	2

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127	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
128	Syndromic Immunodeficiencies: Genetic Syndromes Associated with Immune Abnormalities. Critical Reviews in Clinical Laboratory Sciences, 2003, 40, 587-642.	6.1	35
129	Genetic syndromes associated with immunodeficiency. Immunology and Allergy Clinics of North America, 2002, 22, 261-280.	1.9	3
130	The mutational spectrum of brachydactyly type C. American Journal of Medical Genetics Part A, 2002, 112, 291-296.	2.4	82
131	Broad phenotypic spectrum caused by an identical heterozygous CDMP-1 mutation in three unrelated families. , 2002, 117A, 136-142.		45
132	Spectrum of dolichospondylic dysplasia: Two new patients with distinctive findings. American Journal of Medical Genetics Part A, 2002, 113, 351-361.	2.4	9
133	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
134	Kilnefelter Syndrome and Its Variants: An Update and Review for the Primary Pediatrician. Clinical Pediatrics, 2001, 40, 639-651.	0.8	95
135	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
136	A recognizable syndrome within CHARGE association: Hall-Hittner syndrome. American Journal of Medical Genetics Part A, 2001, 99, 120-123.	2.4	59
137	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
138	Blaschkolinear skin pigmentary variation due to trisomy 7 mosaicism. American Journal of Medical Genetics Part A, 2000, 95, 281-284.	2.4	20
139	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
140	Williams-Beuren Syndrome: An Update and Review for the Primary Physician. Clinical Pediatrics, 1999, 38, 189-208.	0.8	78
141	Proteus syndrome: Diagnostic criteria, differential diagnosis, and patient evaluation. American Journal of Medical Genetics Part A, 1999, 84, 389-395.	2.4	374
142	Clinical and behavioral characteristics in FG syndrome. , 1999, 85, 470-475.		28
143	Familial lissencephaly with cleft palate and severe cerebellar hypoplasia. American Journal of Medical Genetics Part A, 1999, 87, 440-445.	2.4	28
144	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

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145	Syndrome of coronal craniosynostosis with brachydactyly and carpal/tarsal coalition due to Pro250Arg mutation in FGFR3 gene. , 1998, 77, 322-329.		61
146	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
147	Teratogen update: Gestational effects of maternal hyperthermia due to febrile illnesses and resultant patterns of defects in humans. , 1998, 58, 209-221.		177
148	Cumming Syndrome: report of two additional cases. Pediatric Radiology, 1998, 28, 798-801.	2.0	6
149	CHARGE Association: An Update and Review for the Primary Pediatrician. Clinical Pediatrics, 1998, 37, 159-173.	0.8	394
150	P1148A in fibrillin-1 is not a mutation anymore. Nature Genetics, 1997, 15, 12-12.	21.4	17
151	Deletions of 20p12 in Alagille syndrome: Frequency and molecular characterization. , 1997, 70, 80-86.		71
152	Evaluation of mental retardation: Recommendations of a consensus conference. , 1997, 72, 468-477.		344
153	Incidence of fetal alcohol syndrome and prevalence of alcohol-related neurodevelopmental disorder. Teratology, 1997, 56, 317-326.	1.6	706
154	Incidence of fetal alcohol syndrome and prevalence of alcohol-related neurodevelopmental disorder. , 1997, 56, 317.		1
155	Incidence of fetal alcohol syndrome and prevalence of alcoholâ€related neurodevelopmental disorder. Teratology, 1997, 56, 317-326.	1.6	2
156	Immunodeficiency as a component of recognizable syndromes. , 1996, 66, 378-398.		28
157	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
158	Longitudinal observations on 15 children with Wiedemannâ€Beckwith syndrome. American Journal of Medical Genetics Part A, 1995, 56, 366-373.	2.4	89
159	Prenatal diagnosis and dysmorphic findings in mosaic trisomy 16. Prenatal Diagnosis, 1994, 14, 257-266.	2.3	36
160	Trisomy 8 mosaicism in chorionic villus sampling: Case report and counselling issues. Prenatal Diagnosis, 1994, 14, 451-454.	2.3	33
161	Compromise of the spinal canal in Proteus syndrome. American Journal of Medical Genetics Part A, 1993, 47, 656-659.	2.4	46
162	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

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163	Congenital diaphragmatic hernia in the Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1993, 47, 1018-1021.	2.4	53
164	Preaxial ray reduction defects as part of valproic acid embryofetopathy. Prenatal Diagnosis, 1993, 13, 909-918.	2.3	60
165	Fetal akinesia/hypokinesia sequence: Prenatal diagnosis and intra-familial variability. Prenatal Diagnosis, 1993, 13, 1011-1019.	2.3	19
166	Central nervous system malformations in the CHARGE association. American Journal of Medical Genetics Part A, 1990, 37, 304-310.	2.4	95
167	Triploidy: Pregnancy complications and clinical findings in seven cases. Prenatal Diagnosis, 1989, 9, 409-419.	2.3	41
168	Heat- and Alcohol-Induced Neural Tube Defects: Interactions with Folate in a Golden Hamster Model. Pediatric Research, 1985, 19, 247-251.	2.3	25
169	Coloboma, congenital heart disease, and choanal atresia with multiple anomalies: CHARGE association. Journal of Pediatrics, 1981, 99, 223-227.	1.8	780
170	Agnathia, situs inversus, and associated malformations. Teratology, 1981, 23, 85-93.	1.6	61