Moshe Frydman

List of Publications by Year
in descending order

44 papers	4,388 citations	331670 21 h-index	276875 41 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. Frontiers in Pediatrics, 2022, 10, 844845.	1.9	8
2	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene <scp>ATOH1</scp>. Clinical Genetics, 2020, 98, 353-364.	2.0	15
3	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. Genetics in Medicine, 2019, 21, 2442-2452.	2.4	56
4	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
5	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€"Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	2.5	143
6	Autosomalâ€dominant cutis laxa resulting from an intronic mutation in <scp>ELN</scp>. Experimental Dermatology, 2015, 24, 885-887.	2.9	4
7	Late onset fulminant Wilsonâ€™s disease: A case report and review of the literature. World Journal of Gastroenterology, 2014, 20, 17656.	3.3	22
8	Novel myosin mutations for hereditary hearing loss revealed by targeted genomic capture and massively parallel sequencing. European Journal of Human Genetics, 2014, 22, 768-775.	2.8	44
9	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in middle eastern families. Genome Biology, 2011, 12, R89.	9.6	183
10	Mutations in FYCO1 Cause Autosomal-Recessive Congenital Cataracts. American Journal of Human Genetics, 2011, 88, 827-838.	6.2	132
11	Oligodontia, short stature and small head circumference with normal intelligence. Clinical Genetics, 2008, 46, 316-318.	2.0	2
12	The Marfan syndrome. Israel Medical Association Journal, 2008, 10, 175-8.	0.1	12
13	Congenital diaphragmatic hernia in a family segregating a reciprocal translocation t(5;15)(p15.3;q24). American Journal of Medical Genetics Part A, 2000, 90, 120-122.	2.4	23
14	Molecular cytogenetic studies in three patients with partial trisomy 2p, including CGH from paraffin-embedded tissue. American Journal of Medical Genetics Part A, 2000, 91, 74-82.	2.4	29
15	Noonan syndrome: A cryptic condition in early gestation. American Journal of Medical Genetics Part A, 2000, 92, 159-165.	2.4	58
16	Screening for fragile X syndrome in women of reproductive age. Prenatal Diagnosis, 2000, 20, 611-614.	2.3	117
17	Prenatal diagnosis of a novel*COL1A1* mutation in osteogenesis imperfecta type I carried through full term pregnancy. Prenatal Diagnosis, 2000, 20, 876-880.	2.3	18
18	Prenatal diagnosis of nail-patella syndrome. Prenatal Diagnosis, 1999, 19, 287-288.	2.3	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 thepyruvate 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.	2.5	36
22	No founder effect detected in Jewish Ashkenazi patients with fragile-X syndrome. Human Genetics, 1997, 101, 186-189.	3.8	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 RAMBAMâ€"HASHARON SYNDROME. Prenatal Diagnosis, 1996, 16, 266-269.	2.3	9
28	Predominance of null mutations in ataxia-telangiectasia. Human Molecular Genetics, 1996, 5, 433-439.	2.9	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.	12.6	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	VÃ¡radi 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.	2.8	29

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37	A single origin of phenylketonuria in Yemenite Jews. Nature, 1990, 344, 168-170.	27.8	68
38	Possible heterogeneity in spondyloenchondrodysplasia: Quadriparesis, basal ganglia calcifications, and chondrocyte inclusions. American Journal of Medical Genetics Part A, 1990, 36, 279-284.	2.4	30
39	Ambiguous genitalia in the Proteus syndrome. American Journal of Medical Genetics Part A, 1990, 36, 511-512.	2.4	7
40	Genetic aspects of Wilson's disease. Journal of Gastroenterology and Hepatology (Australia), 1990, 5, 483-490.	2.8	81
41	Interstitial deletion 2q14q21. American Journal of Medical Genetics Part A, 1989, 34, 476-479.	2.4	28
42	Familial simple hypohidrosis with abnormal palmar dermal ridges. American Journal of Medical Genetics Part A, 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. American Journal of Medical Genetics Part A, 1986, 25, 245-249.	2.4	21
44	Oculoâ€palatoâ€cerebral dwarfism: a new syndrome. Clinical Genetics, 1985, 27, 414-419.	2.0	16