Marwan S Shinawi

List of Publications by Year in Descending Order

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	Identification of disease-linked hyperactivating mutations in UBE3A through large-scale functional variant analysis. *Nature Communications*, **2021**, 12, 6809	17.4	1
149	Semaphorin-Plexin Signaling: From Axonal Guidance to a New X-Linked Intellectual Disability Syndrome. *Pediatric Neurology*, **2021**, 126, 65-73	2.9	1
148	Biallelic ASCC1 variants including a novel intronic variant result in expanded phenotypic spectrum of spinal muscular atrophy with congenital bone fractures 2 (SMABF2). *American Journal of Medical Genetics, Part A*, **2021**, 185, 2190-2197	2.5	0
147	Immunodeficiency and bone marrow failure with mosaic and germline TLR8 gain of function. *Blood*, **2021**, 137, 2450-2462	2.2	11
146	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	8.1	4
145	Biallelic variants in RNU12 cause CDAGS syndrome. *Human Mutation*, **2021**, 42, 1042-1052	4.7	1
144	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. *Genetics in Medicine*, **2021**, 23, 1912-1921	8.1	1
143	Functional and epigenetic phenotypes of humans and mice with DNMT3A Overgrowth Syndrome. *Nature Communications*, **2021**, 12, 4549	17.4	2
142	Autosomal Dominant ANO5-Related Disorder Associated With Myopathy and Gnathodiaphyseal Dysplasia. *Neurology: Genetics*, **2021**, 7, e612	3.8	0
141	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	4.7	1
140	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. *Genetics in Medicine*, **2021**, 23, 653-660	8.1	5
139	Wilms tumor in patients with osteopathia striata with cranial sclerosis. *European Journal of Human Genetics*, **2021**, 29, 396-401	5.3	6
138	Response to Mounts and Besser. *Genetics in Medicine*, **2021**, 23, 240-242	8.1	1
137	New Cohort of Patients With CEDNIK Syndrome Expands the Phenotypic and Genotypic Spectra. *Neurology: Genetics*, **2021**, 7, e553	3.8	3
136	Paroxysmal Kinesigenic Dyskinesia in Twins With Chromosome 16p11.2 Duplication Syndrome. *Neurology: Genetics*, **2021**, 7, e549	3.8	1
135	A phase 1/2 open label nonrandomized clinical trial of intravenous 2-hydroxypropyl-Ecyclodextrin for acute liver disease in infants with Niemann-Pick C1. *Molecular Genetics and Metabolism Reports*, **2021**, 28, 100772	1.8	3
134	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	4.3	2

133	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. *Genetics in Medicine*, **2020**, 22, 1215-1226	8.1	7
132	Abnormally increased carotid intima media-thickness and elasticity in patients with Morquio A disease. *Orphanet Journal of Rare Diseases*, **2020**, 15, 73	4.2	2
131	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. *Human Genetics*, **2020**, 139, 575-592	6.3	11
130	Pathogenic variants in 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	5.8	6
129	Sorting nexin 27 (SNX27) variants associated with seizures, developmental delay, behavioral disturbance, and subcortical brain abnormalities. *Clinical Genetics*, **2020**, 97, 437-446	4	5
128	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. *American Journal of Human Genetics*, **2020**, 106, 234-245	11	22
127	Overcoming presynaptic effects of VAMP2 mutations with 4-aminopyridine treatment. *Human Mutation*, **2020**, 41, 1999-2011	4.7	3
126	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Cl/H-Exchanger, Causes Early-Onset Neurodegeneration. *American Journal of Human Genetics*, **2020**, 107, 1062-1077	11	7
125	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	11	6
124	Intragenic CNTN4 copy number variants associated with a spectrum of neurobehavioral phenotypes. *European Journal of Medical Genetics*, **2020**, 63, 103736	2.6	5
123	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	8.1	35
122	2-Pyrrolidinone and Succinimide as Clinical Screening Biomarkers for GABA-Transaminase Deficiency: Anti-seizure Medications Impact Accurate Diagnosis. *Frontiers in Neuroscience*, **2019**, 13, 394	5.1	15
121	A mutation in Site-1 Protease is associated with a complex phenotype that includes episodic hyperCKemia and focal myoedema. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00733	2.3	6
120	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. *Neurogenetics*, **2019**, 20, 129-143	3	8
119	Phenotype and response to growth hormone therapy in siblings with B4GALT7 deficiency. *Bone*, **2019**, 124, 14-21	4.7	6
118	Birth Defects Among 788 Children Born to Gulf War Veterans Based on Physical Examination. *Journal of Occupational and Environmental Medicine*, **2019**, 61, 263-270	2	1
117	Variants in DOCK3 cause developmental delay and hypotonia. *European Journal of Human Genetics*, **2019**, 27, 1225-1234	5.3	6
116	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. *Clinical Epigenetics*, **2019**, 11, 60	7.7	12

115	Pathogenic Variants in GPC4 Cause Keipert Syndrome. *American Journal of Human Genetics*, **2019**, 104, 914-924	11	11
114	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. *Genetics in Medicine*, **2019**, 21, 2036-2042	8.1	6
113	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. *Nature Communications*, **2019**, 10, 3094	17.4	76
112	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. *European Journal of Human Genetics*, **2019**, 27, 1611-1618	5.3	22
111	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. *Genetics in Medicine*, **2019**, 21, 2723-2733	8.1	18
110	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	1.9	0
109	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	11	11
108	Special Therapy and Psychosocial Needs Identified in a Multidisciplinary Cancer Predisposition Syndrome Clinic. *Journal of Pediatric Hematology/Oncology*, **2019**, 41, 133-136	1.2	1
107	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. *Bone*, **2019**, 120, 354-363	4.7	7
106	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. *American Journal of Human Genetics*, **2018**, 102, 27-43	11	61
105	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	11	21
104	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. *Brain*, **2018**, 141, 2299-2311	11.2	36
103	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	11.2	42
102	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). *Bone*, **2018**, 107, 161-171	4.7	13
101	Child Neurology: Brown-Vialetto-Van Laere syndrome: Dramatic visual recovery after delayed riboflavin therapy. *Neurology*, **2018**, 91, 938-941	6.5	5
100	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. *Human Mutation*, **2018**, 39, 1875-1884	4.7	15
99	DeSanto-Shinawi Syndrome: First Case in South America. *Molecular Syndromology*, **2018**, 9, 154-158	1.5	7
98	Mutations in the PH Domain of DNM1 are associated with a nonepileptic phenotype characterized by developmental delay and neurobehavioral abnormalities. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 294-300	2.3	10

97	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	3.2	6
96	CpG Island Hypermethylation Mediated by DNMT3A Is a Consequence of AML Progression. *Cell*, **2017**, 168, 801-816.e13	56.2	131
95	Support for the Diagnosis of CHARGE Syndrome-Reply. *JAMA Otolaryngology - Head and Neck Surgery*, **2017**, 143, 635	3.9	
94	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. *Genetics in Medicine*, **2017**, 19, 1040-1048	8.1	58
93	Adult-onset dystonia with marfanoid features. *Neurology: Clinical Practice*, **2017**, 7, e31-e34	1.7	0
92	CEDNIK: Phenotypic and Molecular Characterization of an Additional Patient and Review of the Literature. *Child Neurology Open*, **2017**, 4, 2329048X17733214	1.3	13
91	The spectrum of DNMT3A variants in Tatton-Brown-Rahman syndrome overlaps with that in hematologic malignancies. *American Journal of Medical Genetics, Part A*, **2017**, 173, 3022-3028	2.5	24
90	Survival among children with "Lethal" congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene (GLDN). *Human Mutation*, **2017**, 38, 1477-1484	4.7	14
89	Heterozygous variants in ACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. *Human Mutation*, **2017**, 38, 1365-1371	4.7	17
88	Prevalence of Semicircular Canal Hypoplasia in Patients With CHARGE Syndrome: 3C Syndrome. *JAMA Otolaryngology - Head and Neck Surgery*, **2017**, 143, 168-177	3.9	18
87	The Cognitive and Behavioral Phenotypes of Individuals with CHRNA7 Duplications. *Journal of Autism and Developmental Disorders*, **2017**, 47, 549-562	4.6	44
86	Neuroligin 2 nonsense variant associated with anxiety, autism, intellectual disability, hyperphagia, and obesity. *American Journal of Medical Genetics, Part A*, **2017**, 173, 213-216	2.5	51
85	FBXL4 defects are common in patients with congenital lactic acidemia and encephalomyopathic mitochondrial DNA depletion syndrome. *Clinical Genetics*, **2017**, 91, 634-639	4	14
84	Clinical Characterization of Patients With Autosomal Dominant Short Stature due to Aggrecan Mutations. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 460-469	5.6	68
83	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotype-phenotype correlations. *Genetics in Medicine*, **2016**, 18, 1143-1150	8.1	42
82	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. *American Journal of Human Genetics*, **2016**, 98, 347-57	11	58
81	STXBP1 encephalopathy: A neurodevelopmental disorder including epilepsy. *Neurology*, **2016**, 86, 954-62	8.5	159
80	Multigenerational autosomal dominant inheritance of 5p chromosomal deletions. *American Journal of Medical Genetics, Part A*, **2016**, 170, 583-93	2.5	11

79	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. *Neurogenetics*, **2016**, 17, 173-8	3	18
78	TSHZ3 deletion causes an autism syndrome and defects in cortical projection neurons. *Nature Genetics*, **2016**, 48, 1359-1369	36.3	36
77	De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. *American Journal of Human Genetics*, **2015**, 96, 1009	11	78
76	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 1 Mutations. *Journal of Biological Chemistry*, **2015**, 290, 16132-41	5.4	35
75	De novo mutations in SIK1 cause a spectrum of developmental epilepsies. *American Journal of Human Genetics*, **2015**, 96, 682-90	11	31
74	A 5-month-old boy with delay in growth and development and decreased muscle tone. *Clinical Chemistry*, **2015**, 61, 50-4	5.5	
73	Intragenic CAMTA1 deletions are associated with a spectrum of neurobehavioral phenotypes. *Clinical Genetics*, **2015**, 87, 478-82	4	10
72	Mutations in COQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. *Journal of Medical Genetics*, **2015**, 52, 627-35	5.8	36
71	Digynic triploidy: utility and challenges of noninvasive prenatal testing. *Clinical Case Reports (discontinued)*, **2015**, 3, 406-10	0.7	7
70	Multi-systemic involvement in NGLY1-related disorder caused by two novel mutations. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 816-20	2.5	34
69	WAC loss-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, 754-61	5.8	30
68	TBX6 null variants and a common hypomorphic allele in congenital scoliosis. *New England Journal of Medicine*, **2015**, 372, 341-50	59.2	171
67	Autosomal recessive posterior column ataxia with retinitis pigmentosa caused by novel mutations in the FLVCR1 gene. *International Journal of Neuroscience*, **2015**, 125, 43-9	2	16
66	FGFR3-related condition: a skeletal dysplasia with similarities to thanatophoric dysplasia and SADDAN due to Lys650Met. *Skeletal Radiology*, **2015**, 44, 441-5	2.7	5
65	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**, 164A, 2003-12	2.5	9
64	Molecular and phenotypic characterization of atypical Williams-Beuren syndrome. *Clinical Genetics*, **2014**, 86, 487-91	4	6
63	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, 565-78	11	34
62	Heterozygous 24-polyalanine repeats in the PHOX2B gene with different manifestations across three generations. *Pediatric Pulmonology*, **2014**, 49, E13-6	3.5	14

61	Two novel RAD21 mutations in patients with mild Cornelia de Lange syndrome-like presentation and report of the first familial case. *Gene*, **2014**, 537, 279-84	3.8	25
60	Scoliosis and vertebral anomalies: additional abnormal phenotypes associated with chromosome 16p11.2 rearrangement. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1118-26	2.5	28
59	Transient massive trimethylaminuria associated with food protein-induced enterocolitis syndrome. *JIMD Reports*, **2014**, 12, 11-5	1.9	11
58	NR2F1 haploinsufficiency is associated with optic atrophy, dysmorphism and global developmental delay. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 377-81	2.5	31
57	Duplication of 20p12.3 associated with familial Wolff-Parkinson-White syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 137-44	2.5	10
56	Haploinsufficiency of ZNF238 is associated with corpus callosum abnormalities in 1q44 deletions. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 711-6	2.5	24
55	ADULT syndrome due to an R243W mutation in TP63. *International Journal of Dermatology*, **2012**, 51, 693-6	1.7	9
54	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, 165-79	4.7	36
53	Acute intermittent porphyria: a diagnostic challenge. *Journal of Child Neurology*, **2012**, 27, 917-21	2.5	13
52	Duplication of OCRL and adjacent genes associated with autism but not Lowe syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2602-5	2.5	10
51	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, 7974-81	11.5	94
50	Early-onset hepatic fibrosis in lysinuric protein intolerance. *Journal of Pediatric Gastroenterology and Nutrition*, **2011**, 53, 695-8	2.8	6
49	Expanding the clinical spectrum of the 16p11.2 chromosomal rearrangements: three patients with syringomyelia. *European Journal of Human Genetics*, **2011**, 19, 152-6	5.3	41
48	11p14.1 microdeletions associated with ADHD, autism, developmental delay, and obesity. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1272-80	2.5	69
47	Desmosterolosis-phenotypic and molecular characterization of a third case and review of the literature. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1597-604	2.5	40
46	Cobalamin F disease detected by newborn screening and follow-up on a 14-year-old patient. *Pediatrics*, **2011**, 128, e1636-40	7.4	9
45	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. *Human Molecular Genetics*, **2011**, 20, 1975-88	5.6	61
44	Long-term, open-labeled extension study of idursulfase in the treatment of Hunter syndrome. *Genetics in Medicine*, **2011**, 13, 95-101	8.1	156

43	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. *Journal of Medical Genetics*, **2010**, 47, 332-41	5.8	362
42	Increased homocysteine in a patient diagnosed with Marfan syndrome. *Clinical Chemistry*, **2010**, 56, 1665-8	5.8	3
41	McCune-Albright syndrome presenting with unilateral macroorchidism and bilateral testicular masses. *Pediatric Radiology*, **2010**, 40 Suppl 1, S16-20	2.8	6
40	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. *Human Mutation*, **2010**, 31, 840-50	4.7	101
39	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, 1832-7	2.5	19
38	Progressive myopathy with multiple symmetric lipomatosis. *Archives of Neurology*, **2009**, 66, 1576-7		2
37	Mitochondrial neurogastrointestinal encephalopathy due to mutations in RRM2B. *Archives of Neurology*, **2009**, 66, 1028-32		86
36	Is this the Coffin-Siris syndrome or the BOD syndrome?. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 559-62	2.5	10
35	The Xp contiguous deletion syndrome and autism. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1138-48	2.5	17
34	Atypical presentation of VLCAD deficiency associated with a novel ACADVL splicing mutation. *Muscle and Nerve*, **2009**, 39, 374-82	3.4	18
33	Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. *Nature Genetics*, **2009**, 41, 829-32	36.3	507
32	A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. *Nature Genetics*, **2009**, 41, 1269-71	36.3	155
31	The MTHFR 677C-->T polymorphism and behaviors in children with autism: exploratory genotype-phenotype correlations. *Autism Research*, **2009**, 2, 98-108	5.1	42
30	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, 825-33	5.8	92
29	Microdeletion 15q13.3: a locus with incomplete penetrance for autism, mental retardation, and psychiatric disorders. *Journal of Medical Genetics*, **2009**, 46, 382-8	5.8	191
28	Prader-Willi phenotype caused by paternal deficiency for the HBII-85 C/D box small nucleolar RNA cluster. *Nature Genetics*, **2008**, 40, 719-21	36.3	458
27	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. *Nature Genetics*, **2008**, 40, 1466-71	36.3	457
26	The array CGH and its clinical applications. *Drug Discovery Today*, **2008**, 13, 760-70	8.8	145

25	15q13q14 deletions: phenotypic characterization and molecular delineation by comparative genomic hybridization. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1933-41	2.5	13
24	Syndromic thrombocytopenia and predisposition to acute myelogenous leukemia caused by constitutional microdeletions on chromosome 21q. *Blood*, **2008**, 112, 1042-7	2.2	64
23	Low-level mosaicism of trisomy 14: phenotypic and molecular characterization. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1395-405	2.5	30
22	Delineation of the proximal 3q microdeletion syndrome. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1729-35	2.5	15
21	Lymphedema of the lower extremity: is it genetic or nongenetic?. *Clinical Pediatrics*, **2007**, 46, 835-41	1.2	8
20	Preaxial polydactyly in neurofibromatosis 1. *Clinical Dysmorphology*, **2007**, 16, 193-194	0.9	5
19	Hyperhomocysteinemia and cobalamin disorders. *Molecular Genetics and Metabolism*, **2007**, 90, 113-21	3.7	12
18	Multiple ganglion cysts (Rcystic ganglionosisR): an unusual presentation in a child. *Scandinavian Journal of Rheumatology*, **2007**, 36, 145-8	1.9	14
17	A phase II/III clinical study of enzyme replacement therapy with idursulfase in mucopolysaccharidosis II (Hunter syndrome). *Genetics in Medicine*, **2006**, 8, 465-73	8.1	416
16	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**, 138, 56-60	2.5	17
15	Extensive primary cutaneous herpes simplex virus type 1 infection in an infant following acute rotavirus gastroenteritis. *European Journal of Pediatrics*, **2005**, 164, 175-6	4.1	
14	Autoantibodies against bactericidal/permeability-increasing protein (BPI-ANCA) in cystic fibrosis patients treated with azithromycin. *Clinical and Experimental Medicine*, **2005**, 5, 80-5	4.9	13
13	Splicing mutation in the fibrillin-1 gene associated with neonatal Marfan syndrome and severe pulmonary emphysema with tracheobronchomalacia. *Pediatric Pulmonology*, **2005**, 39, 374-8	3.5	15
12	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	5.8	18
11	Pulmonary manifestations and function tests in children genetically diagnosed with FMF. *Pediatric Pulmonology*, **2003**, 35, 452-5	3.5	7
10	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		13
9	Crouzon syndrome: association with absent pulmonary valve syndrome and severe tracheobronchomalacia. *Pediatric Pulmonology*, **2002**, 34, 478-81	3.5	10
8	The differential contribution of MEFV mutant alleles to the clinical profile of familial Mediterranean fever. *European Journal of Human Genetics*, **2002**, 10, 145-9	5.3	97

7	The musculoskeletal manifestations of familial Mediterranean fever in children genetically diagnosed with the disease. *Arthritis and Rheumatism*, **2001**, 44, 1416-9		60
6	Familial Mediterranean fever: prevalence, penetrance and genetic drift. *European Journal of Human Genetics*, **2001**, 9, 634-7	5.3	131
5	Familial Mediterranean fever: high gene frequency and heterogeneous disease among an Israeli-Arab population. *Journal of Rheumatology*, **2000**, 27, 1492-5	4.1	20
4	Familial Mediterranean fever: clinical and genetic characterization in a mixed pediatric population of Jewish and Arab patients. *Pediatrics*, **1999**, 103, e70	7.4	85
3	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. *Human Mutation*, **1999**, 14, 91	4.7	19
2	CSF levels of carnitine in children with meningitis, neurologic disorders, acute gastroenteritis, and seizure. *Neurology*, **1998**, 50, 1869-71	6.5	21
1	The differential contribution of MEFV mutant alleles to the clinical profile of familial Mediterranean fever		2