Jan M Friedman

List of Publications by Year
in descending order

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Version: 2024-02-01

158 papers	11,210 citations	38720 50 h-index	31818 101 g-index

169 all docs	169 docs citations	169 times ranked	14340 citing authors

#	Article	IF	Citations
1	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. American Journal of Human Genetics, 2010, 86, 749-764.	2.6	2,325
2	Use of Selective Serotonin-Reuptake Inhibitors in Pregnancy and the Risk of Birth Defects. New England Journal of Medicine, 2007, 356, 2684-2692.	13.9	455
3	Clinical and genetic aspects of neurofibromatosis 1. Genetics in Medicine, 2010, 12, 1-11.	1.1	405
4	Evidence for multi-site closure of the neural tube in humans. American Journal of Medical Genetics Part A, 1993, 47, 723-743.	2.4	364
5	Prevalence of Neurofibromatosis 1 in German Children at Elementary School Enrollment. Archives of Dermatology, 2005, 141, 71-4.	1.7	322
6	Massively Parallel Sequencing: The Next Big Thing in Genetic Medicine. American Journal of Human Genetics, 2009, 85, 142-154.	2.6	308
7	Maternal treatment with opioid analgesics and risk for birth defects. American Journal of Obstetrics and Gynecology, 2011, 204, 314.e1-314.e11.	0.7	291
8	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	1.1	290
9	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	13.9	254
10	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817.	2.6	219
11	Assessment of benign tumor burden by whole-body MRI in patients with neurofibromatosis 1. Neuro-Oncology, 2008, 10, 593-598.	0.6	200
12	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2015, 52, 431-437.	1.5	187
13	Human Chromosome 7: DNA Sequence and Biology. Science, 2003, 300, 767-772.	6.0	185
14	PREVALENCE OF COXSACKIE B VIRUS ANTIBODIES IN PATIENTS WITH JUVENILE DERMATOMYOSITIS. Arthritis and Rheumatism, 1986, 29, 1365-1370.	6.7	181
15	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. American Journal of Human Genetics, 2012, 91, 541-547.	2.6	167
16	A novel recurrent mutation in ATP1A3 causes CAPOS syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 15.	1.2	157
17	Medications in the first trimester of pregnancy: most common exposures and critical gaps in understanding fetal risk. Pharmacoepidemiology and Drug Safety, 2013, 22, 1013-1018.	0.9	140
18	Patterns of Antidepressant Medication Use Among Pregnant Women in a United States Population. Journal of Clinical Pharmacology, 2011, 51, 264-270.	1.0	139

#	Article	IF	Citations
19	â€œI want to know what's in Pandora's boxâ€: Comparing stakeholder perspectives on incidental findings in clinical whole genomic sequencing. American Journal of Medical Genetics, Part A, 2012, 158A, 2519-2525.	0.7	135
20	Decreased bone mineral density in patients with neurofibromatosisÂ1. Osteoporosis International, 2005, 16, 1161-1166.	1.3	130
21	Low risk of solid tumors in persons with Down syndrome. Genetics in Medicine, 2016, 18, 1151-1157.	1.1	129
22	Skeletal abnormalities in neurofibromatosis type 1: Approaches to therapeutic options. American Journal of Medical Genetics, Part A, 2009, 149A, 2327-2338.	0.7	128
23	Specific SSRIs and birth defects: bayesian analysis to interpret new data in the context of previous reports. BMJ, The, 2015, 351, h3190.	3.0	123
24	Acute Nonlymphocytic Leukemia. New England Journal of Medicine, 1979, 301, 1-5.	13.9	120
25	Management of Multiple Sclerosis During Pregnancy and the Reproductive Years. Obstetrics and Gynecology, 2014, 124, 1157-1168.	1.2	109
26	Combined immunodeficiency associated with homozygous MALT1 mutations. Journal of Allergy and Clinical Immunology, 2014, 133, 1458-1462.e7.	1.5	103
27	Growth dynamics of plexiform neurofibromas: a retrospective cohort study of 201 patients with neurofibromatosis 1. Orphanet Journal of Rare Diseases, 2012, 7, 75.	1.2	99
28	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. American Journal of Human Genetics, 2017, 101, 65-74.	2.6	99
29	Perceptions of genetic discrimination among people at risk for Huntington's disease: a cross sectional survey. BMJ: British Medical Journal, 2009, 338, b2175-b2175.	2.4	98
30	Safety of Selective Serotonin Reuptake Inhibitors in Pregnancy. CNS Drugs, 2009, 23, 493-509.	2.7	89
31	Empirical development of improved diagnostic criteria for neurofibromatosis 2. Genetics in Medicine, 2011, 13, 576-581.	1.1	89
32	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	2.6	83
33	The cost and diagnostic yield of exome sequencing for children with suspected genetic disorders: a benchmarking study. Genetics in Medicine, 2018, 20, 1-9.	1.1	79
34	Maternal use of bupropion and risk for congenital heart defects. American Journal of Obstetrics and Gynecology, 2010, 203, 52.e1-52.e6.	0.7	78
35	Clinical and molecular predictors of mortality in neurofibromatosis 2: a UK national analysis of 1192 patients. Journal of Medical Genetics, 2015, 52, 699-705.	1.5	78
36	Genomic newborn screening: public health policy considerations and recommendations. BMC Medical Genomics, 2017, 10, 9.	0.7	78

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37	Exome sequencing for gene discovery in lethal fetal disorders â€“ harnessing the value of extreme phenotypes. Prenatal Diagnosis, 2015, 35, 1005-1009.	1.1	76
38	Subcutaneous neurofibromas are associated with mortality in neurofibromatosis 1: A cohort study of 703 patients. American Journal of Medical Genetics, Part A, 2005, 132A, 49-53.	0.7	73
39	Parental perceived value of a diagnosis for intellectual disability (ID): A qualitative comparison of families with and without a diagnosis for their child's ID. American Journal of Medical Genetics, Part A, 2009, 149A, 2393-2402.	0.7	69
40	Novel deletions of 14q11.2 associated with developmental delay, cognitive impairment and similar minor anomalies in three children. Journal of Medical Genetics, 2007, 44, 556-561.	1.5	68
41	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. Brain, 2019, 142, 542-559.	3.7	67
42	The composition and capacity of the clinical genetics workforce in high-income countries: a scoping review. Genetics in Medicine, 2020, 22, 1437-1449.	1.1	64
43	Safety of Selective Serotonin Reuptake Inhibitors in Pregnancy: A Review of Current Evidence. CNS Drugs, 2016, 30, 499-515.	2.7	61
44	Benign whole body tumor volume is a risk factor for malignant peripheral nerve sheath tumors in neurofibromatosis type 1. Journal of Neuro-Oncology, 2014, 116, 307-313.	1.4	59
45	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unitâ€"successes and challenges. European Journal of Pediatrics, 2019, 178, 1207-1218.	1.3	59
46	Osteopoikilosis, short stature and mental retardation as key features of a new microdeletion syndrome on 12q14. Journal of Medical Genetics, 2007, 44, 264-268.	1.5	58
47	Congenital diaphragmatic hernia, coarse facies, and acral hypoplasia: Fryns syndrome. American Journal of Medical Genetics Part A, 1989, 32, 93-99.	2.4	56
48	Value for Money? Array Genomic Hybridization for Diagnostic Testing for Genetic Causes of Intellectual Disability. American Journal of Human Genetics, 2010, 86, 765-772.	2.6	56
49	Approaches to Treating NF1 Tibial Pseudarthrosis. Journal of Pediatric Orthopaedics, 2013, 33, 269-275.	0.6	55
50	The Sensitivity of Massively Parallel Sequencing for Detecting Candidate Infectious Agents Associated with Human Tissue. PLoS ONE, 2011, 6, e19838.	1.1	55
51	Selective Serotonin-Reuptake Inhibitors and Persistent Pulmonary Hypertension of the Newborn. New England Journal of Medicine, 2006, 354, 2188-2190.	13.9	54
52	Prospective study of methylenetetrahydrofolate reductase (MTHFR) variant C677T and risk of all-cause and cardiovascular disease mortality among 6000 US adults. American Journal of Clinical Nutrition, 2012, 95, 1245-1253.	2.2	51
53	Paternalism and the ACMG recommendations on genomic incidental findings: patients seen but not heard. Genetics in Medicine, 2013, 15, 751-752.	1.1	50
54	Patterns of associations of clinical features in neurofibromatosisÂ1 (NF1). Human Genetics, 2003, 112, 289-297.	1.8	49

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55	A different approach to validating screening assays for developmental toxicity. Birth Defects Research Part B: Developmental and Reproductive Toxicology, 2010, 89, 526-530.	1.4	48
56	Exposureâ€Based Validation List for Developmental Toxicity Screening Assays. Birth Defects Research Part B: Developmental and Reproductive Toxicology, 2014, 101, 423-428.	1.4	48
57	Immunogenetic studies of juvenile dermatomyositis. III. Study of antibody to organ-specific and nuclear antigens. Arthritis and Rheumatism, 1985, 28, 151-157.	6.7	47
58	Fetal karyotype following ascertainment of fetal anomalies by ultrasound. Prenatal Diagnosis, 1987, 7, 551-555.	1.1	47
59	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. Journal of Medical Genetics, 2010, 47, 155-161.	1.5	47
60	Impact of BRCA mutations on female fertility and offspring sex ratio. American Journal of Human Biology, 2010, 22, 201-205.	0.8	45
61	Safe lists for medications in pregnancy: inadequate evidence base and inconsistent guidance from Webâ€based information, 2011. Pharmacoepidemiology and Drug Safety, 2013, 22, 324-328.	0.9	42
62	Genome-wide sequencing in acutely ill infants: genomic medicineâ€™s critical application?. Genetics in Medicine, 2019, 21, 498-504.	1.1	42
63	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	3.7	38
64	Beyond the patient: The broader impact of genetic discrimination among individuals at risk of Huntington disease. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 217-226.	1.1	37
65	Recurrent trisomy 21 in a couple with a child presenting trisomy 21 mosaicism and maternal uniparental disomy for chromosome 21 in the euploid cell line. American Journal of Medical Genetics Part A, 2000, 94, 35-41.	2.4	35
66	Single exon-resolution targeted chromosomal microarray analysis of known and candidate intellectual disability genes. European Journal of Human Genetics, 2014, 22, 792-800.	1.4	35
67	A distinct neurodevelopmental syndrome with intellectual disability, autism spectrum disorder, characteristic facies, and macrocephaly is caused by defects in CHD8. Journal of Human Genetics, 2019, 64, 271-280.	1.1	35
68	The cost trajectory of the diagnostic care pathway for children with suspected genetic disorders. Genetics in Medicine, 2020, 22, 292-300.	1.1	35
69	Anatomic correlates of ultrasonographic prenatal diagnosis. Prenatal Diagnosis, 1986, 6, 51-61.	1.1	34
70	The importance of genetic counselling in genome-wide sequencing. Nature Reviews Genetics, 2018, 19, 735-736.	7.7	34
71	Mutations in ILK, encoding integrin-linked kinase, are associated with arrhythmogenic cardiomyopathy. Translational Research, 2019, 208, 15-29.	2.2	33
72	The principles of teratology: Are they still true?. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 766-768.	1.6	32

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73	Non-optic glioma in adults and children with neurofibromatosis 1. Orphanet Journal of Rare Diseases, 2017, 12, 34.	1.2	31
74	Aspartylglycosamine is a biomarker for NGLY1-CDDG, a congenital disorder of deglycosylation. Molecular Genetics and Metabolism, 2019, 127, 368-372.	0.5	31
75	Different Patterns of Mast Cells Distinguish Diffuse from Encapsulated Neurofibromas in Patients with Neurofibromatosis 1. Journal of Histochemistry and Cytochemistry, 2011, 59, 584-590.	1.3	29
76	Key Implications of Data Sharing in Pediatric Genomics. JAMA Pediatrics, 2018, 172, 476.	3.3	29
77	Genetic Mosaics and the Germ Line Lineage. Genes, 2015, 6, 216-237.	1.0	28
78	Genome-wide sequencing as a first-tier screening test for short tandem repeat expansions. Genome Medicine, 2021, 13, 126.	3.6	27
79	Comparison of genome-wide array genomic hybridization platforms for the detection of copy number variants in idiopathic mental retardation. BMC Medical Genomics, 2011, 4, 25.	0.7	26
80	Assessing an Interactive Online Tool to Support Parents' Genomic Testing Decisions. Journal of Genetic Counseling, 2019, 28, 10-17.	0.9	26
81	Growth charts for young children with neurofibromatosis 1 (NF1). , 2000, 92, 224-227.		25
82	Serial MRIs provide novel insight into natural history of optic pathway gliomas in patients with neurofibromatosis 1. Orphanet Journal of Rare Diseases, 2018, 13, 62.	1.2	25
83	Kallmann syndrome associated with choanal atresia. Clinical Genetics, 1987, 31, 224-227.	1.0	24
84	Effect of vitamin D3 treatment on bone density in neurofibromatosis 1 patients: A retrospective clinical study. Joint Bone Spine, 2013, 80, 315-319.	0.8	24
85	Clinical objectives in medical genetics for undergraduate medical students. Genetics in Medicine, 1998, 1, 54-55.	1.1	22
86	Toward the diagnosis of rare childhood genetic diseases: what do parents value most?. European Journal of Human Genetics, 2021, 29, 1491-1501.	1.4	22
87	Straglr: discovering and genotyping tandem repeat expansions using whole genome long-read sequences. Genome Biology, 2021, 22, 224.	3.8	22
88	A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7. European Journal of Medical Genetics, 2009, 52, 436-439.	0.7	21
89	Comparing the ability of OPTION12 and OPTION5 to assess shared decision-making in genetic counselling.. Patient Education and Counseling, 2016, 99, 1717-1723.	1.0	21
90	Hepatocellular carcinoma in a child with familial Russell-Silver syndrome. American Journal of Medical Genetics Part A, 1988, 31, 909-914.	2.4	19

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91	Recurrent subacute post-viral onset of ataxia associated with a PRF1 mutation. European Journal of Human Genetics, 2013, 21, 1232-1239.	1.4	19
92	Genetic heterogeneity in spondyloepiphyseal dysplasia congenita. American Journal of Medical Genetics Part A, 1984, 18, 311-320.	2.4	18
93	The Genomic Consultation Service: A clinical service designed to improve patient selection for genomeâ€wide sequencing in British Columbia. Molecular Genetics & Genomic Medicine, 2018, 6, 592-600.	0.6	18
94	Prevalence of selected genomic deletions and duplications in a <scp>F</scp>renchâ€“<scp>C</scp>anadian populationâ€based sample of newborns. Molecular Genetics & Genomic Medicine, 2013, 1, 87-97.	0.6	17
95	Update from the 2013 international neurofibromatosis conference. American Journal of Medical Genetics, Part A, 2014, 164, 2969-2978.	0.7	17
96	Parentsâ€™ Perspectives on Supporting Their Decision Making in Genomeâ€Wide Sequencing. Journal of Nursing Scholarship, 2016, 48, 265-275.	1.1	17
97	Growth in neurofibromatosis 1 microdeletion patients. Clinical Genetics, 2016, 89, 351-354.	1.0	17
98	Current controversies in prenatal diagnosis 2: should a fetal exome be used in the assessment of a dysmorphic or malformed fetus?. Prenatal Diagnosis, 2016, 36, 15-19.	1.1	16
99	Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability. BMC Genomics, 2017, 18, 403.	1.2	15
100	A novel mouse model for pyridoxine-dependent epilepsy due to antiquitin deficiency. Human Molecular Genetics, 2020, 29, 3266-3284.	1.4	15
101	Specificity of acquired clonal chromosome abnormalities in new zealand black mice. International Journal of Cancer, 1978, 21, 505-510.	2.3	14
102	A translocation t(6;7)(p11â€“p12;q22) associated with autism and mental retardation: localization and identification of candidate genes at the breakpoints. Psychiatric Genetics, 2008, 18, 101-109.	0.6	14
103	A novel de novo 1.1 Mb duplication of 17q21.33 associated with cognitive impairment and other anomalies. American Journal of Medical Genetics, Part A, 2009, 149A, 1257-1262.	0.7	14
104	Factors associated with experiences of genetic discrimination among individuals at risk for huntington disease. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 19-27.	1.1	14
105	ABCDXXX: The obscenity of postmarketing surveillance for teratogenic effects. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 670-676.	1.6	14
106	Intragenic CNVs for epigenetic regulatory genes in intellectual disability: Survey identifies pathogenic and benign single exon changes. American Journal of Medical Genetics, Part A, 2016, 170, 2916-2926.	0.7	14
107	Life-Threatening Status Asthmaticus at 12.5 Weeksâ€™ Gestation. Chest, 1991, 100, 285-286.	0.4	13
108	Compound heterozygous TRPV4 mutations in two siblings with a complex phenotype including severe intellectual disability and neuropathy. American Journal of Medical Genetics, Part A, 2017, 173, 3087-3092.	0.7	13

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109	De novo pathogenic DNM1L variant in a patient diagnosed with atypical hereditary sensory and autonomic neuropathy. Molecular Genetics & Genomic Medicine, 2019, 7, e00961.	0.6	12
110	Atypical antipsychotic use during pregnancy and birth defect risk: National Birth Defects Prevention Study, 1997â€“2011. Schizophrenia Research, 2020, 215, 81-88.	1.1	12
111	Early primary tooth eruption in neurofibromatosis 1 individuals. European Journal of Oral Sciences, 2007, 115, 425-426.	0.7	10
112	A case of splenomegaly in CBL syndrome. European Journal of Medical Genetics, 2017, 60, 374-379.	0.7	10
113	After genomic testing results: Parentsâ€™ longâ€term views. Journal of Genetic Counseling, 2022, 31, 82-95.	0.9	10
114	Quantitative associations of scalp and body subcutaneous neurofibromas with internal plexiform tumors in neurofibromatosis 1. American Journal of Medical Genetics, Part A, 2015, 167, 1518-1524.	0.7	9
115	Exome Sequencing and Clinical Diagnosis. JAMA - Journal of the American Medical Association, 2020, 324, 627.	3.8	9
116	Utilization and uptake of clinical genetics services in high-income countries: A scoping review. Health Policy, 2021, 125, 877-887.	1.4	9
117	Addendum: Sartan treatment during pregnancy. Birth Defects Research Part A: Clinical and Molecular Teratology, 2005, 73, 904-905.	1.6	8
118	Big risks in small groups: The difference between epidemiology and counselling. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 720-724.	1.6	8
119	Individual DNA samples and health information sold by 23andMe. Genetics in Medicine, 2016, 18, 305-306.	1.1	8
120	Evaluating the use of parental reports to estimate health care resource utilization in children with suspected genetic disorders. Journal of Evaluation in Clinical Practice, 2018, 24, 416-422.	0.9	8
121	Somatic mosaicism detected by genome-wide sequencing in 500 parentâ€“child trios with suspected genetic disease: clinical and genetic counseling implications. Journal of Physical Education and Sports Management, 2021, 7, a006125.	0.5	8
122	Prevalence of dental caries in children with neurofibromatosis 1. Clinical Oral Investigations, 2010, 14, 479-480.	1.4	7
123	Use of Affymetrix Mapping Arrays in the Diagnosis of Gene Copy Number Variation. Current Protocols in Human Genetics, 2008, 59, Unit 8.13.	3.5	7
124	Cerebral palsy and related neuromotor disorders: Overview of genetic and genomic studies. Molecular Genetics and Metabolism, 2022, 137, 399-419.	0.5	7
125	Genome-wide sequencing and the clinical diagnosis of genetic disease: The CAUSES study. Human Genetics and Genomics Advances, 2022, 3, 100108.	1.0	7
126	Congenital cataracts in mother, sister, and son of a patient with Hallermann-Streiff syndrome: Coincidence or clue?. American Journal of Medical Genetics Part A, 1991, 41, 500-502.	2.4	6

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127	Neurofibromin haploinsufficiency results in altered spermatogenesis in a mouse model of neurofibromatosis type 1. PLoS ONE, 2018, 13, e0208835.	1.1	6
128	MRI based volumetric measurements of vestibular schwannomas in patients with neurofibromatosis type 2: comparison of three different software tools. Scientific Reports, 2020, 10, 11541.	1.6	6
129	Prescription opioid use during pregnancy and risk for preterm birth or term low birthweight. Journal of Opioid Management, 2021, 17, 215-225.	0.2	6
130	Where is genetic medicine headed? Exploring the perspectives of Canadian genetic professionals on future trends using the Delphi method. European Journal of Human Genetics, 2022, 30, 496-504.	1.4	6
131	A personalized genomic results e-booklet, co-designed and pilot-tested by families. PEC Innovation, 2022, 1, 100039.	0.3	6
132	S100B and neurofibromin immunostaining and Xâ€inactivation patterns of laserâ€microdissected cells indicate a multicellular origin of some NF1â€associated neurofibromas. Journal of Neuroscience Research, 2011, 89, 1451-1460.	1.3	5
133	Lifeâ€history chronicle for a patient with the recently described chromosome 4q21 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2606-2609.	0.7	5
134	Utilization of telehealth in paediatric genome-wide sequencing: Health services implementation issues in the CAUSES Study. Journal of Telemedicine and Telecare, 2023, 29, 318-327.	1.4	5
135	Using genomics for birth defects epidemiology: Can epigenetics cut the GxE gordian knot?. Birth Defects Research Part A: Clinical and Molecular Teratology, 2011, 91, 986-989.	1.6	4
136	Editorial In Bed with The Devil: Recognizing Human Teratogenic Exposures. Birth Defects Research, 2017, 109, 1407-1413.	0.8	4
137	Strabismus in Children With Intellectual Disability: Part of a Broader Motor Control Phenotype?. Pediatric Neurology, 2019, 100, 87-91.	1.0	4
138	Renpenning syndrome in a female. American Journal of Medical Genetics, Part A, 2020, 182, 498-503.	0.7	4
139	White matter is increased in the brains of adults with neurofibromatosis 1. Orphanet Journal of Rare Diseases, 2022, 17, 115.	1.2	4
140	Exome/Genome-Wide Testing in Newborn Screening: A Proportionate Path Forward. Frontiers in Genetics, 0, 13, .	1.1	4
141	ACTIVE JUVENILE DERMATOMYOSITIS (JDMS) IS ASSOCIATED WITH COMPLEMENT AND COAGULATION ACTIVATION, AND INCREASED TITERS TO ANTINUCLEAR (ANA) AND COXSACKIE B VIRAL (COX-B) ANTIGENS. Pediatric Research, 1984, 18, 262A-262A.	1.1	3
142	Genetics in Medicine and informatics for the genetic clinician. Genetics in Medicine, 1998, 1, 52-52.	1.1	2
143	Exome Sequencing as Part of a Multidisciplinary Approach to Diagnosisâ€"Reply. JAMA - Journal of the American Medical Association, 2020, 324, 2445.	3.8	2
144	Alterations in brain morphology by MRI in adults with neurofibromatosis 1. Orphanet Journal of Rare Diseases, 2021, 16, 462.	1.2	2

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145	Emerging issues in teratology: An introduction. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 147-149.	0.7	1
146	Clinical Teratology. , 2013, , 1-39.		1
147	Antineoplastic drugs. , 2015, , 373-399.		1
148	Secondary biogenic amine deficiencies: genetic etiology, therapeutic interventions, and clinical effects. Neurogenetics, 2021, 22, 251-262.	0.7	1
149	Molecular Basis of Cardiovascular Abnormalities in NF1. , 2012, , 353-366.		1
150	Maternal serum Î±-tetoprotein in pregnancy. American Journal of Obstetrics and Gynecology, 1990, 163, 692-693.	0.7	0
151	OTIS special issue preface. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 569-569.	1.6	0
152	Controversy and debate on clinical genomics sequencingâ€"paper 2: clinical genome-wide sequencing: don't throw out the baby with theÂbathwater!. Journal of Clinical Epidemiology, 2017, 92, 7-10.	2.4	0
153	Controversy and debate on clinical genomics sequencingâ€"paper 4: clinical genome-wide sequencing: response to Wilson, Miller, and Rousseau. Journal of Clinical Epidemiology, 2017, 92, 13-15.	2.4	0
154	Mutations in Kv7.5 Channels Associated with Intellectual Disability or Epileptic Encephalopathy. Biophysical Journal, 2018, 114, 123a.	0.2	0
155	Assessment of Case Reports and Clinical Series. , 2018, , 389-396.		0
156	Clinical Teratology. , 2019, , 15-60.		0
157	Physician and Patient Education. , 1994, , 293-295.		0
158	Developmental Toxicology and Teratology. , 0, , 9-17.		0