Sedat IÅ**ž**kay

List of Publications by Year in descending order

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567281 361022 1,418 63 15 35 citations h-index g-index papers 64 64 64 3319 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
2	Prevalence of orthorexia among medical students in Erzurum, Turkey. Comprehensive Psychiatry, 2010, 51, 49-54.	3.1	199
3	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	28.9	189
4	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	2.4	104
5	Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. Journal of Clinical Investigation, 2016, 126, 762-778.	8.2	82
6	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150.	6.2	74
7	Mutations in ANKLE2, a ZIKA Virus Target, Disrupt an Asymmetric Cell Division Pathway in Drosophila Neuroblasts to Cause Microcephaly. Developmental Cell, 2019, 51, 713-729.e6.	7.0	71
8	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	12.8	40
9	Viral etiological causes of febrile seizures for respiratory pathogens (EFES Study). Human Vaccines and Immunotherapeutics, 2019, 15, 496-502.	3.3	38
10	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005.	6.2	38
11	A multicenter cross-sectional study to evaluate the clinical characteristics and nutritional status of children with cerebral palsy. Clinical Nutrition ESPEN, 2018, 26, 27-34.	1.2	31
12	Neuroinfluenza: evaluation of seasonal influenza associated severe neurological complications in children (a multicenter study). Child's Nervous System, 2018, 34, 335-347.	1,1	29
13	Asymptomatic rhythm and conduction abnormalities in children with acute rheumatic fever: 24-hour electrocardiography study. Cardiology in the Young, 2010, 20, 620-630.	0.8	22
14	THE NEUROLOGICAL FACE OF CELIAC DISEASE. Arquivos De Gastroenterologia, 2015, 52, 167-170.	0.8	22
15	Characteristics of pediatric multiple sclerosis: The Turkish pediatric multiple sclerosis database. European Journal of Paediatric Neurology, 2017, 21, 864-872.	1.6	18
16	Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. American Journal of Human Genetics, 2018, 103, 794-807.	6.2	18
17	Restless leg syndrome in children with celiac disease. Turkish Journal of Pediatrics, 2018, 60, 70.	0.6	18
18	Prevalence of Celiac Disease in Children With Idiopathic Epilepsy in Southeast Turkey. Pediatric Neurology, 2014, 50, 479-481.	2.1	14

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19	Eyelid Myoclonia With Absence Seizures in a Child With I-2 Hydroxyglutaric Aciduria: Findings of Magnetic Resonance Imaging. Pediatric Neurology, 2012, 46, 195-197.	2.1	13
20	The Frequency of Epileptiform Discharges in Celiac Disease. Pediatric Neurology, 2015, 53, 78-82.	2.1	13
21	Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317.	6.2	11
22	An infant with trisomy 15 mosaicism. Clinical Dysmorphology, 2013, 22, 172-174.	0.3	9
23	Pseudohypoparathyroidism Presenting with Ventricular Arrhythmia: A Case Report. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2012, 4, 42-44.	0.9	8
24	INCREASED TISSUE TRANSGLUTAMINASE LEVELS ARE ASSOCIATED WITH INCREASED EPILEPTIFORM ACTIVITY IN ELECTROENCEPHALOGRAPHY AMONG PATIENTS WITH CELIAC DISEASE. Arquivos De Gastroenterologia, 2015, 52, 272-277.	0.8	7
25	RANBP2 Mutation in Clinically Undiagnosed Acute Necrotizing Encephalopathy. Indian Journal of Pediatrics, 2018, 85, 820-821.	0.8	7
26	Celiac disease with pulmonary haemosiderosis and cardiomyopathy. BMJ Case Reports, 2012, 2012, bcr2012007262-bcr2012007262.	0.5	6
27	PERIPHERAL NEUROPATHY ELECTROPHYSIOLOGICAL SCREENING IN CHILDREN WITH CELIAC DISEASE. Arquivos De Gastroenterologia, 2015, 52, 134-138.	0.8	6
28	Acute Mercury Poisoning in a Group of School Children. Pediatric Emergency Care, 2016, Publish Ahead of Print, 696-699.	0.9	6
29	Neurobrucellosis developing unilateral oculomotor nerve paralysis. American Journal of Emergency Medicine, 2012, 30, 2085.e5-2085.e7.	1.6	5
30	THE PREVALENCE OF CELIAC DISEASE AMONG PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER. Arquivos De Gastroenterologia, 2015, 52, 55-58.	0.8	5
31	Ecthyma gangrenosum in an infant after liver transplantation. Pediatrics International, 2016, 58, 950-952.	0.5	5
32	Colchicine Treatment in Children With Familial Mediterranean Fever: Is it a Risk Factor for Neuromyopathy?. Pediatric Neurology, 2013, 49, 417-419.	2.1	4
33	L-2 Hydroxyglutaric aciduria presenting with anxiety symptoms. BMJ Case Reports, 2013, 2013, bcr2013009512-bcr2013009512.	0.5	4
34	Imaging Findings Associated With Methylmalonic Aciduria. Pediatric Neurology, 2014, 50, 435-436.	2.1	4
35	Frequency of coeliac disease in children with breathâ€holding spells. Journal of Paediatrics and Child Health, 2014, 50, 916-919.	0.8	4
36	Cerebellar involvement of Griscelli syndrome type 2. BMJ Case Reports, 2014, 2014, bcr2014206703-bcr2014206703.	0.5	4

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37	An infant with glutaric aciduria type IIc diagnosed with a novel mutation. Turkish Journal of Pediatrics, 2017, 59, 315-317.	0.6	4
38	Prevalence of celiac disease in Turkish children with idiopathic epilepsy. Iranian Journal of Pediatrics, 2014, 24, 280-4.	0.3	3
39	Congenital cytomegalovirus infection and finger anomaly. BMJ Case Reports, 2013, 2013, bcr2013009486-bcr2013009486.	0.5	2
40	Cerebral Multicystic Lesions in a Child With L-2 Hydroxyglutaric Aciduria: A Rare Disease and a Rare Association. Pediatric Neurology, 2014, 50, 197-198.	2.1	2
41	Influenza A (H1N1) Infection Associated Acute Necrotizing Encephalopathy in a Child With Periodic Lateralized Epileptiform Discharges. Pediatric Emergency Care, 2016, 32, e14-e15.	0.9	2
42	Late diagnosis of pyridoxine-dependent epilepsy in two adolescent siblings. Annals of Indian Academy of Neurology, 2021, 24, 770.	0.5	2
43	Contribution of brain MRI in a patient diagnosed with 2-hydroxyglutaric aciduria. BMJ Case Reports, 2013, 2013, bcr2013008917-bcr2013008917.	0.5	2
44	Evaluation of two non-myasthenic patients with ptosis. Turk Pediatri Arsivi, 2018, 52, 240-243.	0.9	2
45	A child with L-2 hydroxyglutaric aciduria presenting with dilated cardiomyopathy: Coincidence or a new syndrome?. Anatolian Journal of Cardiology, 2013, 14, 92-3.	0.4	1
46	Cerebral multicystic lesions in a child with neurofibromatosis. BMJ Case Reports, 2013, 2013, bcr-2012-007639-bcr-2012-007639.	0.5	1
47	Cerebellar hypoplasia in a case with neurofibromatosis type 1. BMJ Case Reports, 2013, 2013, bcr2013202160-bcr2013202160.	0.5	1
48	Celiac Disease and Juvenile Absence Epilepsy. Pediatric Emergency Care, 2015, 31, e19-e20.	0.9	1
49	Pendular Nystagmus Associated With Cerebral Pylomixoid Astrocytoma. Pediatric Neurology, 2016, 61, 117-118.	2.1	1
50	Epilepsy and GI Disorders. , 2019, , .		1
51	L-2 Hydroxyglutaric aciduria presenting with status epilepticus. BMJ Case Reports, 2013, 2013, bcr2013010164-bcr2013010164.	0.5	1
52	Nominal dysphasia and euphoria caused by EBV encephalitis. BMJ Case Reports, 2013, 2013, bcr2012007514-bcr2012007514.	0.5	1
53	The behavior pattern of parents of patients with subacute sclerosing panencephalitis concerning alternative medicine. Turkish Journal of Pediatrics, 2017, 59, 288.	0.6	1
54	Occipital cortex dysgenesis with white matter changes due to mutations in Laminin a2. Turkish Journal of Pediatrics, 2017, 59, 338-341.	0.6	1

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55	Dorsal midbrain syndrome in two cases with two different presentations. Child's Nervous System, 2016, 32, 2283-2284.	1.1	0
56	Williams-Beuren Syndrome with Mirror Movements. Indian Journal of Pediatrics, 2016, 83, 1493-1494.	0.8	0
57	Foreign Body in the Scalp: A Rare Cause of Recurrent Headache. Indian Journal of Pediatrics, 2016, 83, 871-872.	0.8	0
58	An L-2-Hydroxyglutaric Aciduria Case Presented With Acute Bacterial Meningitis. Pediatric Emergency Care, 2017, 33, e1.	0.9	0
59	Idiopathic Unilateral Paralysis of the Palate in a Youth. Pediatric Emergency Care, 2018, 34, e104-e105.	0.9	0
60	Unusual hair findings in a child with cardiofaciocutaneous syndrome. International Journal of Dermatology, 2019, 58, 354-356.	1.0	0
61	Spontaneous Pneumocephalus Secondary to Positive Ventilation in an Infant. Indian Journal of Pediatrics, 2019, 86, 390-391.	0.8	0
62	Eating-induced electroclinical and electrographical seizures in a child. Acta Neurologica Belgica, 2021, , 1.	1.1	0
63	Congenital muscular dystrophy due to novel compound heterozygote mutations in POMGNT1 gene. Journal of Pediatric Neurosciences, 2018, 13, 462.	0.3	0