Orly Elpeleg

List of Publications by Year
in descending order

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190	11,522	23544	37183
papers	citations	58 h-index	96 g-index

196	196	196	16618
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutant mitochondrial thymidine kinase in mitochondrial DNA depletion myopathy. Nature Genetics, 2001, 29, 342-344.	9.4	551
2	The deoxyguanosine kinase gene is mutated in individuals with depleted hepatocerebral mitochondrial DNA. Nature Genetics, 2001, 29, 337-341.	9.4	521
3	Deleterious Mutation in the Mitochondrial Arginylâ€“Transfer RNA Synthetase Gene Is Associated with Pontocerebellar Hypoplasia. American Journal of Human Genetics, 2007, 81, 857-862.	2.6	306
4	Deficiency of the ADP-Forming Succinyl-CoA Synthase Activity Is Associated with Encephalomyopathy and Mitochondrial DNA Depletion. American Journal of Human Genetics, 2005, 76, 1081-1086.	2.6	284
5	Demonstration of a New Pathogenic Mutation in Human Complex I Deficiency: A 5-bp Duplication in the Nuclear Gene Encoding the 18-kD (AQDQ) Subunit. American Journal of Human Genetics, 1998, 62, 262-268.	2.6	268
6	A Fatal Mitochondrial Disease Is Associated with Defective NFU1 Function in the Maturation of a Subset of Mitochondrial Fe-S Proteins. American Journal of Human Genetics, 2011, 89, 656-667.	2.6	262
7	A Deleterious Mutation in DNAJC6 Encoding the Neuronal-Specific Clathrin-Uncoating Co-Chaperone Auxilin, Is Associated with Juvenile Parkinsonism. PLoS ONE, 2012, 7, e36458.	1.1	256
8	Acute Infantile Liver Failure Due to Mutations in the TRMU Gene. American Journal of Human Genetics, 2009, 85, 401-407.	2.6	205
9	Mutations in the Fatty Acid 2-Hydroxylase Gene Are Associated with Leukodystrophy with Spastic Paraparesis and Dystonia. American Journal of Human Genetics, 2008, 83, 643-648.	2.6	193
10	Mutations in LPIN1 Cause Recurrent Acute Myoglobinuria in Childhood. American Journal of Human Genetics, 2008, 83, 489-494.	2.6	189
11	Exome sequencing and disease-network analysis of a single family implicate a mutation in KIF1A in hereditary spastic paraparesis. Genome Research, 2011, 21, 658-664.	2.4	172
12	Distinct Clinical Phenotypes Associated with a Mutation in the Mitochondrial Translation Elongation Factor EFTs. American Journal of Human Genetics, 2006, 79, 869-877.	2.6	169
13	Type III 3-Methylglutaconic Aciduria (Optic Atrophy Plus Syndrome, or Costeff Optic Atrophy) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf of Human Genetics, 2001, 69, 1218-1224.	2.6	166
14	The H Syndrome Is Caused by Mutations in the Nucleoside Transporter hENT3. American Journal of Human Genetics, 2008, 83, 529-534.	2.6	166
15	Deficiency of caspase recruitment domain family, memberÂ11 (CARD11), causes profound combined immunodeficiency in human subjects. Journal of Allergy and Clinical Immunology, 2013, 131, 477-485.e1.	1.5	166
16	Mutations in the Mitochondrial Seryl-tRNA Synthetase Cause Hyperuricemia, Pulmonary Hypertension, Renal Failure in Infancy and Alkalosis, HUPRA Syndrome. American Journal of Human Genetics, 2011, 88, 193-200.	2.6	161
17	C6ORF66 Is an Assembly Factor of Mitochondrial Complex I. American Journal of Human Genetics, 2008, 82, 32-38.	2.6	155
18	CD59 deficiency is associated with chronic hemolysis and childhood relapsing immune-mediated polyneuropathy. Blood, 2013, 121, 129-135.	0.6	142

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19	Seemingly Neutral Polymorphic Variants May Confer Immunity to Splicing-Inactivating Mutations: A Synonymous SNP in Exon 5 of MCAD Protects from Deleterious Mutations in a Flanking Exonic Splicing Enhancer. American Journal of Human Genetics, 2007, 80, 416-432.	2.6	140
20	Hereditary sensory autonomic neuropathy caused by a mutation in dystonin. Annals of Neurology, 2012, 71, 569-572.	2.8	128
21	The H syndrome: A genodermatosis characterized by indurated, hyperpigmented, and hypertrichotic skin with systemic manifestations. Journal of the American Academy of Dermatology, 2008, 59, 79-85.	0.6	117
22	LPIN1 gene mutations: a major cause of severe rhabdomyolysis in early childhood. Human Mutation, 2010, 31, E1564-E1573.	1.1	112
23	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
24	CCDC65 Mutation Causes Primary Ciliary Dyskinesia with Normal Ultrastructure and Hyperkinetic Cilia. PLoS ONE, 2013, 8, e72299.	1.1	108
25	Cryptic proteolytic activity of dihydrolipoamide dehydrogenase. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 6158-6163.	3.3	107
26	FASTKD2 Nonsense Mutation in an Infantile Mitochondrial Encephalomyopathy Associated with Cytochrome C Oxidase Deficiency. American Journal of Human Genetics, 2008, 83, 415-423.	2.6	107
27	IL-2-inducible T-cell kinase deficiency: clinical presentation and therapeutic approach. Haematologica, 2011, 96, 472-476.	1.7	105
28	Mitochondrial complex I deficiency caused by a deleterious NDUFA11 mutation. Annals of Neurology, 2008, 63, 405-408.	2.8	103
29	AnSNX10mutation causes malignant osteopetrosis of infancy. Journal of Medical Genetics, 2012, 49, 221-226.	1.5	102
30	Mutations inEFL1, anSBDSpartner, are associated with infantile pancytopenia, exocrine pancreatic insufficiency and skeletal anomalies in aShwachman-Diamond like syndrome. Journal of Medical Genetics, 2017, 54, 558-566.	1.5	101
31	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	3.7	98
32	Autoimmune lymphoproliferative syndrome-like disease in patients with LRBA mutation. Clinical Immunology, 2015, 159, 84-92.	1.4	96
33	Infantile Cerebellar-Retinal Degeneration Associated with a Mutation in Mitochondrial Aconitase, ACO2. American Journal of Human Genetics, 2012, 90, 518-523.	2.6	93
34	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	2.6	93
35	Fatal infantile mitochondrial encephalomyopathy, hypertrophic cardiomyopathy and optic atrophy associated with a homozygousOPA1mutation. Journal of Medical Genetics, 2016, 53, 127-131.	1.5	91
36	mtDNA depletion myopathy: elucidation of the tissue specificity in the mitochondrial thymidine kinase (TK2) deficiency. Molecular Genetics and Metabolism, 2003, 79, 1-5.	0.5	89

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37	Joubert Syndrome 2 (JBTS2) in Ashkenazi Jews Is Associated with a TMEM216 Mutation. American Journal of Human Genetics, 2010, 86, 93-97.	2.6	89
38	Early prenatal ventriculomegaly due to an AIFM1 mutation identified by linkage analysis and whole exome sequencing. Molecular Genetics and Metabolism, 2011, 104, 517-520.	0.5	89
39	Extending the Clinical Phenotype of Adenosine Deaminase 2 Deficiency. Journal of Pediatrics, 2016, 177, 316-320.	0.9	87
40	LRRC6 Mutation Causes Primary Ciliary Dyskinesia with Dynein Arm Defects. PLoS ONE, 2013, 8, e59436.	1.1	87
41	Mutations in SLC33A1 Cause a Lethal Autosomal-Recessive Disorder with Congenital Cataracts, Hearing Loss, and Low Serum Copper and Ceruloplasmin. American Journal of Human Genetics, 2012, 90, 61-68.	2.6	85
42	TRMT10A dysfunction is associated with abnormalities in glucose homeostasis, short stature and microcephaly. Journal of Medical Genetics, 2014, 51, 581-586.	1.5	83
43	Mitochondrial complex IV deficiency, caused by mutated COX6B1, is associated with encephalomyopathy, hydrocephalus and cardiomyopathy. European Journal of Human Genetics, 2015, 23, 159-164.	1.4	82
44	EPT1 (selenoprotein I) is critical for the neural development and maintenance of plasmalogen in humans. Journal of Lipid Research, 2018, 59, 1015-1026.	2.0	79
45	Exocrine Pancreatic Insufficiency, Dyserythropoeitic Anemia, and Calvarial Hyperostosis Are Caused by a Mutation in the COX4I2 Gene. American Journal of Human Genetics, 2009, 84, 412-417.	2.6	78
46	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. American Journal of Human Genetics, 2016, 99, 728-734.	2.6	75
47	Biochemical Assays for Mitochondrial Activity: Assays of TCA Cycle Enzymes and PDHc. Methods in Cell Biology, 2007, 80, 199-222.	0.5	74
48	SLC25A19 mutation as a cause of neuropathy and bilateral striatal necrosis. Annals of Neurology, 2009, 66, 419-424.	2.8	74
49	The Thr224Asn mutation in the VPS45 gene is associated with the congenital neutropenia and primary myelofibrosis of infancy. Blood, 2013, 121, 5078-5087.	0.6	70
50	A Recessive Contiguous Gene Deletion of Chromosome 2p16 Associated with Cystinuria and a Mitochondrial Disease. American Journal of Human Genetics, 2001, 69, 869-875.	2.6	69
51	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	1.1	69
52	Clinical and biochemical characterization of four patients with mutations in ECHS1. Orphanet Journal of Rare Diseases, 2015, 10, 79.	1.2	68
53	Mutations inSLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. Journal of Medical Genetics, 2015, 52, 541-547.	1.5	68
54	Two novel CCDC88C mutations confirm the role of DAPLE in autosomal recessive congenital hydrocephalus. Journal of Medical Genetics, 2012, 49, 708-712.	1.5	67

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55	tRNA N6-adenosine threonylcarbamoyltransferase defect due to KAE1/TCS3 (OSGEP) mutation manifest by neurodegeneration and renal tubulopathy. European Journal of Human Genetics, 2017, 25, 545-551.	1.4	67
56	Early-onset Evans syndrome, immunodeficiency, and premature immunosenescence associated with tripeptidyl-peptidase II deficiency. Blood, 2015, 125, 753-761.	0.6	66
57	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	2.6	66
58	Deleterious mutation in FDX1L gene is associated with a novel mitochondrial muscle myopathy. European Journal of Human Genetics, 2014, 22, 902-906.	1.4	65
59	The 3â€² addition of CCA to mitochondrial tRNASer(AGY) is specifically impaired in patients with mutations in the tRNA nucleotidyl transferase TRNT1. Human Molecular Genetics, 2015, 24, 2841-2847.	1.4	65
60	Short-chain acyl-CoA dehydrogenase gene mutation (c.319C>T) presents with clinical heterogeneity and is candidate founder mutation in individuals of Ashkenazi Jewish origin. Molecular Genetics and Metabolism, 2008, 93, 179-189.	0.5	61
61	TMEM70 mutations are a common cause of nuclear encoded ATP synthase assembly defect: further delineation of a new syndrome. Journal of Medical Genetics, 2011, 48, 177-182.	1.5	61
62	Agenesis of corpus callosum and optic nerve hypoplasia due to mutations inSLC25A1encoding the mitochondrial citrate transporter. Journal of Medical Genetics, 2013, 50, 240-245.	1.5	60
63	Early infantile epileptic encephalopathy associated with a high voltage gated calcium channelopathy. Journal of Medical Genetics, 2013, 50, 118-123.	1.5	60
64	Infantile Cerebral and Cerebellar Atrophy Is Associated with a Mutation in the MED17 Subunit of the Transcription Preinitiation Mediator Complex. American Journal of Human Genetics, 2010, 87, 667-670.	2.6	58
65	West syndrome caused by ST3Galâ€‖ deficiency. Epilepsia, 2013, 54, e24-7.	2.6	58
66	Early onset combined immunodeficiency and autoimmunity in patients with loss-of-function mutation in LAT. Journal of Experimental Medicine, 2016, 213, 1185-1199.	4.2	57
67	Infantile citrullinemia caused by citrin deficiency with increased dibasic amino acids. Molecular Genetics and Metabolism, 2002, 77, 202-208.	0.5	56
68	Evaluation of enzymatic assays and compounds affecting ATP production in mitochondrial respiratory chain complex I deficiency. Analytical Biochemistry, 2004, 335, 66-72.	1.1	56
69	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	2.6	56
70	Mutations in SLC35A3 cause autism spectrum disorder, epilepsy and arthrogryposis. Journal of Medical Genetics, 2013, 50, 733-739.	1.5	55
71	Depletion of the other genome-mitochondrial DNA depletion syndromes in humans. Journal of Molecular Medicine, 2002, 80, 389-396.	1.7	54
72	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	2.6	54

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73	Combined OXPHOS complex I and IV defect, due to mutated complex I assembly factor C20ORF7. Journal of Inherited Metabolic Disease, 2012, 35, 125-131.	1.7	52
74	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. American Journal of Human Genetics, 2016, 98, 782-788.	2.6	50
75	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
76	A human laterality disorder associated with recessiveCCDC11mutation. Journal of Medical Genetics, 2012, 49, 386-390.	1.5	49
77	Deficiency of the alkaline ceramidase ACER3 manifests in early childhood by progressive leukodystrophy. Journal of Medical Genetics, 2016, 53, 389-396.	1.5	49
78	Homozygous loss-of-function mutations in MNS1 cause laterality defects and likely male infertility. PLoS Genetics, 2018, 14, e1007602.	1.5	49
79	Delineation of C12orf65-related phenotypes: a genotypeâ€“phenotype relationship. European Journal of Human Genetics, 2014, 22, 1019-1025.	1.4	48
80	The unique neuroradiology of complex I deficiency due to NDUFA12L defect. Molecular Genetics and Metabolism, 2008, 94, 78-82.	0.5	46
81	A human laterality disorder caused by a homozygous deleterious mutation inMMP21. Journal of Medical Genetics, 2015, 52, 840-847.	1.5	46
82	Loss of ADAMTS19 causes progressive non-syndromic heart valve disease. Nature Genetics, 2020, 52, 40-47.	9.4	46
83	TECPR2 mutations cause a new subtype of familial dysautonomia like hereditary sensory autonomic neuropathy with intellectual disability. European Journal of Paediatric Neurology, 2016, 20, 69-79.	0.7	45
84	Homozygous mutation in MFSD2A, encoding a lysolipid transporter for docosahexanoic acid, is associated with microcephaly and hypomyelination. Neurogenetics, 2018, 19, 227-235.	0.7	45
85	A defect in the retromer accessory protein, SNX27, manifests by infantile myoclonic epilepsy and neurodegeneration. Neurogenetics, 2015, 16, 215-221.	0.7	44
86	Mitochondrial hepato-encephalopathy due to deficiency of QIL1/MIC13 (C19orf70), a MICOS complex subunit. European Journal of Human Genetics, 2016, 24, 1778-1782.	1.4	44
87	Mutations in the phosphatidylinositol glycan C (PIGC) gene are associated with epilepsy and intellectual disability. Journal of Medical Genetics, 2017, 54, 196-201.	1.5	44
88	A human laterality disorder associated with a homozygous WDR16 deletion. European Journal of Human Genetics, 2015, 23, 1262-1265.	1.4	43
89	Compound heterozygosity for severe and hypomorphicNDUFS2mutations cause non-syndromic LHON-like optic neuropathy. Journal of Medical Genetics, 2017, 54, 346-356.	1.5	43
90	N-acetylglutamate synthase deficiency and the treatment of hyperammonemic encephalopathy. Annals of Neurology, 2002, 52, 845-849.	2.8	42

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91	Mitochondrial deoxyribonucleoside triphosphate pools in thymidine kinase 2 deficiency. Biochemical and Biophysical Research Communications, 2003, 310, 963-966.	1.0	41
92	Mutated NDUFS6 is the cause of fatal neonatal lactic acidemia in Caucasus Jews. European Journal of Human Genetics, 2009, 17, 1200-1203.	1.4	41
93	Extending the clinical and immunological phenotype of human interleukin-21 receptor deficiency. Haematologica, 2015, 100, e72-e76.	1.7	41
94	Hypomyelination and developmental delay associated withVPS11mutation in Ashkenazi-Jewish patients. Journal of Medical Genetics, 2015, 52, 749-753.	1.5	41
95	Therapy with eculizumab for patients with CD59 p.Cys89Tyr mutation. Annals of Neurology, 2016, 80, 708-717.	2.8	41
96	Heterozygous De Novo UBTF Gain-of-Function Variant Is Associated with Neurodegeneration in Childhood. American Journal of Human Genetics, 2017, 101, 267-273.	2.6	41
97	Organic solute transporterâ€Î² (SLC51B) deficiency in two brothers with congenital diarrhea and features of cholestasis. Hepatology, 2018, 68, 590-598.	3.6	41
98	Combined loss of LAP1B and LAP1C results in an early onset multisystemic nuclear envelopathy. Nature Communications, 2019, 10, 605.	5.8	40
99	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
100	Nemaline body myopathy caused by a novel mutation in troponin T1 (TNNT1). Muscle and Nerve, 2016, 53, 564-569.	1.0	39
101	TAT-mediated Delivery of LAD Restores Pyruvate Dehydrogenase Complex Activity in the Mitochondria of Patients with LAD Deficiency. Molecular Therapy, 2008, 16, 691-697.	3.7	38
102	Mutation in the COX4I1 gene is associated with short stature, poor weight gain and increased chromosomal breaks, simulating Fanconi anemia. European Journal of Human Genetics, 2017, 25, 1142-1146.	1.4	38
103	Truncating Mutation in the Nitric Oxide Synthase 1 Gene Is Associated With Infantile Achalasia. Gastroenterology, 2015, 148, 533-536.e4.	0.6	37
104	Mutations in TRAPPC12 Manifest in Progressive Childhood Encephalopathy and Golgi Dysfunction. American Journal of Human Genetics, 2017, 101, 291-299.	2.6	37
105	Deleterious mutation in GPR88 is associated with chorea, speech delay, and learning disabilities. Neurology: Genetics, 2016, 2, e64.	0.9	36
106	Congenital valvular defects associated with deleterious mutations in thePLD1gene. Journal of Medical Genetics, 2017, 54, 278-286.	1.5	36
107	Two separate functions of NME3 critical for cell survival underlie a neurodegenerative disorder. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 566-574.	3.3	36
108	CFAP45 deficiency causes situs abnormalities and asthenospermia by disrupting an axonemal adenine nucleotide homeostasis module. Nature Communications, 2020, 11, 5520.	5.8	36

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109	Altered RNA metabolism due to a homozygousRBM7mutation in a patient with spinal motor neuropathy. Human Molecular Genetics, 2016, 25, ddw149.	1.4	35
110	Molecular and biochemical characterization of a unique mutation in CCS, the human copper chaperone to superoxide dismutase. Human Mutation, 2012, 33, 1207-1215.	1.1	34
111	Stem cell transplantation for osteopetrosis in patients beyond the age of 5 years. Blood Advances, 2019, 3, 862-868.	2.5	34
112	Isolated truncus arteriosus associated with a mutation in the plexinâ€Ð1 gene. American Journal of Medical Genetics, Part A, 2013, 161, 3115-3120.	0.7	33
113	Exome sequencing identifies a new mutation in SERAC1 in a patient with 3-methylglutaconic aciduria. Molecular Genetics and Metabolism, 2013, 110, 73-77.	0.5	33
114	Cytokine secretion and NK cell activity in human ADAM17 deficiency. Oncotarget, 2015, 6, 44151-44160.	0.8	33
115	Homozygous mutation in the APOA1BP is associated with a lethal infantile leukoencephalopathy. Neurogenetics, 2016, 17, 187-190.	0.7	32
116	De novo GRIN1 mutations: An emerging cause of severe early infantile encephalopathy. European Journal of Medical Genetics, 2017, 60, 317-320.	0.7	32
117	Deficiency of HTRA2/Omi is associated with infantile neurodegeneration and 3-methylglutaconic aciduria. Journal of Medical Genetics, 2016, 53, 690-696.	1.5	30
118	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30
119	Tocilizumab Promotes Regulatory T-cell Alleviation in STAT3 Gain-of-functionâ^'associated Multi-organ Autoimmune Syndrome. Clinical Therapeutics, 2017, 39, 444-449.	1.1	29
120	Pathogenic Variants in NUP214 Cause â€œPluggedâ€•Nuclear Pore Channels and Acute Febrile Encephalopathy. American Journal of Human Genetics, 2019, 105, 48-64.	2.6	29
121	Conotruncal malformations and absent thymus due to a deleterious NKX2-6 mutation. Journal of Medical Genetics, 2014, 51, 268-270.	1.5	28
122	Inherited Mitochondrial DNA Depletion. Pediatric Research, 2003, 54, 153-159.	1.1	27
123	Infantile Neurodegenerative Disorder Associated with Mutations inTBCD, an Essential Gene in the Tubulin Heterodimer Assembly Pathway. Human Molecular Genetics, 2016, 25, ddw292.	1.4	25
124	Biallelic variants in AGTPBP1, involved in tubulin deglutamylation, are associated with cerebellar degeneration and motor neuropathy. European Journal of Human Genetics, 2019, 27, 1419-1426.	1.4	25
125	West syndrome, microcephaly, grey matter heterotopia and hypoplasia of corpus callosum due to a novel ARFGEF2 mutation. Journal of Medical Genetics, 2013, 50, 772-775.	1.5	24
126	Homozygous mutation, p.Pro304His, in IDH3A, encoding isocitrate dehydrogenase subunit is associated with severe encephalopathy in infancy. Neurogenetics, 2017, 18, 57-61.	0.7	23

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127	Clinical and experimental evidence suggest a link between KIF7 and C5orf42-related ciliopathies through Sonic Hedgehog signaling. European Journal of Human Genetics, 2018, 26, 197-209.	1.4	23
128	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	3.6	23
129	Enteroviral Infection in a Patient with BLNK Adaptor Protein Deficiency. Journal of Clinical Immunology, 2015, 35, 356-360.	2.0	22
130	Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. American Journal of Human Genetics, 2017, 100, 666-675.	2.6	22
131	Novel Mutations in the PEX2 Gene of Four Unrelated Patients with a Peroxisome Biogenesis Disorder. Pediatric Research, 2004, 55, 431-436.	1.1	21
132	Leukoencephalopathy and early death associated with an Ashkenazi-Jewish founder mutation in the Hikeshi gene. Journal of Medical Genetics, 2016, 53, 132-137.	1.5	21
133	A homozygous deleterious CDK10 mutation in a patient with agenesis of corpus callosum, retinopathy, and deafness. American Journal of Medical Genetics, Part A, 2018, 176, 92-98.	0.7	21
134	Respiratory manifestations in LPS-responsive beige-like anchor (LRBA) protein-deficient patients. European Journal of Pediatrics, 2018, 177, 1163-1172.	1.3	20
135	Compound heterozygous variants in PGAP1 causing severe psychomotor retardation, brain atrophy, recurrent apneas and delayed myelination: a case report and literature review. BMC Neurology, 2016, 16, 74.	0.8	19
136	Further delineation of the clinical spectrum of de novo TRIM8 truncating mutations. American Journal of Medical Genetics, Part A, 2018, 176, 2470-2478.	0.7	19
137	A Novel Familial Mutation in the PCSK1 Gene That Alters the Oxyanion Hole Residue of Proprotein Convertase 1/3 and Impairs Its Enzymatic Activity. PLoS ONE, 2014, 9, e108878.	1.1	19
138	Devastating recurrent brain ischemic infarctions and retinal disease in pediatric patients with CD59 deficiency. European Journal of Paediatric Neurology, 2015, 19, 688-693.	0.7	18
139	Homozygous null variant in CRADD, encoding an adaptor protein that mediates apoptosis, is associated with lissencephaly. American Journal of Medical Genetics, Part A, 2017, 173, 2539-2544.	0.7	18
140	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. Genetics in Medicine, 2020, 22, 1598-1605.	1.1	18
141	Cardiac-Targeted Transgenic Mutant Mitochondrial Enzymes: mtDNA Defects, Antiretroviral Toxicity and Cardiomyopathy. Cardiovascular Toxicology, 2008, 8, 57-69.	1.1	17
142	A novel mutation in TTC19 associated with isolated complex III deficiency, cerebellar hypoplasia, and bilateral basal ganglia lesions. Frontiers in Genetics, 2014, 5, 397.	1.1	17
143	PARP10 deficiency manifests by severe developmental delay and DNA repair defect. Neurogenetics, 2016, 17, 227-232.	0.7	17
144	A mutation in the THG1L gene in a family with cerebellar ataxia and developmental delay. Neurogenetics, 2016, 17, 219-225.	0.7	17

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145	A mutation in POLR3E impairs antiviral immune response and RNA polymerase III. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 22113-22121.	3.3	17
146	Intractable epilepsy of infancy due to homozygous mutation in the EFHC1 gene. Epilepsia, 2012, 53, 1436-1440.	2.6	16
147	T+ NK+ IL-2 Receptor Î³ Chain Mutation: a Challenging Diagnosis of Atypical Severe Combined Immunodeficiency. Journal of Clinical Immunology, 2018, 38, 527-536.	2.0	16
148	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. Journal of Clinical Investigation, 2021, 131, .	3.9	16
149	Novel Homozygous Missense Mutation in SPG20 Gene Results in Troyer Syndrome Associated with Mitochondrial Cytochrome c Oxidase Deficiency. JIMD Reports, 2016, 33, 55-60.	0.7	15
150	A homozygous TTN gene variant associated with lethal congenital contracture syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1001-1005.	0.7	15
151	A patient-specific induced pluripotent stem cell model for West syndrome caused by ST3GAL3 deficiency. European Journal of Human Genetics, 2018, 26, 1773-1783.	1.4	15
152	MARS variant associated with both recessive interstitial lung and liver disease and dominant Charcot-Marie-Tooth disease. European Journal of Medical Genetics, 2018, 61, 616-620.	0.7	14
153	CD59 Deficiency Is Associated with Chronic Hemolysis and Childhood Chronic Immune Mediated Neuropathy. Blood, 2012, 120, 3187-3187.	0.6	14
154	Clinical Characteristics and Muscle Pathology in Myopathic Mitochondrial DNA Depletion. Journal of Child Neurology, 2002, 17, 499-504.	0.7	13
155	<scp>VPS</scp> 45â€associated primary infantile myelofibrosis â€“ Successful treatment with hematopoietic stem cell transplantation. Pediatric Transplantation, 2013, 17, 820-825.	0.5	13
156	Deep intronic mis-splicing mutation in JAK3 gene underlies T â^' B + NK â^' severe combined immunodeficiency phenotype. Clinical Immunology, 2016, 163, 91-95.	1.4	13
157	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	1.5	11
158	Bacillus Calmetteâ€“Guerin (BCG) Vaccine-associated Complications in Immunodeficient Patients Following Stem Cell Transplantation. Journal of Clinical Immunology, 2021, 41, 147-162.	2.0	11
159	Type IV 3-Methylglutaconic (3-MGC) aciduria: A new case presenting with hepatic dysfunction. Pediatric Neurology, 1997, 17, 353-355.	1.0	10
160	Hindbrain malformation and myoclonic seizures associated with a deleterious mutation in the INPP4A gene. Neurogenetics, 2015, 16, 23-26.	0.7	10
161	Heterozygous RNF13 Gain-of-Function Variants Are Associated with Congenital Microcephaly, Epileptic Encephalopathy, Blindness, and Failure to Thrive. American Journal of Human Genetics, 2019, 104, 179-185.	2.6	10
162	A human case of GIMAP6 deficiency: a novel primary immune deficiency. European Journal of Human Genetics, 2021, 29, 657-662.	1.4	10

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