Marwan S Shinawi

List of Publications by Year
in descending order

153 papers	8,391 citations	76196 40 h-index	54797 84 g-index
162 all docs	162 docs citations	162 times ranked	14122 citing authors

#	Article	IF	Citations
1	Mutations involved in Aicardi-GoutiÃ¨res syndrome implicate SAMHD1 as regulator of the innate immune response. Nature Genetics, 2009, 41, 829-832.	9.4	610
2	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471.	9.4	535
3	Prader-Willi phenotype caused by paternal deficiency for the HBII-85 C/D box small nucleolar RNA cluster. Nature Genetics, 2008, 40, 719-721.	9.4	533
4	A phase II/III clinical study of enzyme replacement therapy with idursulfase in mucopolysaccharidosis II (Hunter syndrome). Genetics in Medicine, 2006, 8, 465-473.	1.1	499
5	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	1.5	447
6	STXBP1 encephalopathy. Neurology, 2016, 86, 954-962.	1.5	264
7	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	13.9	239
8	Microdeletion 15q13.3: a locus with incomplete penetrance for autism, mental retardation, and psychiatric disorders. Journal of Medical Genetics, 2009, 46, 382-388.	1.5	213
9	Long-term, open-labeled extension study of idursulfase in the treatment of Hunter syndrome. Genetics in Medicine, 2011, 13, 95-101.	1.1	190
10	CpG Island Hypermethylation Mediated by DNMT3A Is a Consequence of AML Progression. Cell, 2017, 168, 801-816.e13.	13.5	177
11	The array CGH and its clinical applications. Drug Discovery Today, 2008, 13, 760-770.	3.2	171
12	A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. Nature Genetics, 2009, 41, 1269-1271.	9.4	171
13	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
14	Familial Mediterranean fever: prevalence, penetrance and genetic drift. European Journal of Human Genetics, 2001, 9, 634-637.	1.4	146
15	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981.	3.3	118
16	The differential contribution of MEFV mutant alleles to the clinical profile of familial Mediterranean fever. European Journal of Human Genetics, 2002, 10, 145-149.	1.4	116
17	Microdeletions including YWHAE in the Miller-Dieker syndrome region on chromosome 17p13.3 result in facial dysmorphisms, growth restriction, and cognitive impairment. Journal of Medical Genetics, 2009, 46, 825-833.	1.5	112
18	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850.	1.1	111

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19	Mitochondrial Neurogastrointestinal Encephalopathy Due to Mutations in RRM2B. Archives of Neurology, 2009, 66, 1028-32.	4.9	103
20	Familial Mediterranean Fever: Clinical and Genetic Characterization in a Mixed Pediatric Population of Jewish and Arab Patients. Pediatrics, 1999, 103, e70-e70.	1.0	101
21	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	2.6	98
22	Clinical Characterization of Patients With Autosomal Dominant Short Stature due to Aggrecan Mutations. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 460-469.	1.8	95
23	Diagnostic testing for uniparental disomy: a points to consider statement from the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1133-1141.	1.1	89
24	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
25	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048.	1.1	85
26	11p14.1 microdeletions associated with ADHD, autism, developmental delay, and obesity. American Journal of Medical Genetics, Part A, 2011, 155, 1272-1280.	0.7	84
27	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	3.7	81
28	The musculoskeletal manifestations of familial Mediterranean fever in children genetically diagnosed with the disease. Arthritis and Rheumatism, 2001, 44, 1416-1419.	6.7	78
29	Syndromic thrombocytopenia and predisposition to acute myelogenous leukemia caused by constitutional microdeletions on chromosome 21q. Blood, 2008, 112, 1042-1047.	0.6	74
30	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988.	1.4	74
31	TSHZ3 deletion causes an autism syndrome and defects in cortical projection neurons. Nature Genetics, 2016, 48, 1359-1369.	9.4	69
32	The Cognitive and Behavioral Phenotypes of Individuals with CHRNA7 Duplications. Journal of Autism and Developmental Disorders, 2017, 47, 549-562.	1.7	68
33	Neuroligin 2 nonsense variant associated with anxiety, autism, intellectual disability, hyperphagia, and obesity. American Journal of Medical Genetics, Part A, 2017, 173, 213-216.	0.7	68
34	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain, 2018, 141, 1998-2013.	3.7	67
35	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150.	1.1	64
36	The MTHFR 677Câ†'T polymorphism and behaviors in children with autism: exploratory genotypeâ€“phenotype correlations. Autism Research, 2009, 2, 98-108.	2.1	57

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37	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245.	2.6	56
38	Desmosterolosisâ€"phenotypic and molecular characterization of a third case and review of the literature. American Journal of Medical Genetics, Part A, 2011, 155, 1597-1604.	0.7	52
39	De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. American Journal of Human Genetics, 2015, 96, 682-690.	2.6	48
40	Mutations inCOQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. Journal of Medical Genetics, 2015, 52, 627-635.	1.5	48
41	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	2.6	48
42	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	1.1	48
43	Expanding the clinical spectrum of the 16p11.2 chromosomal rearrangements: three patients with syringomyelia. European Journal of Human Genetics, 2011, 19, 152-156.	1.4	47
44	Immunodeficiency and bone marrow failure with mosaic and germline TLR8 gain of function. Blood, 2021, 137, 2450-2462.	0.6	47
45	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. Human Mutation, 2012, 33, 165-179.	1.1	45
46	Multiâ€systemic involvement in NGLY1â€related disorder caused by two novel mutations. American Journal of Medical Genetics, Part A, 2015, 167, 816-820.	0.7	45
47	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	1.4	45
48	The spectrum of DNMT3A variants in Tattonâ€"Brownâ€"Rahman syndrome overlaps with that in hematologic malignancies. American Journal of Medical Genetics, Part A, 2017, 173, 3022-3028.	0.7	42
49	WACloss-of-function mutations cause a recognisable syndrome characterised by dysmorphic features, developmental delay and hypotonia and recapitulate 10p11.23 microdeletion syndrome. Journal of Medical Genetics, 2015, 52, 754-761.	1.5	41
50	A Novel Mutation in Isoform 3 of the Plasma Membrane Ca2+ Pump Impairs Cellular Ca2+ Homeostasis in a Patient with Cerebellar Ataxia and Laminin Subunit 1Î± Mutations. Journal of Biological Chemistry, 2015, 290, 16132-16141.	1.6	41
51	NR2F1 haploinsufficiency is associated with optic atrophy, dysmorphism and global developmental delay. American Journal of Medical Genetics, Part A, 2013, 161, 377-381.	0.7	40
52	Dosage Changes of a Segment at 17p13.1 Lead to Intellectual Disability and Microcephaly as a Result of Complex Genetic Interaction of Multiple Genes. American Journal of Human Genetics, 2014, 95, 565-578.	2.6	40
53	Scoliosis and vertebral anomalies: Additional abnormal phenotypes associated with chromosome 16p11.2 rearrangement. American Journal of Medical Genetics, Part A, 2014, 164, 1118-1126.	0.7	38
54	Lowâ€level mosaicism of trisomy 14: Phenotypic and molecular characterization. American Journal of Medical Genetics, Part A, 2008, 146A, 1395-1405.	0.7	34

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55	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178.	0.7	32
56	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	2.6	32
57	Two novel RAD21 mutations in patients with mild Cornelia de Lange syndrome-like presentation and report of the first familial case. Gene, 2014, 537, 279-284.	1.0	31
58	Haploinsufficiency of ZNF238 is associated with corpus callosum abnormalities in 1q44 deletions. American Journal of Medical Genetics, Part A, 2013, 161, 711-716.	0.7	28
59	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	1.1	27
60	Prevalence of Semicircular Canal Hypoplasia in Patients With CHARGE Syndrome. JAMA Otolaryngology - Head and Neck Surgery, 2017, 143, 168.	1.2	24
61	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	1.8	24
62	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483.	1.1	24
63	Atypical presentation of VLCAD deficiency associated with a novel ACADVL splicing mutation. Muscle and Nerve, 2009, 39, 374-382.	1.0	23
64	Mixed gonadal dysgenesis in a child with isodicentric y chromosome: Does the relative proportion of the 45,X line really matter?. American Journal of Medical Genetics, Part A, 2010, 152A, 1832-1837.	0.7	23
65	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). Bone, 2018, 107, 161-171.	1.4	23
66	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	1.1	23
67	2-Pyrrolidinone and Succinimide as Clinical Screening Biomarkers for GABA-Transaminase Deficiency: Anti-seizure Medications Impact Accurate Diagnosis. Frontiers in Neuroscience, 2019, 13, 394.	1.4	23
68	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	2.6	23
69	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	1.1	23
70	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Clâ˜'/H+-Exchanger, Causes Early-Onset Neurodegeneration. American Journal of Human Genetics, 2020, 107, 1062-1077.	2.6	23
71	Familial Mediterranean fever: high gene frequency and heterogeneous disease among an Israeli-Arab population. Journal of Rheumatology, 2000, 27, 1492-5.	1.0	23
72	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22

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73	CSF levels of carnitine in children with meningitis, neurologic disorders, acute gastroenteritis, and seizure. Neurology, 1998, 50, 1869-1871.	1.5	21
74	Direct detection of common mutations in the familial Mediterranean fever gene (MEFV) using naturally occurring and primer mediated restriction fragment analysis. , 1999, 14, 91-91.		21
75	No live individual homozygous for a novel endoglin mutation was found in a consanguineous Arab family with hereditary haemorrhagic telangiectasia. Journal of Medical Genetics, 2004, 41, e119-e119.	1.5	21
76	Autosomal recessive posterior column ataxia with retinitis pigmentosa caused by novel mutations in theFLVCR1gene. International Journal of Neuroscience, 2015, 125, 43-49.	0.8	21
77	Multigenerational autosomal dominant inheritance of 5p chromosomal deletions. American Journal of Medical Genetics, Part A, 2016, 170, 583-593.	0.7	21
78	Functional and epigenetic phenotypes of humans and mice with DNMT3A Overgrowth Syndrome. Nature Communications, 2021, 12, 4549.	5.8	21
79	Autoantibodies against bactericidal/permeabilityâ€“increasing protein (BPIâ€“ANCA) in cystic fibrosis patients treated with azithromycin. Clinical and Experimental Medicine, 2005, 5, 80-85.	1.9	20
80	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
81	Recognition of Smith-Lemli-Opitz syndrome (RSH) in the fetus: Utility of ultrasonography and biochemical analysis in pregnancies with low maternal serum estriol. American Journal of Medical Genetics, Part A, 2005, 138A, 56-60.	0.7	19
82	Splicing mutation in the fibrillin-1 gene associated with neonatal Marfan syndrome and severe pulmonary emphysema with tracheobronchomalacia. Pediatric Pulmonology, 2005, 39, 374-378.	1.0	19
83	Multiple ganglion cysts (â€˜cystic ganglionosisâ€™): an unusual presentation in a child. Scandinavian Journal of Rheumatology, 2007, 36, 145-148.	0.6	19
84	The Xp contiguous deletion syndrome and autism. American Journal of Medical Genetics, Part A, 2009, 149A, 1138-1148.	0.7	19
85	Brain MRI abnormalities and spectrum of neurological and clinical findings in three patients with proximal 16p11.2 microduplication. American Journal of Medical Genetics, Part A, 2014, 164, 2003-2012.	0.7	19
86	Survival among children with â€œLethalâ€•congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene (GLDN). Human Mutation, 2017, 38, 1477-1484.	1.1	19
87	Mutations in the <scp>PH</scp> Domain of <scp>DNM</scp>1 are associated with a nonepileptic phenotype characterized by developmental delay and neurobehavioral abnormalities. Molecular Genetics & Genomic Medicine, 2018, 6, 294-300.	0.6	19
88	De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome. American Journal of Human Genetics, 2019, 104, 542-552.	2.6	19
89	Intragenic <scp>CAMTA1</scp> deletions are associated with a spectrum of neurobehavioral phenotypes. Clinical Genetics, 2015, 87, 478-482.	1.0	18
90	<scp>FBXL4</scp> defects are common in patients with congenital lactic acidemia and encephalomyopathic mitochondrial <scp>DNA</scp> depletion syndrome. Clinical Genetics, 2017, 91, 634-639.	1.0	18

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91	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	1.8	18
92	CEDNIK. Child Neurology Open, 2017, 4, 2329048X1773321.	0.5	16
93	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143.	0.7	16
94	Familial mediterranean fever: The segregation of four different mutations in 13 individuals from one inbred family: Genotype-phenotype correlation and intrafamilial variability. American Journal of Medical Genetics Part A, 2002, 109, 198-201.	2.4	15
95	Delineation of the proximal 3q microdeletion syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 1729-1735.	0.7	15
96	Heterozygous 24-polyalanine repeats in the PHOX2B gene with different manifestations across three generations. Pediatric Pulmonology, 2014, 49, E13-E16.	1.0	15
97	Variants in DOCK3 cause developmental delay and hypotonia. European Journal of Human Genetics, 2019, 27, 1225-1234.	1.4	15
98	Hyperhomocysteinemia and cobalamin disorders. Molecular Genetics and Metabolism, 2007, 90, 113-121.	0.5	14
99	Acute Intermittent Porphyria. Journal of Child Neurology, 2012, 27, 917-921.	0.7	14
100	Pathogenic variants in TNRC6B cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. Journal of Medical Genetics, 2020, 57, 717-724.	1.5	14
101	Lymphedema of the Lower Extremity: Is It Genetic or Nongenetic?. Clinical Pediatrics, 2007, 46, 835-841.	0.4	13
102	15q13q14 deletions: Phenotypic characterization and molecular delineation by comparative genomic hybridization. American Journal of Medical Genetics, Part A, 2008, 146A, 1933-1941.	0.7	13
103	Cobalamin F Disease Detected by Newborn Screening and Follow-up on a 14-Year-Old Patient. Pediatrics, 2011, 128, e1636-e1640.	1.0	13
104	Duplication of OCRL and adjacent genes associated with autism but not Lowe syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2602-2605.	0.7	13
105	Transient Massive Trimethylaminuria Associated with Food Proteinâ€“Induced Enterocolitis Syndrome. JIMD Reports, 2013, 12, 11-15.	0.7	13
106	A mutation in Siteâ€1 Protease is associated with a complex phenotype that includes episodic hyperCKemia and focal myoedema. Molecular Genetics & Genomic Medicine, 2019, 7, e00733.	0.6	13
107	Crouzon syndrome: Association with absent pulmonary valve syndrome and severe tracheobronchomalacia. Pediatric Pulmonology, 2002, 34, 478-481.	1.0	12
108	Is this the Coffinâ€“Siris syndrome or the BOD syndrome?. American Journal of Medical Genetics, Part A, 2009, 149A, 559-562.	0.7	12

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109	ADULT syndrome due to an R243W mutation in TP63. International Journal of Dermatology, 2012, 51, 693-696.	0.5	12
110	Pulmonary manifestations and function tests in children genetically diagnosed with FMF. Pediatric Pulmonology, 2003, 35, 452-455.	1.0	11
111	DeSanto-Shinawi Syndrome: First Case in South America. Molecular Syndromology, 2018, 9, 154-158.	0.3	11
112	Functional characterization of biallelic RTTN variants identified in an infant with microcephaly, simplified gyral pattern, pontocerebellar hypoplasia, and seizures. Pediatric Research, 2018, 84, 435-441.	1.1	11
113	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. Bone, 2019, 120, 354-363.	1.4	11
114	Intragenic CNTN4 copy number variants associated with a spectrum of neurobehavioral phenotypes. European Journal of Medical Genetics, 2020, 63, 103736.	0.7	11
115	Overcoming presynaptic effects of VAMP2 mutations with 4â€aminopyridine treatment. Human Mutation, 2020, 41, 1999-2011.	1.1	11
116	Duplication of 20p12.3 associated with familial Wolffâ€“Parkinsonâ€“White syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 137-144.	0.7	10
117	Child Neurology: Brown-Vialetto-Van Laere syndrome. Neurology, 2018, 91, 938-941.	1.5	10
118	Sorting nexin 27 (SNX27) variants associated with seizures, developmental delay, behavioral disturbance, and subcortical brain abnormalities. Clinical Genetics, 2020, 97, 437-446.	1.0	10
119	Wilms tumor in patients with osteopathia striata with cranial sclerosis. European Journal of Human Genetics, 2021, 29, 396-401.	1.4	10
120	New Cohort of Patients With CEDNIK Syndrome Expands the Phenotypic and Genotypic Spectra. Neurology: Genetics, 2021, 7, e553.	0.9	10
121	Novel exon-skipping variant disrupting the basic domain of HCFC1 causes intellectual disability without metabolic abnormalities in both male and female patients. Journal of Human Genetics, 2021, 66, 717-724.	1.1	10
122	Identification of disease-linked hyperactivating mutations in UBE3A through large-scale functional variant analysis. Nature Communications, 2021, 12, 6809.	5.8	10
123	Phenotype and response to growth hormone therapy in siblings with B4GALT7 deficiency. Bone, 2019, 124, 14-21.	1.4	9
124	DNMT3Aovergrowth syndrome is associated with the development of hematopoietic malignancies in children and young adults. Blood, 2022, 139, 461-464.	0.6	9
125	Digynic triploidy: utility and challenges of noninvasive prenatal testing. Clinical Case Reports (discontinued), 2015, 3, 406-410.	0.2	8
126	A phase 1/2 open label nonrandomized clinical trial of intravenous 2-hydroxypropyl-Î²-cyclodextrin for acute liver disease in infants with Niemann-Pick C1. Molecular Genetics and Metabolism Reports, 2021, 28, 100772.	0.4	8

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127	Semaphorin-Plexin Signaling: From Axonal Guidance to a New X-Linked Intellectual Disability Syndrome. Pediatric Neurology, 2022, 126, 65-73.	1.0	8
128	Molecular and phenotypic characterization of atypical Williamsâ€“Beuren syndrome. Clinical Genetics, 2014, 86, 487-491.	1.0	7
129	De novo missense variants in FBXO11 alter its protein expression and subcellular localization. Human Molecular Genetics, 2022, 31, 440-454.	1.4	7
130	Preaxial polydactyly in neurofibromatosis 1. Clinical Dysmorphology, 2007, 16, 193-194.	0.1	6
131	McCune-Albright syndrome presenting with unilateral macroorchidism and bilateral testicular masses. Pediatric Radiology, 2010, 40, 16-20.	1.1	6
132	Earlyâ€onset Hepatic Fibrosis in Lysinuric Protein Intolerance. Journal of Pediatric Gastroenterology and Nutrition, 2011, 53, 695-698.	0.9	6
133	FGFR3-related condition: a skeletal dysplasia with similarities to thanatophoric dysplasia and SADDAN due to Lys650Met. Skeletal Radiology, 2015, 44, 441-445.	1.2	5
134	Abnormally increased carotid intima media-thickness and elasticity in patients with Morquio A disease. Orphanet Journal of Rare Diseases, 2020, 15, 73.	1.2	5
135	Biallelic variants in RNU12 cause CDAGS syndrome. Human Mutation, 2021, 42, 1042-1052.	1.1	5
136	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 1912-1921.	1.1	5
137	Biallelic <scp>ASCC1</scp> variants including a novel intronic variant result in expanded phenotypic spectrum of spinal muscular atrophy with congenital bone fractures 2 (<scp>SMABF2</scp>). American Journal of Medical Genetics, Part A, 2021, 185, 2190-2197.	0.7	4
138	Autosomal Dominant ANO5-Related Disorder Associated With Myopathy and Gnathodiaphyseal Dysplasia. Neurology: Genetics, 2021, 7, e612.	0.9	4
139	Increased Homocysteine in a Patient Diagnosed with Marfan Syndrome. Clinical Chemistry, 2010, 56, 1665-1668.	1.5	3
140	Inherited Deletion of 1q, Hyperparathyroidism and Signs of Y-chromosomal Influence in a Patient with Turner Syndrome. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 88-93.	0.4	3
141	Progressive Myopathy With Multiple Symmetric Lipomatosis. Archives of Neurology, 2009, 66, 1576-7.	4.9	2
142	Birth Defects Among 788 Children Born to Gulf War Veterans Based on Physical Examination. Journal of Occupational and Environmental Medicine, 2019, 61, 263-270.	0.9	2
143	Delineation of the 1q24.3 microdeletion syndrome provides further evidence for the potential role of non-coding RNAs in regulating the skeletal phenotype. Bone, 2021, 142, 115705.	1.4	2
144	The differential contribution of MEFV mutant alleles to the clinical profile of familial Mediterranean fever. , 0, .		2

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145	De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. American Journal of Human Genetics, 2015, 96, 1009.	2.6	1
146	Adult-onset dystonia with marfanoid features. Neurology: Clinical Practice, 2017, 7, e31-e34.	0.8	1
147	Special Therapy and Psychosocial Needs Identified in a Multidisciplinary Cancer Predisposition Syndrome Clinic. Journal of Pediatric Hematology/Oncology, 2019, 41, 133-136.	0.3	1
148	Response to Mounts and Besser. Genetics in Medicine, 2021, 23, 240-242.	1.1	1
149	Paroxysmal Kinesigenic Dyskinesia in Twins With Chromosome 16p11.2 Duplication Syndrome. Neurology: Genetics, 2021, 7, e549.	0.9	1
150	Extensive primary cutaneous herpes simplex virus type 1 infection in an infant following acute rotavirus gastroenteritis. European Journal of Pediatrics, 2005, 164, 175-176.	1.3	0
151	A 5-Month-Old Boy with Delay in Growth and Development and Decreased Muscle Tone. Clinical Chemistry, 2015, 61, 50-54.	1.5	0
152	Support for the Diagnosis of CHARGE Syndromeâ€"Reply. JAMA Otolaryngology - Head and Neck Surgery, 2017, 143, 635.	1.2	0
153	Known and Possible Roles of Epigenetics in Autism. , 2011, , 737-755.		0