

Jan M Friedman

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

151
papers

8,436
citations

45
h-index

90
g-index

169
ext. papers

9,905
ext. citations

7
avg, IF

5.68
L-index

#	Paper	IF	Citations
151	White matter is increased in the brains of adults with neurofibromatosis 1.. <i>Orphanet Journal of Rare Diseases</i> , 2022 , 17, 115	4.2	0
150	Genome-wide Sequencing and the Clinical Diagnosis of Genetic Disease: The CAUSES Study. <i>Human Genetics and Genomics Advances</i> , 2022 , 100108	0.8	0
149	A personalized genomic results e-booklet, co-designed and pilot-tested by families. <i>PEC Innovation</i> , 2022 , 100039		0
148	Alterations in brain morphology by MRI in adults with neurofibromatosis 1. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 462	4.2	1
147	Toward the diagnosis of rare childhood genetic diseases: what do parents value most?. <i>European Journal of Human Genetics</i> , 2021 , 29, 1491-1501	5.3	2
146	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. <i>Genetics in Medicine</i> , 2021 , 23, 1506-1513	8.1	43
145	After genomic testing results: Parents' long-term views. <i>Journal of Genetic Counseling</i> , 2021 ,	2.5	1
144	Secondary biogenic amine deficiencies: genetic etiology, therapeutic interventions, and clinical effects. <i>Neurogenetics</i> , 2021 , 22, 251-262	3	
143	Utilization and uptake of clinical genetics services in high-income countries: A scoping review. <i>Health Policy</i> , 2021 , 125, 877-887	3.2	1
142	Prescription opioid use during pregnancy and risk for preterm birth or term low birthweight. <i>Journal of Opioid Management</i> , 2021 , 17, 215-225	0.8	0
141	Straglr: discovering and genotyping tandem repeat expansions using whole genome long-read sequences. <i>Genome Biology</i> , 2021 , 22, 224	18.3	2
140	Genome-wide sequencing as a first-tier screening test for short tandem repeat expansions. <i>Genome Medicine</i> , 2021 , 13, 126	14.4	4
139	Utilization of telehealth in paediatric genome-wide sequencing: Health services implementation issues in the CAUSES Study. <i>Journal of Telemedicine and Telecare</i> , 2021 , 1357633X20982737	6.8	2
138	The composition and capacity of the clinical genetics workforce in high-income countries: a scoping review. <i>Genetics in Medicine</i> , 2020 , 22, 1437-1449	8.1	16
137	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. <i>Brain</i> , 2020 , 143, 55-68	11.2	18
136	Renpenning syndrome in a female. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 498-503	2.5	1
135	Atypical antipsychotic use during pregnancy and birth defect risk: National Birth Defects Prevention Study, 1997-2011. <i>Schizophrenia Research</i> , 2020 , 215, 81-88	3.6	6

134	A novel mouse model for pyridoxine-dependent epilepsy due to antiquitin deficiency. <i>Human Molecular Genetics</i> , 2020 , 29, 3266-3284	5.6	5
133	MRI based volumetric measurements of vestibular schwannomas in patients with neurofibromatosis type 2: comparison of three different software tools. <i>Scientific Reports</i> , 2020 , 10, 11541	4.9	1
132	Exome Sequencing and Clinical Diagnosis. <i>JAMA - Journal of the American Medical Association</i> , 2020 ,	27.4	6
131	The cost trajectory of the diagnostic care pathway for children with suspected genetic disorders. <i>Genetics in Medicine</i> , 2020 , 22, 292-300	8.1	14
130	Exome Sequencing as Part of a Multidisciplinary Approach to Diagnosis-Reply. <i>JAMA - Journal of the American Medical Association</i> , 2020 , 324, 2445-2446	27.4	1
129	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. <i>Brain</i> , 2019 , 142, 542-559	11.2	41
128	A distinct neurodevelopmental syndrome with intellectual disability, autism spectrum disorder, characteristic facies, and macrocephaly is caused by defects in CHD8. <i>Journal of Human Genetics</i> , 2019 , 64, 271-280	4.3	20
127	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unit-successes and challenges. <i>European Journal of Pediatrics</i> , 2019 , 178, 1207-1218	4.1	29
126	Strabismus in Children With Intellectual Disability: Part of a Broader Motor Control Phenotype?. <i>Pediatric Neurology</i> , 2019 , 100, 87-91	2.9	1
125	Mutations in ILK, encoding integrin-linked kinase, are associated with arrhythmogenic cardiomyopathy. <i>Translational Research</i> , 2019 , 208, 15-29	11	14
124	Genome-wide sequencing in acutely ill infants: genomic medicine's critical application?. <i>Genetics in Medicine</i> , 2019 , 21, 498-504	8.1	28
123	Aspartylglycosamine is a biomarker for NGLY1-CDDG, a congenital disorder of deglycosylation. <i>Molecular Genetics and Metabolism</i> , 2019 , 127, 368-372	3.7	12
122	De novo pathogenic DNMT1L variant in a patient diagnosed with atypical hereditary sensory and autonomic neuropathy. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e00961	2.3	8
121	Clinical Teratology 2019 , 15-60		
120	The cost and diagnostic yield of exome sequencing for children with suspected genetic disorders: a benchmarking study. <i>Genetics in Medicine</i> , 2018 , 20, 1013-1021	8.1	55
119	Serial MRIs provide novel insight into natural history of optic pathway gliomas in patients with neurofibromatosis 1. <i>Orphanet Journal of Rare Diseases</i> , 2018 , 13, 62	4.2	17
118	Evaluating the use of parental reports to estimate health care resource utilization in children with suspected genetic disorders. <i>Journal of Evaluation in Clinical Practice</i> , 2018 , 24, 416-422	2.5	5
117	Key Implications of Data Sharing in Pediatric Genomics. <i>JAMA Pediatrics</i> , 2018 , 172, 476-481	8.3	22

116 Assessment of Case Reports and Clinical Series **2018**, 389-396

115 Assessing an Interactive Online Tool to Support Parents' Genomic Testing Decisions. *Journal of Genetic Counseling*, **2018**, 28, 10 2.5 13

114 Neurofibromin haploinsufficiency results in altered spermatogenesis in a mouse model of neurofibromatosis type 1. *PLoS ONE*, **2018**, 13, e0208835 3.7 2

113 The importance of genetic counselling in genome-wide sequencing. *Nature Reviews Genetics*, **2018**, 19, 735-736 30.1 21

112 The Genomic Consultation Service: A clinical service designed to improve patient selection for genome-wide sequencing in British Columbia. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 592 2.3 16

111 Genomic newborn screening: public health policy considerations and recommendations. *BMC Medical Genomics*, **2017**, 10, 9 3.7 44

110 Non-optic glioma in adults and children with neurofibromatosis 1. *Orphanet Journal of Rare Diseases*, **2017**, 12, 34 4.2 21

109 A case of splenomegaly in CBL syndrome. *European Journal of Medical Genetics*, **2017**, 60, 374-379 2.6 5

108 Controversy and debate on clinical genomics sequencing-paper 2: clinical genome-wide sequencing: don't throw out the baby with the bathwater!. *Journal of Clinical Epidemiology*, **2017**, 92, 7-10 5.7

107 Controversy and debate on clinical genomics sequencing-paper 4: clinical genome-wide sequencing: response to Wilson, Miller, and Rousseau. *Journal of Clinical Epidemiology*, **2017**, 92, 13-15 5.7

106 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 2.5 7

105 ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. *American Journal of Human Genetics*, **2017**, 101, 1021-1033 11 50

104 Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability. *BMC Genomics*, **2017**, 18, 403 4.5 9

103 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 11 58

102 Individual DNA samples and health information sold by 23andMe. *Genetics in Medicine*, **2016**, 18, 305-6 8.1 7

101 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 2.5 11

100 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, 15-9 3.2 15

99 Parents' Perspectives on Supporting Their Decision Making in Genome-Wide Sequencing. *Journal of Nursing Scholarship*, **2016**, 48, 265-75 3.6 14

98	Growth in neurofibromatosis 1 microdeletion patients. <i>Clinical Genetics</i> , 2016 , 89, 351-4	4	14
97	Low risk of solid tumors in persons with Down syndrome. <i>Genetics in Medicine</i> , 2016 , 18, 1151-1157	8.1	83
96	Exome Sequencing and the Management of Neurometabolic Disorders. <i>New England Journal of Medicine</i> , 2016 , 374, 2246-55	59.2	197
95	Comparing the ability of OPTION(12) and OPTION(5) to assess shared decision-making in genetic counselling. <i>Patient Education and Counseling</i> , 2016 , 99, 1717-23	3.1	12
94	Safety of Selective Serotonin Reuptake Inhibitors in Pregnancy: A Review of Current Evidence. <i>CNS Drugs</i> , 2016 , 30, 499-515	6.7	43
93	Exome sequencing for gene discovery in lethal fetal disorders--harnessing the value of extreme phenotypes. <i>Prenatal Diagnosis</i> , 2015 , 35, 1005-9	3.2	63
92	Specific SSRIs and birth defects: Bayesian analysis to interpret new data in the context of previous reports. <i>BMJ, The</i> , 2015 , 351, h3190	5.9	96
91	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. <i>Journal of Medical Genetics</i> , 2015 , 52, 431-7	5.8	137
90	Clinical and molecular predictors of mortality in neurofibromatosis 2: a UK national analysis of 1192 patients. <i>Journal of Medical Genetics</i> , 2015 , 52, 699-705	5.8	52
89	Quantitative associations of scalp and body subcutaneous neurofibromas with internal plexiform tumors in neurofibromatosis 1. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167, 1518-24	2.5	6
88	Genetic mosaics and the germ line lineage. <i>Genes</i> , 2015 , 6, 216-37	4.2	22
87	Antineoplastic drugs 2015 , 373-399		1
86	Combined immunodeficiency associated with homozygous MALT1 mutations. <i>Journal of Allergy and Clinical Immunology</i> , 2014 , 133, 1458-62, 1462.e1-7	11.5	91
85	Single exon-resolution targeted chromosomal microarray analysis of known and candidate intellectual disability genes. <i>European Journal of Human Genetics</i> , 2014 , 22, 792-800	5.3	30
84	Update from the 2013 International Neurofibromatosis Conference. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 2969-78	2.5	11
83	A novel recurrent mutation in ATP1A3 causes CAPOS syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 15	4.2	121
82	FORGE Canada Consortium: outcomes of a 2-year national rare-disease gene-discovery project. <i>American Journal of Human Genetics</i> , 2014 , 94, 809-17	11	174
81	Exposure-based validation list for developmental toxicity screening assays. <i>Birth Defects Research Part B: Developmental and Reproductive Toxicology</i> , 2014 , 101, 423-8		35

80	Management of multiple sclerosis during pregnancy and the reproductive years: a systematic review. <i>Obstetrics and Gynecology</i> , 2014 , 124, 1157-1168	4.9	87
79	Benign whole body tumor volume is a risk factor for malignant peripheral nerve sheath tumors in neurofibromatosis type 1. <i>Journal of Neuro-Oncology</i> , 2014 , 116, 307-13	4.8	49
78	Clinical Teratology 2013 , 1-39		1
77	Recurrent subacute post-viral onset of ataxia associated with a PRF1 mutation. <i>European Journal of Human Genetics</i> , 2013 , 21, 1232-9	5.3	14
76	Paternalism and the ACMG recommendations on genomic incidental findings: patients seen but not heard. <i>Genetics in Medicine</i> , 2013 , 15, 751-2	8.1	41
75	Effet de la vitamine D3 sur la densité osseuse chez les patients porteurs d'une neurofibromatose 1: une étude clinique rétrospective. <i>Revue Du Rhumatisme (Edition Francaise)</i> , 2013 , 80, 262-266	0.1	
74	Effect of vitamin D3 treatment on bone density in neurofibromatosis 1 patients: a retrospective clinical study. <i>Joint Bone Spine</i> , 2013 , 80, 315-9	2.9	20
73	Approaches to treating NF1 tibial pseudarthrosis: consensus from the Children's Tumor Foundation NF1 Bone Abnormalities Consortium. <i>Journal of Pediatric Orthopaedics</i> , 2013 , 33, 269-75	2.4	42
72	Prevalence of selected genomic deletions and duplications in a French-Canadian population-based sample of newborns. <i>Molecular Genetics & Genomic Medicine</i> , 2013 , 1, 87-97	2.3	13
71	Safe lists for medications in pregnancy: inadequate evidence base and inconsistent guidance from Web-based information, 2011. <i>Pharmacoepidemiology and Drug Safety</i> , 2013 , 22, 324-8	2.6	35
70	Medications in the first trimester of pregnancy: most common exposures and critical gaps in understanding fetal risk. <i>Pharmacoepidemiology and Drug Safety</i> , 2013 , 22, 1013-8	2.6	103
69	Beyond the patient: the broader impact of genetic discrimination among individuals at risk of Huntington disease. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012 , 159B, 217-26	3.5	32
68	"I want to know what's in Pandora's Box": comparing stakeholder perspectives on incidental findings in clinical whole genomic sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 2519-25	2.5	128
67	Life-history chronicle for a patient with the recently described chromosome 4q21 microdeletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 2606-9	2.5	4
66	Growth dynamics of plexiform neurofibromas: a retrospective cohort study of 201 patients with neurofibromatosis 1. <i>Orphanet Journal of Rare Diseases</i> , 2012 , 7, 75	4.2	75
65	Exome sequencing and functional validation in zebrafish identify GTDC2 mutations as a cause of Walker-Warburg syndrome. <i>American Journal of Human Genetics</i> , 2012 , 91, 541-7	11	144
64	Prospective study of methylenetetrahydrofolate reductase (MTHFR) variant C677T and risk of all-cause and cardiovascular disease mortality among 6000 US adults. <i>American Journal of Clinical Nutrition</i> , 2012 , 95, 1245-53	7	40
63	Molecular Basis of Cardiovascular Abnormalities in NF1 2012 , 353-366		1

62 Massively Parallel Sequencing **2011**, 114-134

61	Empirical development of improved diagnostic criteria for neurofibromatosis 2. <i>Genetics in Medicine</i> , 2011 , 13, 576-81	8.1	67
60	Maternal treatment with opioid analgesics and risk for birth defects. <i>American Journal of Obstetrics and Gynecology</i> , 2011 , 204, 314.e1-11	6.4	235
59	Comparison of genome-wide array genomic hybridization platforms for the detection of copy number variants in idiopathic mental retardation. <i>BMC Medical Genomics</i> , 2011 , 4, 25	3.7	25
58	S100B and neurofibromin immunostaining and X-inactivation patterns of laser-microdissected cells indicate a multicellular origin of some NF1-associated neurofibromas. <i>Journal of Neuroscience Research</i> , 2011 , 89, 1451-60	4.4	5
57	Factors associated with experiences of genetic discrimination among individuals at risk for Huntington disease. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2011 , 156B, 19-27	3.5	14
56	Emerging issues in teratology: an introduction. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2011 , 157C, 147-9	3.1	1
55	Patterns of antidepressant medication use among pregnant women in a United States population. <i>Journal of Clinical Pharmacology</i> , 2011 , 51, 264-70	2.9	122
54	Different patterns of mast cells distinguish diffuse from encapsulated neurofibromas in patients with neurofibromatosis 1. <i>Journal of Histochemistry and Cytochemistry</i> , 2011 , 59, 584-90	3.4	24
53	The sensitivity of massively parallel sequencing for detecting candidate infectious agents associated with human tissue. <i>PLoS ONE</i> , 2011 , 6, e19838	3.7	51
52	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. <i>Journal of Medical Genetics</i> , 2010 , 47, 155-61	5.8	45
51	Clinical and genetic aspects of neurofibromatosis 1. <i>Genetics in Medicine</i> , 2010 , 12, 1-11	8.1	302
50	Impact of BRCA mutations on female fertility and offspring sex ratio. <i>American Journal of Human Biology</i> , 2010 , 22, 201-5	2.7	36
49	Value for money? Array genomic hybridization for diagnostic testing for genetic causes of intellectual disability. <i>American Journal of Human Genetics</i> , 2010 , 86, 765-72	11	46
48	Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. <i>American Journal of Human Genetics</i> , 2010 , 86, 749-64	11	1831
47	Prevalence of dental caries in children with neurofibromatosis 1. <i>Clinical Oral Investigations</i> , 2010 , 14, 479-80; author reply 480	4.2	6
46	Maternal use of bupropion and risk for congenital heart defects. <i>American Journal of Obstetrics and Gynecology</i> , 2010 , 203, 52.e1-6	6.4	63
45	The principles of teratology: are they still true?. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2010 , 88, 766-8		28

44	A different approach to validating screening assays for developmental toxicity. <i>Birth Defects Research Part B: Developmental and Reproductive Toxicology</i> , 2010 , 89, 526-30		39
43	Perceptions of genetic discrimination among people at risk for Huntington's disease: a cross sectional survey. <i>BMJ, The</i> , 2009 , 338, b2175	5.9	77
42	A novel de novo 1.1 Mb duplication of 17q21.33 associated with cognitive impairment and other anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 1257-62	2.5	10
41	Skeletal abnormalities in neurofibromatosis type 1: approaches to therapeutic options. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 2327-38	2.5	97
40	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. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 2393-402	2.5	52
39	Big risks in small groups: The difference between epidemiology and counselling. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2009 , 85, 720-4		6
38	Massively parallel sequencing: the next big thing in genetic medicine. <i>American Journal of Human Genetics</i> , 2009 , 85, 142-54	11	257
37	A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7. <i>European Journal of Medical Genetics</i> , 2009 , 52, 436-9	2.6	20
36	Safety of selective serotonin reuptake inhibitors in pregnancy. <i>CNS Drugs</i> , 2009 , 23, 493-509	6.7	69
35	Assessment of benign tumor burden by whole-body MRI in patients with neurofibromatosis 1. <i>Neuro-Oncology</i> , 2008 , 10, 593-8	1	151
34	A translocation t(6;7)(p11-p12;q22) associated with autism and mental retardation: localization and identification of candidate genes at the breakpoints. <i>Psychiatric Genetics</i> , 2008 , 18, 101-9	2.9	11
33	Use of Affymetrix mapping arrays in the diagnosis of gene copy number variation. <i>Current Protocols in Human Genetics</i> , 2008 , Chapter 8, Unit 8.13	3.2	7
32	Early primary tooth eruption in neurofibromatosis 1 individuals. <i>European Journal of Oral Sciences</i> , 2007 , 115, 425-6	2.3	9
31	Novel deletions of 14q11.2 associated with developmental delay, cognitive impairment and similar minor anomalies in three children. <i>Journal of Medical Genetics</i> , 2007 , 44, 556-61	5.8	58
30	Osteopoikilosis, short stature and mental retardation as key features of a new microdeletion syndrome on 12q14. <i>Journal of Medical Genetics</i> , 2007 , 44, 264-8	5.8	47
29	Use of selective serotonin-reuptake inhibitors in pregnancy and the risk of birth defects. <i>New England Journal of Medicine</i> , 2007 , 356, 2684-92	59.2	384
28	Selective serotonin-reuptake inhibitors and persistent pulmonary hypertension of the newborn. <i>New England Journal of Medicine</i> , 2006 , 354, 2188-90; author reply 2188-90	59.2	49
27	Addendum: sartan treatment during pregnancy. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2005 , 73, 904-5		6

26	Subcutaneous neurofibromas are associated with mortality in neurofibromatosis 1: a cohort study of 703 patients. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 132A, 49-53	2.5	57
25	Decreased bone mineral density in patients with neurofibromatosis 1. <i>Osteoporosis International</i> , 2005 , 16, 1161-6	5.3	110
24	Prevalence of neurofibromatosis 1 in German children at elementary school enrollment. <i>Archives of Dermatology</i> , 2005 , 141, 71-4		243
23	Patterns of associations of clinical features in neurofibromatosis 1 (NF1). <i>Human Genetics</i> , 2003 , 112, 289-97	6.3	43
22	Human chromosome 7: DNA sequence and biology. <i>Science</i> , 2003 , 300, 767-72	33.3	159
21	Growth charts for young children with neurofibromatosis 1 (NF1) 2000 , 92, 224-227		22
20	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. <i>American Journal of Medical Genetics Part A</i> , 2000 , 94, 35-41		33
19	Clinical objectives in medical genetics for undergraduate medical students. Association of Professors of Human Genetics, Clinical Objectives Task Force. <i>Genetics in Medicine</i> , 1998 , 1, 54-5	8.1	18
18	Genetics in Medicine and informatics for the genetic clinician. <i>Genetics in Medicine</i> , 1998 , 1, 52-52	8.1	2
17	Evidence for multi-site closure of the neural tube in humans. <i>American Journal of Medical Genetics Part A</i> , 1993 , 47, 723-43		317
16	Life-threatening status asthmaticus at 12.5 weeks' gestation. Report of a normal pregnancy outcome. <i>Chest</i> , 1991 , 100, 285-6	5.3	11
15	Congenital cataracts in mother, sister, and son of a patient with Hallermann-Streiff syndrome: coincidence or clue?. <i>American Journal of Medical Genetics Part A</i> , 1991 , 41, 500-2		4
14	Maternal serum alpha-fetoprotein in pregnancy. <i>American Journal of Obstetrics and Gynecology</i> , 1990 , 163, 692-3	6.4	
13	Congenital diaphragmatic hernia, coarse facies, and acral hypoplasia: Fryns syndrome. <i>American Journal of Medical Genetics Part A</i> , 1989 , 32, 93-9		51
12	Hepatocellular carcinoma in a child with familial Russell-Silver syndrome. <i>American Journal of Medical Genetics Part A</i> , 1988 , 31, 909-14		16
11	Kallmann syndrome associated with choanal atresia. <i>Clinical Genetics</i> , 1987 , 31, 224-7	4	21
10	Fetal karyotype following ascertainment of fetal anomalies by ultrasound. <i>Prenatal Diagnosis</i> , 1987 , 7, 551-5	3.2	45
9	Prevalence of Coxsackie B virus antibodies in patients with juvenile dermatomyositis. <i>Arthritis and Rheumatism</i> , 1986 , 29, 1365-70		154

8	Anatomic correlates of ultrasonographic prenatal diagnosis. <i>Prenatal Diagnosis</i> , 1986 , 6, 51-61	3.2	31
7	Immunogenetic studies of juvenile dermatomyositis. III. Study of antibody to organ-specific and nuclear antigens. <i>Arthritis and Rheumatism</i> , 1985 , 28, 151-7		41
6	Genetic heterogeneity in spondyloepiphyseal dysplasia congenita. <i>American Journal of Medical Genetics Part A</i> , 1984 , 18, 311-20		13
5	ACTIVE JUVENILE DERMATOMYOSITIS (JDMS) IS ASSOCIATED WITH COMPLEMENT AND COAGULATION ACTIVATION, AND INCREASED TITERS TO ANTINUCLEAR (ANA) AND COXSACKIE B VIRAL (COX-B) ANTIGENS. <i>Pediatric Research</i> , 1984 , 18, 262A-262A	3.2	3
4	Acute nonlymphocytic leukemia: expression in cells restricted to granulocytic and monocytic differentiation. <i>New England Journal of Medicine</i> , 1979 , 301, 1-5	59.2	71
3	Specificity of acquired clonal chromosome abnormalities in New Zealand Black mice. <i>International Journal of Cancer</i> , 1978 , 21, 505-10	7.5	11
2	White Matter is Increased in the Brains of Adults with Neurofibromatosis 1		1
1	Developmental Toxicology and Teratology9-17		