Adnan YÃ¼ksel

List of Publications by Year
in descending order

55 papers	1,999 citations	361296 20 h-index	265120 42 g-index
56 all docs	56 docs citations	56 times ranked	4765 citing authors

#	Article	IF	Citations
1	ASCâ€1 Is a Cell Cycle Regulator Associated with Severe and Mild Forms of Myopathy. Annals of Neurology, 2020, 87, 217-232.	2.8	12
2	The New CIC Mutation Associates with Mental Retardation and Severity of Seizure in Turkish Child with a Rare Class I Glucose-6-Phosphate Dehydrogenase Deficiency. Journal of Molecular Neuroscience, 2020, 70, 2077-2084.	1.1	3
3	New mutations in KCNT2 gene causing early infantile epileptic encephalopathy type 57: Case study and literature review. Acta Biochimica Polonica, 2020, 67, 431-434.	0.3	5
4	A novel mutation in the SERAC1 gene correlates with the severe manifestation of the MEGDEL phenotype, as revealed by wholeâ€‘exome sequencing. Experimental and Therapeutic Medicine, 2020, 19, 3505-3512.	0.8	4
5	New Genetic Approaches for Early Diagnosis and Treatment of Autism Spectrum Disorders. Review Journal of Autism and Developmental Disorders, 2019, 6, 367-380.	2.2	2
6	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. Nature Genetics, 2016, 48, 1071-1076.	9.4	314
7	Revealing the function of a novel splice-site mutation of CHD7 in CHARGE syndrome. Gene, 2016, 576, 776-781.	1.0	6
8	NovelPOC1Amutation in primordial dwarfism reveals new insights for centriole biogenesis. Human Molecular Genetics, 2015, 24, 5378-5387.	1.4	26
9	Whole-exome sequencing revealed two novel mutations in Usher syndrome. Gene, 2015, 563, 215-218.	1.0	6
10	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	3.8	258
11	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. Nature Genetics, 2015, 47, 73-77.	9.4	130
12	Melatonin attenuates phenytoin sodium-induced DNA damage. Drug and Chemical Toxicology, 2014, 37, 233-239.	1.2	16
13	Report of a patient with Templeâ€“Baraitser syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 848-851.	0.7	8
14	Keutel syndrome: Report of two novel MGP mutations and discussion of clinical overlap with arylsulfatase E deficiency and relapsing polychondritis. American Journal of Medical Genetics, Part A, 2014, 164, 1062-1068.	0.7	18
15	The drug-transporter gene MDR1 C3435T and G2677T/A polymorphisms and the risk of multidrug-resistant epilepsy in Turkish children. Molecular Biology Reports, 2014, 41, 331-336.	1.0	37
16	The Effect of Genetic Polymorphisms of Cytochrome P450 CYP2C9, CYP2C19, and CYP2D6 on Drug-Resistant Epilepsy in Turkish Children. Molecular Diagnosis and Therapy, 2014, 18, 229-236.	1.6	21
17	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	13.5	189
18	Poikiloderma with neutropenia: Genotypeâ€ethnic origin correlation, expanding phenotype and literature review. American Journal of Medical Genetics, Part A, 2014, 164, 2535-2540.	0.7	18

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19	Therapeutic Plasma Exchange for Malignant Refractory Status Epilepticus: A Case Report. Pediatric Neurology, 2014, 50, 407-410.	1.0	15
20	Demographic and Clinical Findings of Cerebral Palsy Patients in Istanbul: A Multicenter Study. FTR - Turkiye Fiziksel Tip Ve Rehabilitasyon Dergisi, 2014, 60, 134-138.	0.1	3
21	Deficiency of selenium and zinc as a causative factor for idiopathic intractable epilepsy. Epilepsy Research, 2013, 104, 35-39.	0.8	63
22	A novel EFNB1 mutation in a patient with craniofrontonasal syndrome and right hallux duplication. Gene, 2013, 527, 675-678.	1.0	5
23	Effects of memantine and melatonin on signal transduction pathways vascular leakage and brain injury after focal cerebral ischemia in mice. Neuroscience, 2013, 237, 268-276.	1.1	57
24	MicroRNA profiling in lymphocytes and serum of tyrosinemia type-I patients. Molecular Biology Reports, 2013, 40, 4619-4623.	1.0	5
25	Identification of a novel mutation in ZAP70 and prenatal diagnosis in a Turkish family with severe combined immunodeficiency disorder. Gene, 2013, 512, 189-193.	1.0	26
26	Circumferential skin folds and multiple anomalies. Clinical Dysmorphology, 2013, 22, 87-90.	0.1	15
27	A rare case of split hand/foot malformation with sensorineural hearing loss and Mondini dysplasia. Clinical Dysmorphology, 2013, 22, 33-35.	0.1	2
28	Vanishing white matter leukodystrophy, a rare case report. Turk Pediatri Arsivi, 2013, 48, 78-79.	0.9	0
29	Letter to the Editor Involvement of the corpus callosum splenium in a case with SSPE: magnetic resonance spectroscopy findings. Archives of Medical Science, 2013, 2, 386-387.	0.4	3
30	Evidence that membraneâ€bound G proteinâ€coupled melatonin receptors MT1 and MT2 are not involved in the neuroprotective effects of melatonin in focal cerebral ischemia. Journal of Pineal Research, 2012, 52, 228-235.	3.4	97
31	Glutathione S-Transferase M1, <i>GSTT1</i> and <i>GSTP1</i> Genetic Polymorphisms and the Risk of Age-Related Macular Degeneration. Ophthalmic Research, 2011, 46, 31-37.	1.0	25
32	Polymorphisms of the DNA Repair GenesXPDandXRCC1and the Risk of Age-Related Macular Degeneration. , 2010, 51, 4732.		37
33	Marked Improvement in Segawa Syndrome After l-Dopa and Selegiline Treatment. Pediatric Neurology, 2010, 42, 348-350.	1.0	7
34	Magnetic Resonance Imaging, Magnetic Resonance Spectroscopy, and Facial Dysmorphism in a Case of Lowe Syndrome with Novel OCRL1 Gene Mutation. Journal of Child Neurology, 2009, 24, 93-96.	0.7	8
35	Molecular genetic screening of MBS1 locus on chromosome 13 for microdeletions and exclusion of FGF9, GSH1 and CDX2 as causative genes in patients with Moebius syndrome. European Journal of Medical Genetics, 2009, 52, 315-320.	0.7	16
36	Mutational screening of BASP1 and transcribed processed pseudogene TPÎ¨g-BASP1 in patients with MÃ¶bius syndrome. Journal of Genetics and Genomics, 2009, 36, 251-256.	1.7	3

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37	Spontaneous Intracranial Hypotension Syndrome in a Patient With Marfan Syndrome and Autosomal Dominant Polycystic Kidney Disease. Headache, 2008, 48, 632-636.	1.8	12
38	Warburg Micro syndrome in a Turkish boy. Clinical Dysmorphology, 2007, 16, 89-93.	0.1	18
39	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	2.6	137
40	Evaluation of mental retardation - Part 1: Etiologic classification of 4659 patients with mental retardation or multiple congenital abnormality and mental retardation. Journal of Pediatric Neurosciences, 2007, 2, 45.	0.2	2
41	Evaluation of mental retardation - Part 2: The factors that elucidate the etiologic diagnosis of the patients with mental retardation or multiple congenital abnormality and mental retardation. Journal of Pediatric Neurosciences, 2007, 2, 53.	0.2	0
42	Facial Dysmorphism in Leigh Syndrome With SURF-1 Mutation and COX Deficiency. Pediatric Neurology, 2006, 34, 486-489.	1.0	14
43	The Effects of Vigabatrin on Rat Liver Antioxidant Status. Drug Metabolism and Drug Interactions, 2005, 21, 109-15.	0.3	4
44	Epilepsy in vacuolating megalencephalic leukoencephalopathy with subcortical cysts. Seizure: the Journal of the British Epilepsy Association, 2003, 12, 388-396.	0.9	25
45	Neuroblastoma in a dysmorphic girl with a partial duplication of 2p caused by an unbalanced translocation. Clinical Dysmorphology, 2002, 11, 39-42.	0.1	10
46	Changes in the Antioxidant System in Epileptic Children Receiving Antiepileptic Drugs: Two-Year Prospective Studies. Journal of Child Neurology, 2001, 16, 603-606.	0.7	47
47	Erythrocyte Glutathione, Glutathione Peroxidase, Superoxide Dismutase and Serum Lipid Peroxidation in Epileptic Children With Valproate and Carbamazepine Monotherapy. Journal of Basic and Clinical Physiology and Pharmacology, 2000, 11, 73-81.	0.7	57
48	Siblings With Cystic Leukoencephalopathy and Megalencephaly. Journal of Child Neurology, 2000, 15, 690-693.	0.7	7
49	THE EFFECTS OF CARBAMAZEPINE AND VALPROIC ACID ON THE ERYTHROCYTE GLUTATHIONE, GLUTATHIONE PEROXIDASE, SUPEROXIDE DISMUTASE AND SERUM LIPID PEROXIDATION IN EPILEPTIC CHILDREN. Pharmacological Research, 2000, 41, 423-425.	3.1	81
50	N-acetyl-Î²-glucosaminidase and Î²-galactosidase activity in children receiving antiepileptic drugs. Pediatric Neurology, 1999, 20, 24-26.	1.0	19
51	Neuroimaging findings of four patients with Sandhoff disease. Pediatric Neurology, 1999, 21, 562-565.	1.0	29
52	Effects of carbamazepine and valproate on brainstem auditory evoked potentials in epileptic children. Child's Nervous System, 1995, 11, 474-477.	0.6	21
53	Effect of valproate and carbamazepine on visual evoked potentials in epileptic children. Pediatrics International, 1995, 37, 358-361.	0.2	19
54	Serum thyroid hormones and pituitary response to thyrotropinâ€releasing hormone in epileptic children receiving antiâ€epileptic medication. Pediatrics International, 1993, 35, 108-112.	0.2	20

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55	Influence of longâ€term carbamazepine treatment on thyroid function. Pediatrics International, 1993, 35, 229-232.	0.2	10