Haluk A Topaloglu

List of Publications by Year
in descending order

153	13,389	26626	23530
papers	citations	56 h-index	111 g-index

162	162	162	13018
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2017, 377, 1723-1732.	27.0	1,533
2	Muscular Dystrophy and Neuronal Migration Disorder Caused by Mutations in a Glycosyltransferase, POMGnT1. Developmental Cell, 2001, 1, 717-724.	7.0	675
3	Mutations in the laminin Î±2â€“chain gene (LAMA2) cause merosinâ€“deficient congenital muscular dystrophy. Nature Genetics, 1995, 11, 216-218.	21.4	603
4	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders, 2018, 28, 103-115.	0.6	584
5	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. Neuromuscular Disorders, 2019, 29, 842-856.	0.6	401
6	Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. Brain, 2007, 130, 2725-2735.	7.6	385
7	The gene encoding gigaxonin, a new member of the cytoskeletal BTB/kelch repeat family, is mutated in giant axonal neuropathy. Nature Genetics, 2000, 26, 370-374.	21.4	372
8	Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. Lancet, The, 2017, 390, 1489-1498.	13.7	365
9	Mutations in SEPN1 cause congenital muscular dystrophy with spinal rigidity and restrictive respiratory syndrome. Nature Genetics, 2001, 29, 17-18.	21.4	326
10	Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies. JAMA - Journal of the American Medical Association, 2014, 312, 68.	7.4	304
11	The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene. Brain, 2007, 130, 2037-2044.	7.6	298
12	Safety and efficacy of drisapersen for the treatment of Duchenne muscular dystrophy (DEMAND II): an exploratory, randomised, placebo-controlled phase 2 study. Lancet Neurology, The, 2014, 13, 987-996.	10.2	279
13	Perlecan, the major proteoglycan of basement membranes, is altered in patients with Schwartz-Jampel syndrome (chondrodystrophic myotonia). Nature Genetics, 2000, 26, 480-483.	21.4	274
14	Deciphering the Glycosylome of Dystroglycanopathies Using Haploid Screens for Lassa Virus Entry. Science, 2013, 340, 479-483.	12.6	262
15	Spinal Muscular Atrophy Associated with Progressive Myoclonic Epilepsy Is Caused by Mutations in ASAH1. American Journal of Human Genetics, 2012, 91, 5-14.	6.2	250
16	A Dystroglycan Mutation Associated with Limb-Girdle Muscular Dystrophy. New England Journal of Medicine, 2011, 364, 939-946.	27.0	246
17	Mutations in SIL1 cause Marinesco-SjÃ¶gren syndrome, a cerebellar ataxia with cataract and myopathy. Nature Genetics, 2005, 37, 1312-1314.	21.4	232
18	Localization of merosin-negative congenital muscular dystrophy to chromosome 6q2 by homozygosity mapping. Human Molecular Genetics, 1994, 3, 1657-1661.	2.9	214

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19	Mutations in FAM134B, encoding a newly identified Golgi protein, cause severe sensory and autonomic neuropathy. Nature Genetics, 2009, 41, 1179-1181.	21.4	205
20	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	6.2	186
21	Mutations in a Gene Encoding a Novel SH3/TPR Domain Protein Cause Autosomal Recessive Charcot-Marie-Tooth Type 4C Neuropathy. American Journal of Human Genetics, 2003, 73, 1106-1119.	6.2	185
22	European Academy of Neurology/Peripheral Nerve Society guideline on diagnosis and treatment of chronic inflammatory demyelinating polyradiculoneuropathy: Report of a joint Task Forceâ€"Second revision. Journal of the Peripheral Nervous System, 2021, 26, 242-268.	3.1	176
23	Mutations in COL6A3 Cause Severe and Mild Phenotypes of Ullrich Congenital Muscular Dystrophy. American Journal of Human Genetics, 2002, 70, 1446-1458.	6.2	165
24	European Academy of Neurology/Peripheral Nerve Society guideline on diagnosis and treatment of chronic inflammatory demyelinating polyradiculoneuropathy: Report of a joint Task Forceâ€"Second revision. European Journal of Neurology, 2021, 28, 3556-3583.	3.3	153
25	Recessive TTN truncating mutations define novel forms of core myopathy with heart disease. Human Molecular Genetics, 2014, 23, 980-991.	2.9	149
26	Mitochondrial serine protease HTRA2 p.G399S in a kindred with essential tremor and Parkinson disease. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 18285-18290.	7.1	147
27	Worldwide distribution and broader clinical spectrum of muscle-eye-brain disease. Human Molecular Genetics, 2003, 12, 527-534.	2.9	133
28	Assignment of the Muscle-Eye-Brain Disease Gene to 1p32-p34 by Linkage Analysis and Homozygosity Mapping. American Journal of Human Genetics, 1999, 64, 126-135.	6.2	128
29	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	6.2	118
30	Deficiency of Î±-Dystroglycan in Muscleâ€"Eyeâ€"Brain Disease. Biochemical and Biophysical Research Communications, 2002, 291, 1283-1286.	2.1	115
31	A Congenital Muscular Dystrophy with Mitochondrial Structural Abnormalities Caused by Defective De Novo Phosphatidylcholine Biosynthesis. American Journal of Human Genetics, 2011, 88, 845-851.	6.2	115
32	Early onset collagen VI myopathies: Genetic and clinical correlations. Annals of Neurology, 2010, 68, 511-520.	5.3	112
33	Genetic spectrum of hereditary neuropathies with onset in the first year of life. Brain, 2011, 134, 2664-2676.	7.6	112
34	Missense mutation in the ATPase, aminophospholipid transporter protein ATP8A2 is associated with cerebellar atrophy and quadrupedal locomotion. European Journal of Human Genetics, 2013, 21, 281-285.	2.8	110
35	A Comparative Study of Î±â€Dystroglycan Glycosylation in Dystroglycanopathies Suggests that the Hypoglycosylation of Î±â€Dystroglycan Does Not Consistently Correlate with Clinical Severity. Brain Pathology, 2009, 19, 596-611.	4.1	107
36	Mutation in Exon 1f of PLEC, Leading to Disruption of Plectin Isoform 1f, Causes Autosomal-Recessive Limb-Girdle Muscular Dystrophy. American Journal of Human Genetics, 2010, 87, 834-841.	6.2	104

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37	Loss-of-function mutations in HINT1 cause axonal neuropathy with neuromyotonia. Nature Genetics, 2012, 44, 1080-1083.	21.4	102
38	Spectrum ofHSPG2(Perlecan) mutations in patients with Schwartz-Jampel syndrome. Human Mutation, 2006, 27, 1082-1091.	2.5	98
39	Spectrum of Brain Changes in Patients With Congenital Muscular Dystrophy and FKRP Gene Mutations. Archives of Neurology, 2006, 63, 251.	4.5	97
40	Identification of a New Locus for a Peculiar Form of Congenital Muscular Dystrophy with Early Rigidity of the Spine, on Chromosome 1p35-36. American Journal of Human Genetics, 1998, 62, 1439-1445.	6.2	95
41	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. Neuromuscular Disorders, 2021, 31, 574-582.	0.6	94
42	Assignment of a Form of Congenital Muscular Dystrophy with Secondary Merosin Deficiency to Chromosome 1q42. American Journal of Human Genetics, 2000, 66, 428-435.	6.2	90
43	Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. Journal of Neurology, 2014, 261, 152-163.	3.6	76
44	Dominant and recessive COL6A1 mutations in Ullrich scleroatonic muscular dystrophy. Annals of Neurology, 2005, 58, 400-410.	5.3	72
45	Mutation in TOR1AIP1 encoding LAP1B in a form of muscular dystrophy: A novel gene related to nuclear envelopathies. Neuromuscular Disorders, 2014, 24, 624-633.	0.6	71
46	Different types of upper extremity exercise training in Duchenne muscular dystrophy: Effects on functional performance, strength, endurance, and ambulation. Muscle and Nerve, 2015, 51, 697-705.	2.2	71
47	Late onset muscular dystrophy with cerebral white matter changes due to partial merosin deficiency. Neuromuscular Disorders, 1997, 7, 85-89.	0.6	69
48	Natural History of Congenital Generalized Lipodystrophy: A Nationwide Study From Turkey. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2759-2767.	3.6	67
49	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. Brain, 2011, 134, 183-195.	7.6	66
50	SIL1 mutations and clinical spectrum in patients with Marinesco-SjÃ¶gren syndrome. Brain, 2013, 136, 3634-3644.	7.6	65
51	MAN1B1 Deficiency: An Unexpected CDG-II. PLoS Genetics, 2013, 9, e1003989.	3.5	63
52	Homozygosity mapping and targeted genomic sequencing reveal the gene responsible for cerebellar hypoplasia and quadrupedal locomotion in a consanguineous kindred. Genome Research, 2011, 21, 1995-2003.	5.5	62
53	Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations. European Journal of Paediatric Neurology, 2010, 14, 326-333.	1.6	61
54	Acute Disseminated Encephalomyelitis in Childhood: Report of 10 Cases. Journal of Child Neurology, 1999, 14, 198-201.	1.4	60

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55	Novel POMGnT1 mutations define broader phenotypic spectrum of muscleâ€“eyeâ€“brain disease. Neurogenetics, 2007, 8, 279-288.	1.4	60
56	Recessive PIEZO2 stop mutation causes distal arthrogryposis with distal muscle weakness, scoliosis and proprioception defects. Journal of Human Genetics, 2017, 62, 497-501.	2.3	60
57	Private SACS mutations in autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) families from Turkey. Neurogenetics, 2004, 5, 165-170.	1.4	57
58	Enzymatic diagnostic test for Muscle-Eye-Brain type congenital muscular dystrophy using commercially available reagents. Clinical Biochemistry, 2003, 36, 339-344.	1.9	54
59	A novel form of recessive limb girdle muscular dystrophy with mental retardation and abnormal expression of Î±-dystroglycan. Neuromuscular Disorders, 2003, 13, 771-778.	0.6	49
60	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. American Journal of Human Genetics, 2016, 99, 1086-1105.	6.2	45
61	Targeted sequencing with expanded gene profile enables high diagnostic yield in non-5q-spinal muscular atrophies. Human Mutation, 2018, 39, 1284-1298.	2.5	42
62	Early-onset chronic axonal neuropathy, strokes, and hemolysis. Neurology, 2015, 84, 1220-1224.	1.1	40
63	Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation. Brain, 2013, 136, e228-e228.	7.6	38
64	Gender Prevalence in Childhood Multiple Sclerosis and Myasthenia Gravis. Journal of Child Neurology, 2002, 17, 390-392.	1.4	36
65	A novel homozygous missense mutation in the myotubularin-related protein 2 gene associated with recessive Charcotâ€“Marieâ€“Tooth disease with irregularly folded myelin sheaths. Neuromuscular Disorders, 2002, 12, 869-873.	0.6	36
66	Identification of a founder mutation in TPM3 in nemaline myopathy patients of Turkish origin. European Journal of Human Genetics, 2008, 16, 1055-1061.	2.8	36
67	Prenatal diagnosis in laminin Î±2 chain (merosin)-deficient congenital muscular dystrophy: A collective experience of five international centers. Neuromuscular Disorders, 2005, 15, 588-594.	0.6	35
68	Novel mutations in genes causing hereditary spastic paraplegia and Charcot-Marie-Tooth neuropathy identified by an optimized protocol for homozygosity mapping based on whole-exome sequencing. Genetics in Medicine, 2016, 18, 600-607.	2.4	35
69	The genomic and clinical landscape of fetal akinesia. Genetics in Medicine, 2020, 22, 511-523.	2.4	35
70	Calpain-3 mutations in Turkey. European Journal of Pediatrics, 2006, 165, 293-298.	2.7	34
71	Protein O-mannosyltransferase activities in lymphoblasts from patients with Î±-dystroglycanopathies. Neuromuscular Disorders, 2008, 18, 45-51.	0.6	33
72	Genetic heterogeneity of congenital muscular dystrophy with rigid spine syndrome. Neuromuscular Disorders, 1999, 9, 376-382.	0.6	32

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73	Glycosylation defects in muscular dystrophies. Current Opinion in Neurology, 2004, 17, 521-527.	3.6	31
74	Arthrogryposis and fetal hypomobility syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1311-1319.	1.8	29
75	Genetic Landscape of Congenital Myasthenic Syndromes From Turkey: Novel Mutations and Clinical Insights. Journal of Child Neurology, 2017, 32, 759-765.	1.4	29
76	Recessive Schwartz-Jampel syndrome (SJS): confirmation of linkage to chromosome 1p, evidence of genetic homogeneity and reduction of the SJS locus to a 3-cM interval. Human Genetics, 1996, 98, 380-385.	3.8	28
77	Neurologic features as initial presentations of childhood malignancies. Pediatric Neurology, 1994, 10, 40-43.	2.1	27
78	Cerebral infarct associated with factor V Leiden mutation in a boy with hemophilia A. American Journal of Hematology, 1997, 56, 189-190.	4.1	27
79	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27
80	A Novel Missense Variant in the AGRN Gene; Congenital Myasthenic Syndrome Presenting With Head Drop. Journal of Clinical Neuromuscular Disease, 2017, 18, 147-151.	0.7	26
81	Two siblings with nemaline myopathy presenting with rigid spine syndrome. Neuromuscular Disorders, 1994, 4, 263-267.	0.6	24
82	Giant axonal neuropathy locus refinement to a < 590 kb critical interval. European Journal of Human Genetics, 2000, 8, 527-534.	2.8	23
83	Spectrum of clinical manifestations in two young Turkish patients with congenital generalized lipodystrophy type 4. European Journal of Medical Genetics, 2016, 59, 320-324.	1.3	23
84	Merosin-deficient congenital muscular dystrophy with mental retardation and cerebellar cysts unlinked to the LAMA2, FCMD and MEB loci. Neuromuscular Disorders, 2000, 10, 548-552.	0.6	22
85	Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency. Pediatric Neurology, 2019, 99, 69-75.	2.1	22
86	Expression of HLA Class I Antigens in Skeletal Muscle Is a Diagnostic Marker in Juvenile Dermatomyositis. Journal of Child Neurology, 1997, 12, 60-63.	1.4	21
87	The role of immunocytochemistry and linkage analysis in the prenatal diagnosis of merosin-deficient congenital muscular dystrophy. Human Genetics, 1997, 99, 535-540.	3.8	20
88	Clinical spectra of neuromuscular manifestations in patients with lipodystrophy: A multicenter study. Neuromuscular Disorders, 2017, 27, 923-930.	0.6	20
89	Effect of muscle weakness distribution on balance in neuromuscular disease. Pediatrics International, 2015, 57, 92-97.	0.5	19
90	Deletion analysis in Turkish patients with spinal muscular atrophy. Brain and Development, 1999, 21, 86-89.	1.1	18

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91	Electroencephalographic findings in anti-N-methyl-d-aspartate receptor encephalitis in children: A series of 12 patients. Epilepsy and Behavior, 2018, 78, 118-123.	1.7	18
92	Clinical long-time course, novel mutations and genotype-phenotype correlation in a cohort of 27 families with POMT1-related disorders. Orphanet Journal of Rare Diseases, 2019, 14, 179.	2.7	18
93	Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling. Neuromuscular Disorders, 2019, 29, 601-613.	0.6	18
94	Selenoprotein Nâ€related myopathy: a retrospective natural history study to guide clinical trials. Annals of Clinical and Translational Neurology, 2020, 7, 2288-2296.	3.7	18
95	Monomelic amyotrophy in siblings. Pediatric Neurology, 1991, 7, 220-222.	2.1	17
96	Results of Botilinum Toxin. Journal of Craniofacial Surgery, 2006, 17, 656-660.	0.7	17
97	Transcript levels of plastin 3 and neuritin 1 modifier genes in spinal muscular atrophy siblings. Pediatrics International, 2017, 59, 53-56.	0.5	17
98	Effects of Arm Cycling Exercise in Spinal Muscular Atrophy Type II Patients: A Pilot Study. Journal of Child Neurology, 2018, 33, 209-215.	1.4	17
99	Neonatal-Onset Recurrent Guillainâ€“BarrÃ© Syndrome-Like Disease: Clues for Inherited CD59 Deficiency. Neuropediatrics, 2017, 48, 477-481.	0.6	16
100	Current Outline of Exon Skipping Trials in Duchenne Muscular Dystrophy. Genes, 2022, 13, 1241.	2.4	16
101	Genes for spinocerebellar ataxia with blindness and deafness (SCABD/SCAR3, MIM# 271250 and SCABD2). European Journal of Human Genetics, 2016, 24, 1154-1159.	2.8	15
102	Clinical presentation of anti-N-methyl- d -aspartate receptor and anti-voltage-gated potassium channel complex antibodies in children: A series of 24 cases. European Journal of Paediatric Neurology, 2018, 22, 135-142.	1.6	15
103	ATP8A2-related disorders as recessive cerebellar ataxia. Journal of Neurology, 2020, 267, 203-213.	3.6	15
104	Clinical and Histopathological Study of Merosin-deficient and Merosin-positive Congenital Muscular Dystrophy. Pediatric and Developmental Pathology, 2000, 3, 168-176.	1.0	14
105	Genotypeâ€“phenotype correlation in seven motor neuron disease families with novel <scp>ALS2</scp> mutations. American Journal of Medical Genetics, Part A, 2021, 185, 344-354.	1.2	14
106	Whole Exome Sequencing Reveals DYSF, FKTN, and ISPD Mutations in Congenital Muscular Dystrophy Without Brain or Eye Involvement. Journal of Neuromuscular Diseases, 2015, 2, 87-92.	2.6	13
107	Screening of deletions and RFLP analysis in Turkish DMD/BMD families by PCR. Clinical Genetics, 1993, 43, 261-266.	2.0	12
108	Congenital mirror movements in a patient with alpha-dystroglycanopathy due to a novel POMK mutation. Neuromuscular Disorders, 2017, 27, 239-242.	0.6	12

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109	Neurologic Involvement in Primary Immunodeficiency Disorders. Journal of Child Neurology, 2018, 33, 320-328.	1.4	12
110	Genetic and phenotypic features of patients with childhood ataxias diagnosed by next-generation sequencing gene panel. Brain and Development, 2020, 42, 6-18.	1.1	12
111	The Common miRNA Signatures Associated with Mitochondrial Dysfunction in Different Muscular Dystrophies. American Journal of Pathology, 2020, 190, 2136-2145.	3.8	11
112	Clinical features, muscle biopsy scores, myositis specific antibody profiles and outcome in juvenile dermatomyositis. Seminars in Arthritis and Rheumatism, 2021, 51, 95-100.	3.4	11
113	Challenges in pediatric chronic inflammatory demyelinating polyneuropathy. Neuromuscular Disorders, 2016, 26, 817-824.	0.6	10
114	Autosomal recessive spinocerebellar ataxia 18 caused by homozygous exon 14 duplication in GRID2 and review of the literature. Acta Neurologica Belgica, 2021, 121, 1457-1462.	1.1	10
115	Epidemiology of muscular dystrophies in the Mediterranean area. Acta Myologica, 2013, 32, 138-41.	1.5	10
116	Heart disease in Friedreich's ataxia: A clinical and echocardiographic study. Pediatrics International, 1996, 38, 308-311.	0.5	9
117	Assessment of left ventricular systolic and diastolic functions in children with merosin-positive congenital muscular dystrophy. International Journal of Cardiology, 2003, 87, 129-133.	1.7	9
118	A Boy With Spastic Paraparesis and Dyspnea. Journal of Child Neurology, 2004, 19, 397-398.	1.4	8
119	The association between trunk control and upper limb functions of children with Duchenne muscular dystrophy. Physiotherapy Theory and Practice, 2022, 38, 46-54.	1.3	8
120	Inflammatory milieu of muscle biopsies in juvenile dermatomyositis. Rheumatology International, 2021, 41, 77-85.	3.0	8
121	Reduced mitochondrial fission and impaired energy metabolism in human primary skeletal muscle cells of Megaconial Congenital Muscular Dystrophy. Scientific Reports, 2021, 11, 18161.	3.3	8
122	Effect of topiramate on enlargement of head in Canavan disease: a new option for treatment of megalencephaly. Turkish Journal of Pediatrics, 2004, 46, 67-71.	0.6	8
123	A novel mutation in the DGUOK gene in a Turkish newborn with mitochondrial depletion syndrome. Turkish Journal of Pediatrics, 2011, 53, 79-82.	0.6	8
124	Uncontrolled inflammation of the nervous system. Neurology: Clinical Practice, 2018, 8, e18-e20.	1.6	7
125	Biallelic hypomorphic mutations in HEATR5B, encoding HEAT repeat-containing protein 5B, in a neurological syndrome with pontocerebellar hypoplasia. European Journal of Human Genetics, 2021, 29, 957-964.	2.8	7
126	Is oxidative damage in operation in patients with hereditary spastic paraparesis?. Brain and Development, 2010, 32, 130-136.	1.1	6

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127	Disseminated Alveolar Rhabdomyosarcoma in a Child With Spinal Muscular Atrophy. Journal of Pediatric Hematology/Oncology, 2002, 24, 508-509.	0.6	5
128	Andermann Syndrome in a Turkish Patient. Journal of Child Neurology, 2003, 18, 76-79.	1.4	4
129	Infantile anti-MuSK positive myasthenia gravis in a patient with autoimmune polyendocrinopathy type 3. European Journal of Paediatric Neurology, 2014, 18, 526-528.	1.6	4
130	Diagnosis of Duchenne muscular dystrophy in Italy in the last decade: Critical issues and areas for improvements. Neuromuscular Disorders, 2017, 27, 973.	0.6	4
131	Assessment of Neurologic Disorders and Rare Intracranial Anomalies Associated With Cleft Lip and Palate. Journal of Craniofacial Surgery, 2018, 29, 2195-2197.	0.7	4
132	Common deletion of mitochondrial DNA in a 5-year-old girl with failure to thrive, ptosis, ophthalmoplegia and ragged-red fibers. Brain and Development, 1999, 21, 413-415.	1.1	3
133	Clinical, electrophysiological and neuropsychological findings of twenty-two children with mesial temporal sclerosis. Turkish Journal of Pediatrics, 2003, 45, 221-30.	0.6	3
134	An unusual presentation of gastrointestinal obstruction in a three-year-old boy. Turkish Journal of Pediatrics, 2009, 51, 195-8.	0.6	3
135	3-phosphoglycerate dehydrogenase deficiency: a case report of a treatable cause of seizures. Turkish Journal of Pediatrics, 2009, 51, 587-92.	0.6	3
136	Ultrasonographic assessment of lower limb muscle architecture in children with early-stage Duchenne muscular dystrophy. Arquivos De Neuro-Psiquiatria, 2022, 80, 475-481.	0.8	3
137	The effect of aerobic training on motor function and muscle architecture in children with Duchenne muscular dystrophy: A randomized controlled study. Clinical Rehabilitation, 2022, , 026921552210954.	2.2	3
138	Good clinical observation is essential before molecular studies. Lancet, The, 1995, 346, 1490.	13.7	2
139	Mystery Case: Pontine tegmental cap dysplasia in a neonate. Neurology, 2018, 91, e2100-e2101.	1.1	2
140	Merosin deficient congenital muscular dystrophy type 1A: An international workshop on the road to therapy 15-17 November 2019, Maastricht, the Netherlands. Neuromuscular Disorders, 2021, 31, 673-680.	0.6	2
141	Reply to Tzoulis et al.: Genetic and clinical heterogeneity of essential tremor. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E2269-E2269.	7.1	1
142	Core myopathies - a short review. Acta Myologica, 2020, 39, 266-273.	1.5	1
143	Treatment of a severe conversion disorder in a 10-year-old boy: a case study and overview. Turkish Journal of Pediatrics, 2012, 54, 413-8.	0.6	1
144	Expensive molecular therapies for rare genetic disorders: carrier detection or newborn screening should be the strategy. A personal opinion.. Journal of International Child Neurology Association, 2021, 1, .	0.0	1

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145	ALLEGATIONS OF TORTURE IN TURKEY. Lancet, The, 1989, 334, 220-221.	13.7	0
146	The Muscular Dystrophies. European Journal of Paediatric Neurology, 2003, 7, 91.	1.6	0
147	Nesprinopathy: A multi-faceted genetic disorder. European Journal of Paediatric Neurology, 2019, 23, 231.	1.6	0
148	Novel Missense ALDH3A2 Mutation in a Patient with SjÃ¶grenâ€“Larsson Syndrome. Journal of Pediatric Neurology, 2020, 18, 166-168.	0.2	0
149	Spinal muscular atrophy. Neurology, 2020, 95, 11-12.	1.1	0
150	RNA-based treatments in spinal muscular atrophy. Journal of the Neurological Sciences, 2021, 429, 117911.	0.6	0
151	Diagnosis of Quantitative Mitochondrial DNA Defects by Rapidly Prepared Whole Mitochondrial DNA Probe. Diagnostic Molecular Pathology, 2000, 9, 81-83.	2.1	0
152	Novel Lysosomal Positioning Defects Due to Biallelic Mutations in BORCS7 Causes a Neurodegenerative Disease Presenting as Hereditary-Spastic Paraplegia. Neuropediatrics, 2021, 52, .	0.6	0
153	The outcome of two SMA cases treated with nusinersen at seven hours and at three days of life: the earliest ever. Neuromuscular Disorders, 2022, 32, 575-577.	0.6	0