

Marwan S Shinawi

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

150
papers

6,325
citations

36
h-index

77
g-index

162
ext. papers

7,557
ext. citations

6.7
avg, IF

5.06
L-index

#	Paper	IF	Citations
150	Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. <i>Nature Genetics</i> , 2009 , 41, 829-32	36.3	507
149	Prader-Willi phenotype caused by paternal deficiency for the HBII-85 C/D box small nucleolar RNA cluster. <i>Nature Genetics</i> , 2008 , 40, 719-21	36.3	458
148	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. <i>Nature Genetics</i> , 2008 , 40, 1466-71	36.3	457
147	A phase II/III clinical study of enzyme replacement therapy with idursulfase in mucopolysaccharidosis II (Hunter syndrome). <i>Genetics in Medicine</i> , 2006 , 8, 465-73	8.1	416
146	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. <i>Journal of Medical Genetics</i> , 2010 , 47, 332-41	5.8	362
145	Microdeletion 15q13.3: a locus with incomplete penetrance for autism, mental retardation, and psychiatric disorders. <i>Journal of Medical Genetics</i> , 2009 , 46, 382-8	5.8	191
144	TBX6 null variants and a common hypomorphic allele in congenital scoliosis. <i>New England Journal of Medicine</i> , 2015 , 372, 341-50	59.2	171
143	STXBP1 encephalopathy: A neurodevelopmental disorder including epilepsy. <i>Neurology</i> , 2016 , 86, 954-62.5	6.5	159
142	Long-term, open-labeled extension study of idursulfase in the treatment of Hunter syndrome. <i>Genetics in Medicine</i> , 2011 , 13, 95-101	8.1	156
141	A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. <i>Nature Genetics</i> , 2009 , 41, 1269-71	36.3	155
140	The array CGH and its clinical applications. <i>Drug Discovery Today</i> , 2008 , 13, 760-70	8.8	145
139	CpG Island Hypermethylation Mediated by DNMT3A Is a Consequence of AML Progression. <i>Cell</i> , 2017 , 168, 801-816.e13	56.2	131
138	Familial Mediterranean fever: prevalence, penetrance and genetic drift. <i>European Journal of Human Genetics</i> , 2001 , 9, 634-7	5.3	131
137	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. <i>Human Mutation</i> , 2010 , 31, 840-50	4.7	101
136	The differential contribution of MEFV mutant alleles to the clinical profile of familial Mediterranean fever. <i>European Journal of Human Genetics</i> , 2002 , 10, 145-9	5.3	97
135	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012 , 109, 7974-81.5	11.5	94
134	Microdeletions including YWHAE in the Miller-Dieker syndrome region on chromosome 17p13.3 result in facial dysmorphisms, growth restriction, and cognitive impairment. <i>Journal of Medical Genetics</i> , 2009 , 46, 825-33	5.8	92

133	Mitochondrial neurogastrointestinal encephalopathy due to mutations in RRM2B. <i>Archives of Neurology</i> , 2009 , 66, 1028-32		86
132	Familial Mediterranean fever: clinical and genetic characterization in a mixed pediatric population of Jewish and Arab patients. <i>Pediatrics</i> , 1999 , 103, e70	7.4	85
131	De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. <i>American Journal of Human Genetics</i> , 2015 , 96, 1009	11	78
130	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. <i>Nature Communications</i> , 2019 , 10, 3094	17.4	76
129	11p14.1 microdeletions associated with ADHD, autism, developmental delay, and obesity. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 1272-80	2.5	69
128	Clinical Characterization of Patients With Autosomal Dominant Short Stature due to Aggrecan Mutations. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017 , 102, 460-469	5.6	68
127	Syndromic thrombocytopenia and predisposition to acute myelogenous leukemia caused by constitutional microdeletions on chromosome 21q. <i>Blood</i> , 2008 , 112, 1042-7	2.2	64
126	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. <i>American Journal of Human Genetics</i> , 2018 , 102, 27-43	11	61
125	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. <i>Human Molecular Genetics</i> , 2011 , 20, 1975-88	5.6	61
124	The musculoskeletal manifestations of familial Mediterranean fever in children genetically diagnosed with the disease. <i>Arthritis and Rheumatism</i> , 2001 , 44, 1416-9		60
123	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. <i>Genetics in Medicine</i> , 2017 , 19, 1040-1048	8.1	58
122	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. <i>American Journal of Human Genetics</i> , 2016 , 98, 347-57	11	58
121	Neurologin 2 nonsense variant associated with anxiety, autism, intellectual disability, hyperphagia, and obesity. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 213-216	2.5	51
120	The Cognitive and Behavioral Phenotypes of Individuals with CHRNA7 Duplications. <i>Journal of Autism and Developmental Disorders</i> , 2017 , 47, 549-562	4.6	44
119	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotype-phenotype correlations. <i>Genetics in Medicine</i> , 2016 , 18, 1143-1150	8.1	42
118	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. <i>Brain</i> , 2018 , 141, 1998-2013	11.2	42
117	The MTHFR 677C-->T polymorphism and behaviors in children with autism: exploratory genotype-phenotype correlations. <i>Autism Research</i> , 2009 , 2, 98-108	5.1	42
116	Expanding the clinical spectrum of the 16p11.2 chromosomal rearrangements: three patients with syringomyelia. <i>European Journal of Human Genetics</i> , 2011 , 19, 152-6	5.3	41

115	Desmosterolosis-phenotypic and molecular characterization of a third case and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 1597-604	2.5	40
114	Mutations in COQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. <i>Journal of Medical Genetics</i> , 2015 , 52, 627-35	5.8	36
113	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. <i>Brain</i> , 2018 , 141, 2299-2311	11.2	36
112	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. <i>Human Mutation</i> , 2012 , 33, 165-79	4.7	36
111	TSHZ3 deletion causes an autism syndrome and defects in cortical projection neurons. <i>Nature Genetics</i> , 2016 , 48, 1359-1369	36.3	36
110	A Novel Mutation in Isoform 3 of the Plasma Membrane Ca ²⁺ Pump Impairs Cellular Ca ²⁺ Homeostasis in a Patient with Cerebellar Ataxia and Laminin Subunit 1 Mutations. <i>Journal of Biological Chemistry</i> , 2015 , 290, 16132-41	5.4	35
109	Diagnostic testing for uniparental disomy: a points to consider statement from the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2020 , 22, 1133-1141	8.1	35
108	Dosage changes of a segment at 17p13.1 lead to intellectual disability and microcephaly as a result of complex genetic interaction of multiple genes. <i>American Journal of Human Genetics</i> , 2014 , 95, 565-78 ¹¹	11	34
107	Multi-systemic involvement in NGLY1-related disorder caused by two novel mutations. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 816-20	2.5	34
106	De novo mutations in SIK1 cause a spectrum of developmental epilepsies. <i>American Journal of Human Genetics</i> , 2015 , 96, 682-90	11	31
105	NR2F1 haploinsufficiency is associated with optic atrophy, dysmorphism and global developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 377-81	2.5	31
104	WAC loss-of-function mutations cause a recognisable syndrome characterised by dysmorphic features, developmental delay and hypotonia and recapitulate 10p11.23 microdeletion syndrome. <i>Journal of Medical Genetics</i> , 2015 , 52, 754-61	5.8	30
103	Low-level mosaicism of trisomy 14: phenotypic and molecular characterization. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 1395-405	2.5	30
102	Scoliosis and vertebral anomalies: additional abnormal phenotypes associated with chromosome 16p11.2 rearrangement. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1118-26	2.5	28
101	Two novel RAD21 mutations in patients with mild Cornelia de Lange syndrome-like presentation and report of the first familial case. <i>Gene</i> , 2014 , 537, 279-84	3.8	25
100	The spectrum of DNMT3A variants in Tatton-Brown-Rahman syndrome overlaps with that in hematologic malignancies. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 3022-3028	2.5	24
99	Haploinsufficiency of ZNF238 is associated with corpus callosum abnormalities in 1q44 deletions. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 711-6	2.5	24
98	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. <i>European Journal of Human Genetics</i> , 2019 , 27, 1611-1618	5.3	22

97	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. <i>American Journal of Human Genetics</i> , 2020 , 106, 234-245	11	22
96	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018 , 103, 305-316	11	21
95	CSF levels of carnitine in children with meningitis, neurologic disorders, acute gastroenteritis, and seizure. <i>Neurology</i> , 1998 , 50, 1869-71	6.5	21
94	Familial Mediterranean fever: high gene frequency and heterogeneous disease among an Israeli-Arab population. <i>Journal of Rheumatology</i> , 2000 , 27, 1492-5	4.1	20
93	Mixed gonadal dysgenesis in a child with isodicentric Y chromosome: Does the relative proportion of the 45,X line really matter?. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 1832-7	2.5	19
92	Direct detection of common mutations in the familial Mediterranean fever gene (MEFV) using naturally occurring and primer mediated restriction fragment analysis. Mutation in brief no. 257. Online. <i>Human Mutation</i> , 1999 , 14, 91	4.7	19
91	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. <i>Genetics in Medicine</i> , 2019 , 21, 2723-2733	8.1	18
90	Prevalence of Semicircular Canal Hypoplasia in Patients With CHARGE Syndrome: 3C Syndrome. <i>JAMA Otolaryngology - Head and Neck Surgery</i> , 2017 , 143, 168-177	3.9	18
89	Atypical presentation of VLCAD deficiency associated with a novel ACADVL splicing mutation. <i>Muscle and Nerve</i> , 2009 , 39, 374-82	3.4	18
88	No live individual homozygous for a novel endoglin mutation was found in a consanguineous Arab family with hereditary haemorrhagic telangiectasia. <i>Journal of Medical Genetics</i> , 2004 , 41, e119	5.8	18
87	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. <i>Neurogenetics</i> , 2016 , 17, 173-8	3	18
86	Heterozygous variants in ACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. <i>Human Mutation</i> , 2017 , 38, 1365-1371	4.7	17
85	The Xp contiguous deletion syndrome and autism. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 1138-48	2.5	17
84	Recognition of Smith-Lemli-Opitz syndrome (RSH) in the fetus: utility of ultrasonography and biochemical analysis in pregnancies with low maternal serum estriol. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 138, 56-60	2.5	17
83	Autosomal recessive posterior column ataxia with retinitis pigmentosa caused by novel mutations in the FLVCR1 gene. <i>International Journal of Neuroscience</i> , 2015 , 125, 43-9	2	16
82	2-Pyrrolidinone and Succinimide as Clinical Screening Biomarkers for GABA-Transaminase Deficiency: Anti-seizure Medications Impact Accurate Diagnosis. <i>Frontiers in Neuroscience</i> , 2019 , 13, 394	5.1	15
81	Delineation of the proximal 3q microdeletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 1729-35	2.5	15
80	Splicing mutation in the fibrillin-1 gene associated with neonatal Marfan syndrome and severe pulmonary emphysema with tracheobronchomalacia. <i>Pediatric Pulmonology</i> , 2005 , 39, 374-8	3.5	15

79	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. <i>Human Mutation</i> , 2018 , 39, 1875-1884	4.7	15
78	Heterozygous 24-polyalanine repeats in the PHOX2B gene with different manifestations across three generations. <i>Pediatric Pulmonology</i> , 2014 , 49, E13-6	3.5	14
77	Survival among children with "Lethal" congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene (GLDN). <i>Human Mutation</i> , 2017 , 38, 1477-1484	4.7	14
76	FBXL4 defects are common in patients with congenital lactic acidemia and encephalomyopathic mitochondrial DNA depletion syndrome. <i>Clinical Genetics</i> , 2017 , 91, 634-639	4	14
75	Multiple ganglion cysts (Rystic ganglionosis): an unusual presentation in a child. <i>Scandinavian Journal of Rheumatology</i> , 2007 , 36, 145-8	1.9	14
74	CEDNIK: Phenotypic and Molecular Characterization of an Additional Patient and Review of the Literature. <i>Child Neurology Open</i> , 2017 , 4, 2329048X17733214	1.3	13
73	Acute intermittent porphyria: a diagnostic challenge. <i>Journal of Child Neurology</i> , 2012 , 27, 917-21	2.5	13
72	15q13q14 deletions: phenotypic characterization and molecular delineation by comparative genomic hybridization. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 1933-41	2.5	13
71	Familial Mediterranean fever: the segregation of four different mutations in 13 individuals from one inbred family: genotype-phenotype correlation and intrafamilial variability. <i>American Journal of Medical Genetics Part A</i> , 2002 , 109, 198-201		13
70	Autoantibodies against bactericidal/permeability-increasing protein (BPI-ANCA) in cystic fibrosis patients treated with azithromycin. <i>Clinical and Experimental Medicine</i> , 2005 , 5, 80-5	4.9	13
69	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). <i>Bone</i> , 2018 , 107, 161-171	4.7	13
68	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. <i>Clinical Epigenetics</i> , 2019 , 11, 60	7.7	12
67	Hyperhomocysteinemia and cobalamin disorders. <i>Molecular Genetics and Metabolism</i> , 2007 , 90, 113-21	3.7	12
66	Pathogenic Variants in GPC4 Cause Keipert Syndrome. <i>American Journal of Human Genetics</i> , 2019 , 104, 914-924	11	11
65	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. <i>Human Genetics</i> , 2020 , 139, 575-592	6.3	11
64	Transient massive trimethylaminuria associated with food protein-induced enterocolitis syndrome. <i>JIMD Reports</i> , 2014 , 12, 11-5	1.9	11
63	Immunodeficiency and bone marrow failure with mosaic and germline TLR8 gain of function. <i>Blood</i> , 2021 , 137, 2450-2462	2.2	11
62	Multigenerational autosomal dominant inheritance of 5p chromosomal deletions. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 583-93	2.5	11

61	De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome. <i>American Journal of Human Genetics</i> , 2019 , 104, 542-552	11	11
60	Intragenic CAMTA1 deletions are associated with a spectrum of neurobehavioral phenotypes. <i>Clinical Genetics</i> , 2015 , 87, 478-82	4	10
59	Duplication of 20p12.3 associated with familial Wolff-Parkinson-White syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 137-44	2.5	10
58	Duplication of OCRL and adjacent genes associated with autism but not Lowe syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 2602-5	2.5	10
57	Is this the Coffin-Siris syndrome or the BOD syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 559-62	2.5	10
56	Crouzon syndrome: association with absent pulmonary valve syndrome and severe tracheobronchomalacia. <i>Pediatric Pulmonology</i> , 2002 , 34, 478-81	3.5	10
55	Mutations in the PH Domain of DNM1 are associated with a nonepileptic phenotype characterized by developmental delay and neurobehavioral abnormalities. <i>Molecular Genetics & Genomic Medicine</i> , 2018 , 6, 294-300	2.3	10
54	Brain MRI abnormalities and spectrum of neurological and clinical findings in three patients with proximal 16p11.2 microduplication. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 2003-12	2.5	9
53	ADULT syndrome due to an R243W mutation in TP63. <i>International Journal of Dermatology</i> , 2012 , 51, 693-6	1.7	9
52	Cobalamin F disease detected by newborn screening and follow-up on a 14-year-old patient. <i>Pediatrics</i> , 2011 , 128, e1636-40	7.4	9
51	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. <i>Neurogenetics</i> , 2019 , 20, 129-143	3	8
50	Lymphedema of the lower extremity: is it genetic or nongenetic?. <i>Clinical Pediatrics</i> , 2007 , 46, 835-41	1.2	8
49	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. <i>Genetics in Medicine</i> , 2020 , 22, 1215-1226	8.1	7
48	Digynic triploidy: utility and challenges of noninvasive prenatal testing. <i>Clinical Case Reports (discontinued)</i> , 2015 , 3, 406-10	0.7	7
47	Pulmonary manifestations and function tests in children genetically diagnosed with FMF. <i>Pediatric Pulmonology</i> , 2003 , 35, 452-5	3.5	7
46	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the CLC-6 Cl/H-Exchanger, Causes Early-Onset Neurodegeneration. <i>American Journal of Human Genetics</i> , 2020 , 107, 1062-1077	11	7
45	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. <i>Bone</i> , 2019 , 120, 354-363	4.7	7
44	DeSanto-Shinawi Syndrome: First Case in South America. <i>Molecular Syndromology</i> , 2018 , 9, 154-158	1.5	7

43	A mutation in Site-1 Protease is associated with a complex phenotype that includes episodic hyperCKemia and focal myoedema. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e00733	2.3	6
42	Phenotype and response to growth hormone therapy in siblings with B4GALT7 deficiency. <i>Bone</i> , 2019 , 124, 14-21	4.7	6
41	Variants in DOCK3 cause developmental delay and hypotonia. <i>European Journal of Human Genetics</i> , 2019 , 27, 1225-1234	5.3	6
40	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. <i>Genetics in Medicine</i> , 2019 , 21, 2036-2042	8.1	6
39	Pathogenic variants in cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. <i>Journal of Medical Genetics</i> , 2020 , 57, 717-724	5.8	6
38	Molecular and phenotypic characterization of atypical Williams-Beuren syndrome. <i>Clinical Genetics</i> , 2014 , 86, 487-91	4	6
37	Early-onset hepatic fibrosis in lysinuric protein intolerance. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2011 , 53, 695-8	2.8	6
36	McCune-Albright syndrome presenting with unilateral macroorchidism and bilateral testicular masses. <i>Pediatric Radiology</i> , 2010 , 40 Suppl 1, S16-20	2.8	6
35	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. <i>American Journal of Human Genetics</i> , 2020 , 107, 311-324	11	6
34	Wilms tumor in patients with osteopathia striata with cranial sclerosis. <i>European Journal of Human Genetics</i> , 2021 , 29, 396-401	5.3	6
33	Functional characterization of biallelic RTTN variants identified in an infant with microcephaly, simplified gyral pattern, pontocerebellar hypoplasia, and seizures. <i>Pediatric Research</i> , 2018 , 84, 435-441	3.2	6
32	FGFR3-related condition: a skeletal dysplasia with similarities to thanatophoric dysplasia and SADDAN due to Lys650Met. <i>Skeletal Radiology</i> , 2015 , 44, 441-5	2.7	5
31	Preaxial polydactyly in neurofibromatosis 1. <i>Clinical Dysmorphology</i> , 2007 , 16, 193-194	0.9	5
30	Sorting nexin 27 (SNX27) variants associated with seizures, developmental delay, behavioral disturbance, and subcortical brain abnormalities. <i>Clinical Genetics</i> , 2020 , 97, 437-446	4	5
29	Intragenic CNTN4 copy number variants associated with a spectrum of neurobehavioral phenotypes. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103736	2.6	5
28	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , 2021 , 23, 653-660	8.1	5
27	Child Neurology: Brown-Vialetto-Van Laere syndrome: Dramatic visual recovery after delayed riboflavin therapy. <i>Neurology</i> , 2018 , 91, 938-941	6.5	5
26	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. <i>Genetics in Medicine</i> , 2021 , 23, 1474-1483	8.1	4

25	Increased homocysteine in a patient diagnosed with Marfan syndrome. <i>Clinical Chemistry</i> , 2010 , 56, 1665-8	3
24	Overcoming presynaptic effects of VAMP2 mutations with 4-aminopyridine treatment. <i>Human Mutation</i> , 2020 , 41, 1999-2011	4.7 3
23	New Cohort of Patients With CEDNIK Syndrome Expands the Phenotypic and Genotypic Spectra. <i>Neurology: Genetics</i> , 2021 , 7, e553	3.8 3
22	A phase 1/2 open label nonrandomized clinical trial of intravenous 2-hydroxypropyl-β-cyclodextrin for acute liver disease in infants with Niemann-Pick C1. <i>Molecular Genetics and Metabolism Reports</i> , 2021 , 28, 100772	1.8 3
21	Abnormally increased carotid intima media-thickness and elasticity in patients with Morquio A disease. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 73	4.2 2
20	Progressive myopathy with multiple symmetric lipomatosis. <i>Archives of Neurology</i> , 2009 , 66, 1576-7	2
19	The differential contribution of MEFV mutant alleles to the clinical profile of familial Mediterranean fever	2
18	Functional and epigenetic phenotypes of humans and mice with DNMT3A Overgrowth Syndrome. <i>Nature Communications</i> , 2021 , 12, 4549	17.4 2
17	Novel exon-skipping variant disrupting the basic domain of HCFC1 causes intellectual disability without metabolic abnormalities in both male and female patients. <i>Journal of Human Genetics</i> , 2021 , 66, 717-724	4.3 2
16	Birth Defects Among 788 Children Born to Gulf War Veterans Based on Physical Examination. <i>Journal of Occupational and Environmental Medicine</i> , 2019 , 61, 263-270	2 1
15	Identification of disease-linked hyperactivating mutations in UBE3A through large-scale functional variant analysis. <i>Nature Communications</i> , 2021 , 12, 6809	17.4 1
14	Semaphorin-Plexin Signaling: From Axonal Guidance to a New X-Linked Intellectual Disability Syndrome. <i>Pediatric Neurology</i> , 2021 , 126, 65-73	2.9 1
13	Biallelic variants in RNU12 cause CDAGS syndrome. <i>Human Mutation</i> , 2021 , 42, 1042-1052	4.7 1
12	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2021 , 23, 1912-1921	8.1 1
11	Special Therapy and Psychosocial Needs Identified in a Multidisciplinary Cancer Predisposition Syndrome Clinic. <i>Journal of Pediatric Hematology/Oncology</i> , 2019 , 41, 133-136	1.2 1
10	Delineation of the 1q24.3 microdeletion syndrome provides further evidence for the potential role of non-coding RNAs in regulating the skeletal phenotype. <i>Bone</i> , 2021 , 142, 115705	4.7 1
9	Response to Mounds and Besser. <i>Genetics in Medicine</i> , 2021 , 23, 240-242	8.1 1
8	Paroxysmal Kinesigenic Dyskinesia in Twins With Chromosome 16p11.2 Duplication Syndrome. <i>Neurology: Genetics</i> , 2021 , 7, e549	3.8 1

7	Adult-onset dystonia with marfanoid features. <i>Neurology: Clinical Practice</i> , 2017 , 7, e31-e34	1.7	○
6	Inherited Deletion of 1q, Hyperparathyroidism and Signs of Y-chromosomal Influence in a Patient with Turner Syndrome. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2019 , 11, 88-93	1.9	○
5	Biallelic ASCC1 variants including a novel intronic variant result in expanded phenotypic spectrum of spinal muscular atrophy with congenital bone fractures 2 (SMABF2). <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 2190-2197	2.5	○
4	Autosomal Dominant ANO5-Related Disorder Associated With Myopathy and Gnathodiaphyseal Dysplasia. <i>Neurology: Genetics</i> , 2021 , 7, e612	3.8	○
3	Support for the Diagnosis of CHARGE Syndrome-Reply. <i>JAMA Otolaryngology - Head and Neck Surgery</i> , 2017 , 143, 635	3.9	
2	A 5-month-old boy with delay in growth and development and decreased muscle tone. <i>Clinical Chemistry</i> , 2015 , 61, 50-4	5.5	
1	Extensive primary cutaneous herpes simplex virus type 1 infection in an infant following acute rotavirus gastroenteritis. <i>European Journal of Pediatrics</i> , 2005 , 164, 175-6	4.1	