Jan M Friedman

List of Publications by Citations

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	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, 749-64	11	1831
150	Use of selective serotonin-reuptake inhibitors in pregnancy and the risk of birth defects. *New England Journal of Medicine*, **2007**, 356, 2684-92	59.2	384
149	Evidence for multi-site closure of the neural tube in humans. *American Journal of Medical Genetics Part A*, **1993**, 47, 723-43		317
148	Clinical and genetic aspects of neurofibromatosis 1. *Genetics in Medicine*, **2010**, 12, 1-11	8.1	302
147	Massively parallel sequencing: the next big thing in genetic medicine. *American Journal of Human Genetics*, **2009**, 85, 142-54	11	257
146	Prevalence of neurofibromatosis 1 in German children at elementary school enrollment. *Archives of Dermatology*, **2005**, 141, 71-4		243
145	Maternal treatment with opioid analgesics and risk for birth defects. *American Journal of Obstetrics and Gynecology*, **2011**, 204, 314.e1-11	6.4	235
144	Exome Sequencing and the Management of Neurometabolic Disorders. *New England Journal of Medicine*, **2016**, 374, 2246-55	59.2	197
143	FORGE Canada Consortium: outcomes of a 2-year national rare-disease gene-discovery project. *American Journal of Human Genetics*, **2014**, 94, 809-17	11	174
142	Human chromosome 7: DNA sequence and biology. *Science*, **2003**, 300, 767-72	33.3	159
141	Prevalence of Coxsackie B virus antibodies in patients with juvenile dermatomyositis. *Arthritis and Rheumatism*, **1986**, 29, 1365-70		154
140	Assessment of benign tumor burden by whole-body MRI in patients with neurofibromatosis 1. *Neuro-Oncology*, **2008**, 10, 593-8	1	151
139	Exome sequencing and functional validation in zebrafish identify GTDC2 mutations as a cause of Walker-Warburg syndrome. *American Journal of Human Genetics*, **2012**, 91, 541-7	11	144
138	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, 431-7	5.8	137
137	"I want to know what's in Pandora's Box": comparing stakeholder perspectives on incidental findings in clinical whole genomic sequencing. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2519-25	2.5	128
136	Patterns of antidepressant medication use among pregnant women in a United States population. *Journal of Clinical Pharmacology*, **2011**, 51, 264-70	2.9	122
135	A novel recurrent mutation in ATP1A3 causes CAPOS syndrome. *Orphanet Journal of Rare Diseases*, **2014**, 9, 15	4.2	121

134	Decreased bone mineral density in patients with neurofibromatosis 1. *Osteoporosis International*, **2005**, 16, 1161-6	5.3	110
133	Medications in the first trimester of pregnancy: most common exposures and critical gaps in understanding fetal risk. *Pharmacoepidemiology and Drug Safety*, **2013**, 22, 1013-8	2.6	103
132	Skeletal abnormalities in neurofibromatosis type 1: approaches to therapeutic options. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2327-38	2.5	97
131	Specific SSRIs and birth defects: Bayesian analysis to interpret new data in the context of previous reports. *BMJ, The*, **2015**, 351, h3190	5.9	96
130	Combined immunodeficiency associated with homozygous MALT1 mutations. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 1458-62, 1462.e1-7	11.5	91
129	Management of multiple sclerosis during pregnancy and the reproductive years: a systematic review. *Obstetrics and Gynecology*, **2014**, 124, 1157-1168	4.9	87
128	Low risk of solid tumors in persons with Down syndrome. *Genetics in Medicine*, **2016**, 18, 1151-1157	8.1	83
127	Perceptions of genetic discrimination among people at risk for Huntington's disease: a cross sectional survey. *BMJ, The*, **2009**, 338, b2175	5.9	77
126	Growth dynamics of plexiform neurofibromas: a retrospective cohort study of 201 patients with neurofibromatosis 1. *Orphanet Journal of Rare Diseases*, **2012**, 7, 75	4.2	75
125	Acute nonlymphocytic leukemia: expression in cells restricted to granulocytic and monocytic differentiation. *New England Journal of Medicine*, **1979**, 301, 1-5	59.2	71
124	Safety of selective serotonin reuptake inhibitors in pregnancy. *CNS Drugs*, **2009**, 23, 493-509	6.7	69
123	Empirical development of improved diagnostic criteria for neurofibromatosis 2. *Genetics in Medicine*, **2011**, 13, 576-81	8.1	67
122	Exome sequencing for gene discovery in lethal fetal disorders--harnessing the value of extreme phenotypes. *Prenatal Diagnosis*, **2015**, 35, 1005-9	3.2	63
121	Maternal use of bupropion and risk for congenital heart defects. *American Journal of Obstetrics and Gynecology*, **2010**, 203, 52.e1-6	6.4	63
120	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
119	Novel deletions of 14q11.2 associated with developmental delay, cognitive impairment and similar minor anomalies in three children. *Journal of Medical Genetics*, **2007**, 44, 556-61	5.8	58
118	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	2.5	57
117	The cost and diagnostic yield of exome sequencing for children with suspected genetic disorders: a benchmarking study. *Genetics in Medicine*, **2018**, 20, 1013-1021	8.1	55

116	Clinical and molecular predictors of mortality in neurofibromatosis 2: a UK national analysis of 1192 patients. *Journal of Medical Genetics*, **2015**, 52, 699-705	5.8	52
115	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, 2393-402	2.5	52
114	Congenital diaphragmatic hernia, coarse facies, and acral hypoplasia: Fryns syndrome. *American Journal of Medical Genetics Part A*, **1989**, 32, 93-9		51
113	The sensitivity of massively parallel sequencing for detecting candidate infectious agents associated with human tissue. *PLoS ONE*, **2011**, 6, e19838	3.7	51
112	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. *American Journal of Human Genetics*, **2017**, 101, 1021-1033	11	50
111	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, 307-13	4.8	49
110	Selective serotonin-reuptake inhibitors and persistent pulmonary hypertension of the newborn. *New England Journal of Medicine*, **2006**, 354, 2188-90; author reply 2188-90	59.2	49
109	Osteopoikilosis, short stature and mental retardation as key features of a new microdeletion syndrome on 12q14. *Journal of Medical Genetics*, **2007**, 44, 264-8	5.8	47
108	Value for money? Array genomic hybridization for diagnostic testing for genetic causes of intellectual disability. *American Journal of Human Genetics*, **2010**, 86, 765-72	11	46
107	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. *Journal of Medical Genetics*, **2010**, 47, 155-61	5.8	45
106	Fetal karyotype following ascertainment of fetal anomalies by ultrasound. *Prenatal Diagnosis*, **1987**, 7, 551-5	3.2	45
105	Genomic newborn screening: public health policy considerations and recommendations. *BMC Medical Genomics*, **2017**, 10, 9	3.7	44
104	Patterns of associations of clinical features in neurofibromatosis 1 (NF1). *Human Genetics*, **2003**, 112, 289-97	6.3	43
103	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. *Genetics in Medicine*, **2021**, 23, 1506-1513	8.1	43
102	Safety of Selective Serotonin Reuptake Inhibitors in Pregnancy: A Review of Current Evidence. *CNS Drugs*, **2016**, 30, 499-515	6.7	43
101	Approaches to treating NF1 tibial pseudarthrosis: consensus from the Children's Tumor Foundation NF1 Bone Abnormalities Consortium. *Journal of Pediatric Orthopaedics*, **2013**, 33, 269-75	2.4	42
100	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. *Brain*, **2019**, 142, 542-559	11.2	41
99	Paternalism and the ACMG recommendations on genomic incidental findings: patients seen but not heard. *Genetics in Medicine*, **2013**, 15, 751-2	8.1	41

98	Immunogenetic studies of juvenile dermatomyositis. III. Study of antibody to organ-specific and nuclear antigens. *Arthritis and Rheumatism*, **1985**, 28, 151-7		41
97	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, 1245-53	7	40
96	A different approach to validating screening assays for developmental toxicity. *Birth Defects Research Part B: Developmental and Reproductive Toxicology*, **2010**, 89, 526-30		39
95	Impact of BRCA mutations on female fertility and offspring sex ratio. *American Journal of Human Biology*, **2010**, 22, 201-5	2.7	36
94	Exposure-based validation list for developmental toxicity screening assays. *Birth Defects Research Part B: Developmental and Reproductive Toxicology*, **2014**, 101, 423-8		35
93	Safe lists for medications in pregnancy: inadequate evidence base and inconsistent guidance from Web-based information, 2011. *Pharmacoepidemiology and Drug Safety*, **2013**, 22, 324-8	2.6	35
92	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		33
91	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, 217-26	3.5	32
90	Anatomic correlates of ultrasonographic prenatal diagnosis. *Prenatal Diagnosis*, **1986**, 6, 51-61	3.2	31
89	Single exon-resolution targeted chromosomal microarray analysis of known and candidate intellectual disability genes. *European Journal of Human Genetics*, **2014**, 22, 792-800	5.3	30
88	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unit-successes and challenges. *European Journal of Pediatrics*, **2019**, 178, 1207-1218	4.1	29
87	Genome-wide sequencing in acutely ill infants: genomic medicine's critical application?. *Genetics in Medicine*, **2019**, 21, 498-504	8.1	28
86	The principles of teratology: are they still true?. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2010**, 88, 766-8		28
85	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	3.7	25
84	Different patterns of mast cells distinguish diffuse from encapsulated neurofibromas in patients with neurofibromatosis 1. *Journal of Histochemistry and Cytochemistry*, **2011**, 59, 584-90	3.4	24
83	Key Implications of Data Sharing in Pediatric Genomics. *JAMA Pediatrics*, **2018**, 172, 476-481	8.3	22
82	Genetic mosaics and the germ line lineage. *Genes*, **2015**, 6, 216-37	4.2	22
81	Growth charts for young children with neurofibromatosis 1 (NF1) **2000**, 92, 224-227		22

80	Non-optic glioma in adults and children with neurofibromatosis 1. *Orphanet Journal of Rare Diseases*, **2017**, 12, 34	4.2	21
79	Kallmann syndrome associated with choanal atresia. *Clinical Genetics*, **1987**, 31, 224-7	4	21
78	The importance of genetic counselling in genome-wide sequencing. *Nature Reviews Genetics*, **2018**, 19, 735-736	30.1	21
77	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	4.3	20
76	Effect of vitamin D3 treatment on bone density in neurofibromatosis 1 patients: a retrospective clinical study. *Joint Bone Spine*, **2013**, 80, 315-9	2.9	20
75	A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7. *European Journal of Medical Genetics*, **2009**, 52, 436-9	2.6	20
74	Clinical objectives in medical genetics for undergraduate medical students. Association of Professors of Human Genetics, Clinical Objectives Task Force. *Genetics in Medicine*, **1998**, 1, 54-5	8.1	18
73	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. *Brain*, **2020**, 143, 55-68	11.2	18
72	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	4.2	17
71	The composition and capacity of the clinical genetics workforce in high-income countries: a scoping review. *Genetics in Medicine*, **2020**, 22, 1437-1449	8.1	16
70	Hepatocellular carcinoma in a child with familial Russell-Silver syndrome. *American Journal of Medical Genetics Part A*, **1988**, 31, 909-14		16
69	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
68	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
67	Mutations in ILK, encoding integrin-linked kinase, are associated with arrhythmogenic cardiomyopathy. *Translational Research*, **2019**, 208, 15-29	11	14
66	Recurrent subacute post-viral onset of ataxia associated with a PRF1 mutation. *European Journal of Human Genetics*, **2013**, 21, 1232-9	5.3	14
65	Factors associated with experiences of genetic discrimination among individuals at risk for Huntington disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156B, 19-27	3.5	14
64	Parents' Perspectives on Supporting Their Decision Making in Genome-Wide Sequencing. *Journal of Nursing Scholarship*, **2016**, 48, 265-75	3.6	14
63	Growth in neurofibromatosis 1 microdeletion patients. *Clinical Genetics*, **2016**, 89, 351-4	4	14

62	The cost trajectory of the diagnostic care pathway for children with suspected genetic disorders. *Genetics in Medicine*, **2020**, 22, 292-300	8.1	14
61	Assessing an Interactive Online Tool to Support Parents' Genomic Testing Decisions. *Journal of Genetic Counseling*, **2018**, 28, 10	2.5	13
60	Prevalence of selected genomic deletions and duplications in a French-Canadian population-based sample of newborns. *Molecular Genetics & Genomic Medicine*, **2013**, 1, 87-97	2.3	13
59	Genetic heterogeneity in spondyloepiphyseal dysplasia congenita. *American Journal of Medical Genetics Part A*, **1984**, 18, 311-20		13
58	Aspartylglycosamine is a biomarker for NGLY1-CDDG, a congenital disorder of deglycosylation. *Molecular Genetics and Metabolism*, **2019**, 127, 368-372	3.7	12
57	Comparing the ability of OPTION(12) and OPTION(5) to assess shared decision-making in genetic counselling. *Patient Education and Counseling*, **2016**, 99, 1717-23	3.1	12
56	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
55	Update from the 2013 International Neurofibromatosis Conference. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2969-78	2.5	11
54	A translocation t(6;7)(p11-p12;q22) associated with autism and mental retardation: localization and identification of candidate genes at the breakpoints. *Psychiatric Genetics*, **2008**, 18, 101-9	2.9	11
53	Life-threatening status asthmaticus at 12.5 weeks' gestation. Report of a normal pregnancy outcome. *Chest*, **1991**, 100, 285-6	5.3	11
52	Specificity of acquired clonal chromosome abnormalities in New Zealand Black mice. *International Journal of Cancer*, **1978**, 21, 505-10	7.5	11
51	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, 1257-62	2.5	10
50	Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability. *BMC Genomics*, **2017**, 18, 403	4.5	9
49	Early primary tooth eruption in neurofibromatosis 1 individuals. *European Journal of Oral Sciences*, **2007**, 115, 425-6	2.3	9
48	De novo pathogenic DNM1L variant in a patient diagnosed with atypical hereditary sensory and autonomic neuropathy. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00961	2.3	8
47	Individual DNA samples and health information sold by 23andMe. *Genetics in Medicine*, **2016**, 18, 305-6	8.1	7
46	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
45	Use of Affymetrix mapping arrays in the diagnosis of gene copy number variation. *Current Protocols in Human Genetics*, **2008**, Chapter 8, Unit 8.13	3.2	7

44	Quantitative associations of scalp and body subcutaneous neurofibromas with internal plexiform tumors in neurofibromatosis 1. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1518-24	2.5	6
43	Big risks in small groups: The difference between epidemiology and counselling. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2009**, 85, 720-4		6
42	Prevalence of dental caries in children with neurofibromatosis 1. *Clinical Oral Investigations*, **2010**, 14, 479-80; author reply 480	4.2	6
41	Addendum: sartan treatment during pregnancy. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2005**, 73, 904-5		6
40	Atypical antipsychotic use during pregnancy and birth defect risk: National Birth Defects Prevention Study, 1997-2011. *Schizophrenia Research*, **2020**, 215, 81-88	3.6	6
39	Exome Sequencing and Clinical Diagnosis. *JAMA - Journal of the American Medical Association*, **2020**,	27.4	6
38	A case of splenomegaly in CBL syndrome. *European Journal of Medical Genetics*, **2017**, 60, 374-379	2.6	5
37	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	2.5	5
36	S100B and neurofibromin immunostaining and X-inactivation patterns of laser-microdissected cells indicate a multicellular origin of some NF1-associated neurofibromas. *Journal of Neuroscience Research*, **2011**, 89, 1451-60	4.4	5
35	A novel mouse model for pyridoxine-dependent epilepsy due to antiquitin deficiency. *Human Molecular Genetics*, **2020**, 29, 3266-3284	5.6	5
34	Life-history chronicle for a patient with the recently described chromosome 4q21 microdeletion syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2606-9	2.5	4
33	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, 500-2		4
32	Genome-wide sequencing as a first-tier screening test for short tandem repeat expansions. *Genome Medicine*, **2021**, 13, 126	14.4	4
31	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	3.2	3
30	Genetics in Medicine and informatics for the genetic clinician. *Genetics in Medicine*, **1998**, 1, 52-52	8.1	2
29	Toward the diagnosis of rare childhood genetic diseases: what do parents value most?. *European Journal of Human Genetics*, **2021**, 29, 1491-1501	5.3	2
28	Neurofibromin haploinsufficiency results in altered spermatogenesis in a mouse model of neurofibromatosis type 1. *PLoS ONE*, **2018**, 13, e0208835	3.7	2
27	Straglr: discovering and genotyping tandem repeat expansions using whole genome long-read sequences. *Genome Biology*, **2021**, 22, 224	18.3	2

26	Utilization of telehealth in paediatric genome-wide sequencing: Health services implementation issues in the CAUSES Study. *Journal of Telemedicine and Telecare*, **2021**, 1357633X20982737	6.8	2
25	Strabismus in Children With Intellectual Disability: Part of a Broader Motor Control Phenotype?. *Pediatric Neurology*, **2019**, 100, 87-91	2.9	1
24	Clinical Teratology **2013**, 1-39		1
23	Antineoplastic drugs **2015**, 373-399		1
22	Emerging issues in teratology: an introduction. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2011**, 157C, 147-9	3.1	1
21	Alterations in brain morphology by MRI in adults with neurofibromatosis 1. *Orphanet Journal of Rare Diseases*, **2021**, 16, 462	4.2	1
20	Molecular Basis of Cardiovascular Abnormalities in NF1 **2012**, 353-366		1
19	Renpenning syndrome in a female. *American Journal of Medical Genetics, Part A*, **2020**, 182, 498-503	2.5	1
18	MRI based volumetric measurements of vestibular schwannomas in patients with neurofibromatosis type 2: comparison of three different software tools. *Scientific Reports*, **2020**, 10, 11541	4.9	1
17	White Matter is Increased in the Brains of Adults with Neurofibromatosis 1		1
16	After genomic testing results: Parents' long-term views. *Journal of Genetic Counseling*, **2021**,	2.5	1
15	Utilization and uptake of clinical genetics services in high-income countries: A scoping review. *Health Policy*, **2021**, 125, 877-887	3.2	1
14	Exome Sequencing as Part of a Multidisciplinary Approach to Diagnosis-Reply. *JAMA - Journal of the American Medical Association*, **2020**, 324, 2445-2446	27.4	1
13	Prescription opioid use during pregnancy and risk for preterm birth or term low birthweight. *Journal of Opioid Management*, **2021**, 17, 215-225	0.8	0
12	Genome-wide Sequencing and the Clinical Diagnosis of Genetic Disease: The CAUSES Study. *Human Genetics and Genomics Advances*, **2022**, 100108	0.8	0
11	A personalized genomic results e-booklet, co-designed and pilot-tested by families. *PEC Innovation*, **2022**, 100039		0
10	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	
9	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	

8	Assessment of Case Reports and Clinical Series **2018**, 389-396	
7	Effet de la vitamine D3´sur la densit osseuse chez les patients porteurs d une neurofibromatose 1´: une ude clinique r rospective. *Revue Du Rhumatisme (Edition Francaise)*, **2013**, 80, 262-266	0.1
6	Massively Parallel Sequencing **2011**, 114-134	
5	Maternal serum alpha-fetoprotein in pregnancy. *American Journal of Obstetrics and Gynecology*, **1990**, 163, 692-3	6.4
4	Secondary biogenic amine deficiencies: genetic etiology, therapeutic interventions, and clinical effects. *Neurogenetics*, **2021**, 22, 251-262	3
3	Clinical Teratology **2019**, 15-60	
2	White matter is increased in the brains of adults with neurofibromatosis 1.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 115	4.2
1	Developmental Toxicology and Teratology9-17	