Moshe Frydman

List of Publications by Year
in descending order

44	4,388	331670 21	276875 41
papers	citations	h-index	g-index

47	47	47	4634
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A Single Ataxia Telangiectasia Gene with a Product Similar to PI-3 Kinase. Science, 1995, 268, 1749-1753.	12.6	2,634
2	Predominance of null mutations in ataxia-telangiectasia. Human Molecular Genetics, 1996, 5, 433-439.	2.9	247
3	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
4	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
5	Mutations in FYCO1 Cause Autosomal-Recessive Congenital Cataracts. American Journal of Human Genetics, 2011, 88, 827-838.	6.2	132
6	Screening for fragile X syndrome in women of reproductive age. Prenatal Diagnosis, 2000, 20, 611-614.	2.3	117
7	Genetic aspects of Wilson's disease. Journal of Gastroenterology and Hepatology (Australia), 1990, 5, 483-490.	2.8	81
8	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
9	A single origin of phenylketonuria in Yemenite Jews. Nature, 1990, 344, 168-170.	27.8	68
10	Velocardiofacial manifestations and microdeletions in schizophrenic inpatients. , 1997, 72, 455-461.		68
11	Noonan syndrome: A cryptic condition in early gestation. American Journal of Medical Genetics Part A, 2000, 92, 159-165.	2.4	58
12	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
13	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
14	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
15	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
16	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
17	Schizophrenia and Marfan Syndrome. British Journal of Psychiatry, 1990, 157, 433-436.	2.8	29
18	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

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19	Interstitial deletion 2q14q21. American Journal of Medical Genetics Part A, 1989, 34, 476-479.	2.4	28
20	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
21	Late onset fulminant Wilsonâ€™s disease: A case report and review of the literature. World Journal of Gastroenterology, 2014, 20, 17656.	3.3	22
22	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
23	MODED: Microcephaly-oculo-digito-esophageal-duodenal syndrome. , 1997, 71, 251-257.		21
24	Leukonychia totalis in two sibs. American Journal of Medical Genetics Part A, 1993, 47, 540-541.	2.4	20
25	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
26	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
27	Prenatal diagnosis of a novelCOL1A1 mutation in osteogenesis imperfecta type I carried through full term pregnancy. Prenatal Diagnosis, 2000, 20, 876-880.	2.3	18
28	Oculoâ€palatoâ€cerebral dwarfism: a new syndrome. Clinical Genetics, 1985, 27, 414-419.	2.0	16
29	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
30	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
31	The Marfan syndrome. Israel Medical Association Journal, 2008, 10, 175-8.	0.1	12
32	Prenatal diagnosis of nail-patella syndrome. Prenatal Diagnosis, 1999, 19, 287-288.	2.3	11
33	Duchenne muscular dystrophy and idiopathic hyperCKemia segregating in a family. American Journal of Medical Genetics Part A, 1995, 58, 209-212.	2.4	10
34	PRENATAL DIAGNOSIS OF RAMBAMâ€“HASHARON SYNDROME. Prenatal Diagnosis, 1996, 16, 266-269.	2.3	9
35	No founder effect detected in Jewish Ashkenazi patients with fragile-X syndrome. Human Genetics, 1997, 101, 186-189.	3.8	8
36	Linkage disequilibrium of common Gaucher disease mutations with a polymorphic site in thepyruvate kinase (PKLR) gene. , 1998, 78, 233-236.		8

#	Article	IF	Citations
37	Refining the Phenotypic Spectrum of KMT5B-Associated Developmental Delay. Frontiers in Pediatrics, 2022, 10, 844845.	1.9	8
38	Ambiguous genitalia in the Proteus syndrome. American Journal of Medical Genetics Part A, 1990, 36, 511-512.	2.4	7
39	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
40	Familial simple hypohidrosis with abnormal palmar dermal ridges. American Journal of Medical Genetics Part A, 1988, 31, 591-596.	2.4	4
41	Syndrome of alopecia totalis and 17b-hydroxysteroid dehydrogenase deficiency. American Journal of Medical Genetics Part A, 1998, 76, 28-31.	2.4	4
42	Autosomalâ€dominant cutis laxa resulting from an intronic mutation in <scp>ELN</scp>. Experimental Dermatology, 2015, 24, 885-887.	2.9	4
43	Velocardiofacial manifestations and microdeletions in schizophrenic inpatients. American Journal of Medical Genetics Part A, 1997, 72, 455-461.	2.4	3
44	Oligodontia, short stature and small head circumference with normal intelligence. Clinical Genetics, 2008, 46, 316-318.	2.0	2