

# Moshe Frydman

## List of Publications by Year in descending order

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44  
papers

4,388  
citations

393982

19  
h-index

329751

37  
g-index

47  
all docs

47  
docs citations

47  
times ranked

4634  
citing authors

#	ARTICLE	IF	CITATIONS
1	Refining the Phenotypic Spectrum of KMT5B-Associated Developmental Delay. <i>Frontiers in Pediatrics</i> , 2022, 10, 844845.	0.9	8
2	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene <i>ATOH1</i> . <i>Clinical Genetics</i> , 2020, 98, 353-364.	1.0	15
3	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. <i>Genetics in Medicine</i> , 2019, 21, 2442-2452.	1.1	56
4	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. <i>Journal of Clinical Immunology</i> , 2016, 36, 220-234.	2.0	71
5	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotype-Phenotype Correlation. <i>Human Mutation</i> , 2015, 36, 1052-1063.	1.1	143
6	Autosomal-dominant cutis laxa resulting from an intronic mutation in <i>ELN</i> . <i>Experimental Dermatology</i> , 2015, 24, 885-887.	1.4	4
7	Late onset fulminant Wilson's disease: A case report and review of the literature. <i>World Journal of Gastroenterology</i> , 2014, 20, 17656.	1.4	22
8	Novel myosin mutations for hereditary hearing loss revealed by targeted genomic capture and massively parallel sequencing. <i>European Journal of Human Genetics</i> , 2014, 22, 768-775.	1.4	44
9	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in middle eastern families. <i>Genome Biology</i> , 2011, 12, R89.	13.9	183
10	Mutations in FYCO1 Cause Autosomal-Recessive Congenital Cataracts. <i>American Journal of Human Genetics</i> , 2011, 88, 827-838.	2.6	132
11	Oligodontia, short stature and small head circumference with normal intelligence. <i>Clinical Genetics</i> , 2008, 46, 316-318.	1.0	2
12	The Marfan syndrome. <i>Israel Medical Association Journal</i> , 2008, 10, 175-8.	0.1	12
13	Congenital diaphragmatic hernia in a family segregating a reciprocal translocation t(5;15)(p15.3;q24). , 2000, 90, 120-122.		23
14	Molecular cytogenetic studies in three patients with partial trisomy 2p, including CGH from paraffin-embedded tissue. , 2000, 91, 74-82.		29
15	Noonan syndrome: A cryptic condition in early gestation. , 2000, 92, 159-165.		58
16	Screening for fragile X syndrome in women of reproductive age. <i>Prenatal Diagnosis</i> , 2000, 20, 611-614.	1.1	117
17	Prenatal diagnosis of a novel COL1A1 mutation in osteogenesis imperfecta type I carried through full term pregnancy. <i>Prenatal Diagnosis</i> , 2000, 20, 876-880.	1.1	18
18	Prenatal diagnosis of nail-patella syndrome. , 1999, 19, 287-288.		11

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19	Syndrome of alopecia totalis and 17b-hydroxysteroid dehydrogenase deficiency. American Journal of Medical Genetics Part A, 1998, 76, 28-31.	2.4	4
20	Linkage disequilibrium of common Gaucher disease mutations with a polymorphic site in the pyruvate kinase (PKLR) gene. , 1998, 78, 233-236.		8
21	Identification of ATM mutations using extended RT-PCR and restriction endonuclease fingerprinting, and elucidation of the repertoire of A-T mutations in Israel. Human Mutation, 1998, 11, 69-75.	1.1	36
22	No founder effect detected in Jewish Ashkenazi patients with fragile-X syndrome. Human Genetics, 1997, 101, 186-189.	1.8	8
23	MODED: Microcephaly-oculo-digito-esophageal-duodenal syndrome. , 1997, 71, 251-257.		21
24	Velocardiofacial manifestations and microdeletions in schizophrenic inpatients. , 1997, 72, 455-461.		68
25	Velocardiofacial manifestations and microdeletions in schizophrenic inpatients. American Journal of Medical Genetics Part A, 1997, 72, 455-461.	2.4	3
26	t(15;21)(q15;q22.1)pat resulting in partial trisomy and partial monosomy of chromosomes 15 and 21 in two offspring. , 1996, 66, 45-51.		6
27	PRENATAL DIAGNOSIS OF RAMBAM'S "HASHARON SYNDROME. Prenatal Diagnosis, 1996, 16, 266-269.	1.1	9
28	Predominance of null mutations in ataxia-telangiectasia. Human Molecular Genetics, 1996, 5, 433-439.	1.4	247
29	Duchenne muscular dystrophy and idiopathic hyperCKemia segregating in a family. American Journal of Medical Genetics Part A, 1995, 58, 209-212.	2.4	10
30	A single ataxia telangiectasia gene with a product similar to PI-3 kinase. Science, 1995, 268, 1749-1753.	6.0	2,634
31	Familial segregation of cervical ribs, Sprengel anomaly, preaxial polydactyly, anal atresia, and urethral obstruction: A new syndrome?. American Journal of Medical Genetics Part A, 1993, 45, 717-720.	2.4	18
32	Alpers progressive infantile neuronal poliodystrophy: An acute neonatal form with findings of the fetal akinesia syndrome. American Journal of Medical Genetics Part A, 1993, 47, 31-36.	2.4	12
33	Vici syndrome (OFD VI) or opitz trigonocephaly syndrome: Overlapping manifestations in two cousins. American Journal of Medical Genetics Part A, 1993, 47, 451-455.	2.4	18
34	Leukonychia totalis in two sibs. American Journal of Medical Genetics Part A, 1993, 47, 540-541.	2.4	20
35	Autosomal recessive Peters anomaly, typical facial appearance, failure to thrive, hydrocephalus, and other anomalies: Further delineation of the Krause-Kivlin syndrome. American Journal of Medical Genetics Part A, 1991, 40, 34-40.	2.4	30
36	Schizophrenia and Marfan Syndrome. British Journal of Psychiatry, 1990, 157, 433-436.	1.7	29

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37	A single origin of phenylketonuria in Yemenite Jews. <i>Nature</i> , 1990, 344, 168-170.	13.7	68
38	Possible heterogeneity in spondyloenchondrodysplasia: Quadriparesis, basal ganglia calcifications, and chondrocyte inclusions. <i>American Journal of Medical Genetics Part A</i> , 1990, 36, 279-284.	2.4	30
39	Ambiguous genitalia in the Proteus syndrome. <i>American Journal of Medical Genetics Part A</i> , 1990, 36, 511-512.	2.4	7
40	Genetic aspects of Wilson's disease. <i>Journal of Gastroenterology and Hepatology (Australia)</i> , 1990, 5, 483-490.	1.4	81
41	Interstitial deletion 2q14q21. <i>American Journal of Medical Genetics Part A</i> , 1989, 34, 476-479.	2.4	28
42	Familial simple hypohidrosis with abnormal palmar dermal ridges. <i>American Journal of Medical Genetics Part A</i> , 1988, 31, 591-596.	2.4	4
43	Interstitial 7q deletion [46,XY,del (7) (pter â†’ cen::q112 â†’ qter)] in a retarded quadriplegic boy with normal beta glucuronidase. <i>American Journal of Medical Genetics Part A</i> , 1986, 25, 245-249.	2.4	21
44	Oculoâ€palatoâ€cerebral dwarfism: a new syndrome. <i>Clinical Genetics</i> , 1985, 27, 414-419.	1.0	16