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	Late diagnosis of pyridoxine-dependent epilepsy in two adolescent siblings. Annals of Indian Academy of Neurology, 2021, 24, 770.	0.5	2
2	Eating-induced electroclinical and electrographical seizures in a child. Acta Neurologica Belgica, 2021, , 1.	1.1	0
3	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
4	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
5	Unusual hair findings in a child with cardiofaciocutaneous syndrome. International Journal of Dermatology, 2019, 58, 354-356.	1.0	O
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	Spontaneous Pneumocephalus Secondary to Positive Ventilation in an Infant. Indian Journal of Pediatrics, 2019, 86, 390-391.	0.8	O
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	Epilepsy and GI Disorders. , 2019, , .		1
10	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
11	Viral etiological causes of febrile seizures for respiratory pathogens (EFES Study). Human Vaccines and Immunotherapeutics, 2019, 15, 496-502.	3.3	38
12	RANBP2 Mutation in Clinically Undiagnosed Acute Necrotizing Encephalopathy. Indian Journal of Pediatrics, 2018, 85, 820-821.	0.8	7
13	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	2.4	104
14	Idiopathic Unilateral Paralysis of the Palate in a Youth. Pediatric Emergency Care, 2018, 34, e104-e105.	0.9	0
15	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
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	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
18	Restless leg syndrome in children with celiac disease. Turkish Journal of Pediatrics, 2018, 60, 70.	0.6	18

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19	Evaluation of two non-myasthenic patients with ptosis. Turk Pediatri Arsivi, 2018, 52, 240-243.	0.9	2
20	Congenital muscular dystrophy due to novel compound heterozygote mutations in POMGNT1 gene. Journal of Pediatric Neurosciences, 2018, 13, 462.	0.3	0
21	An L-2-Hydroxyglutaric Aciduria Case Presented With Acute Bacterial Meningitis. Pediatric Emergency Care, 2017, 33, e1.	0.9	0
22	Characteristics of pediatric multiple sclerosis: The Turkish pediatric multiple sclerosis database. European Journal of Paediatric Neurology, 2017, 21, 864-872.	1.6	18
23	An infant with glutaric aciduria type Ilc diagnosed with a novel mutation. Turkish Journal of Pediatrics, 2017, 59, 315-317.	0.6	4
24	The behavior pattern of parents of patients with subacute sclerosing panencephalitis concerning alternative medicine. Turkish Journal of Pediatrics, 2017, 59, 288.	0.6	1
25	Occipital cortex dysgenesis with white matter changes due to mutations in Laminin a2. Turkish Journal of Pediatrics, 2017, 59, 338-341.	0.6	1
26	Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. Journal of Clinical Investigation, 2016 , 126 , $762-778$.	8.2	82
27	Dorsal midbrain syndrome in two cases with two different presentations. Child's Nervous System, 2016, 32, 2283-2284.	1.1	0
28	Acute Mercury Poisoning in a Group of School Children. Pediatric Emergency Care, 2016, Publish Ahead of Print, 696-699.	0.9	6
29	Pendular Nystagmus Associated With Cerebral Pylomixoid Astrocytoma. Pediatric Neurology, 2016, 61, 117-118.	2.1	1
30	Ecthyma gangrenosum in an infant after liver transplantation. Pediatrics International, 2016, 58, 950-952.	0.5	5
31	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
32	Williams-Beuren Syndrome with Mirror Movements. Indian Journal of Pediatrics, 2016, 83, 1493-1494.	0.8	0
33	Foreign Body in the Scalp: A Rare Cause of Recurrent Headache. Indian Journal of Pediatrics, 2016, 83, 871-872.	0.8	0
34	Celiac Disease and Juvenile Absence Epilepsy. Pediatric Emergency Care, 2015, 31, e19-e20.	0.9	1
35	THE NEUROLOGICAL FACE OF CELIAC DISEASE. Arquivos De Gastroenterologia, 2015, 52, 167-170.	0.8	22
36	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

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37	THE PREVALENCE OF CELIAC DISEASE AMONG PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER. Arquivos De Gastroenterologia, 2015, 52, 55-58.	0.8	5
38	PERIPHERAL NEUROPATHY ELECTROPHYSIOLOGICAL SCREENING IN CHILDREN WITH CELIAC DISEASE. Arquivos De Gastroenterologia, 2015, 52, 134-138.	0.8	6
39	The Frequency of Epileptiform Discharges in Celiac Disease. Pediatric Neurology, 2015, 53, 78-82.	2.1	13
40	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
41	Prevalence of Celiac Disease in Children With Idiopathic Epilepsy in Southeast Turkey. Pediatric Neurology, 2014, 50, 479-481.	2.1	14
42	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
43	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	28.9	189
44	Imaging Findings Associated With Methylmalonic Aciduria. Pediatric Neurology, 2014, 50, 435-436.	2.1	4
45	Frequency of coeliac disease in children with breathâ€holding spells. Journal of Paediatrics and Child Health, 2014, 50, 916-919.	0.8	4
46	Cerebellar involvement of Griscelli syndrome type 2. BMJ Case Reports, 2014, 2014, bcr2014206703.	0.5	4
47	Prevalence of celiac disease in Turkish children with idiopathic epilepsy. Iranian Journal of Pediatrics, 2014, 24, 280-4.	0.3	3
48	Colchicine Treatment in Children With Familial Mediterranean Fever: Is it a Risk Factor for Neuromyopathy?. Pediatric Neurology, 2013, 49, 417-419.	2.1	4
49	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
50	An infant with trisomy 15 mosaicism. Clinical Dysmorphology, 2013, 22, 172-174.	0.3	9
51	Congenital cytomegalovirus infection and finger anomaly. BMJ Case Reports, 2013, 2013, bcr2013009486-bcr2013009486.	0.5	2
52	L-2 Hydroxyglutaric aciduria presenting with anxiety symptoms. BMJ Case Reports, 2013, 2013, bcr2013009512-bcr2013009512.	0.5	4
53	Cerebral multicystic lesions in a child with neurofibromatosis. BMJ Case Reports, 2013, 2013, bcr-2012-007639-bcr-2012-007639.	0.5	1
54	Cerebellar hypoplasia in a case with neurofibromatosis type 1. BMJ Case Reports, 2013, 2013, bcr2013202160-bcr2013202160.	0.5	1

SEDAT IÅŽIKAY

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55	Contribution of brain MRI in a patient diagnosed with 2-hydroxyglutaric aciduria. BMJ Case Reports, 2013, 2013, bcr2013008917-bcr2013008917.	0.5	2
56	L-2 Hydroxyglutaric aciduria presenting with status epilepticus. BMJ Case Reports, 2013, 2013, bcr2013010164-bcr2013010164.	0.5	1
57	Nominal dysphasia and euphoria caused by EBV encephalitis. BMJ Case Reports, 2013, 2013, bcr2012007514-bcr2012007514.	0.5	1
58	Pseudohypoparathyroidism Presenting with Ventricular Arrhythmia: A Case Report. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2012, 4, 42-44.	0.9	8
59	Celiac disease with pulmonary haemosiderosis and cardiomyopathy. BMJ Case Reports, 2012, 2012, bcr2012007262-bcr2012007262.	0.5	6
60	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
61	Neurobrucellosis developing unilateral oculomotor nerve paralysis. American Journal of Emergency Medicine, 2012, 30, 2085.e5-2085.e7.	1.6	5
62	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
63	Prevalence of orthorexia among medical students in Erzurum, Turkey. Comprehensive Psychiatry, 2010, 51, 49-54.	3.1	199