

AyÅe NazlÄ± BaÅak

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/946238/publications.pdf>

Version: 2024-02-01

46
papers

1,938
citations

471509

17
h-index

289244

40
g-index

48
all docs

48
docs citations

48
times ranked

3704
citing authors

#	ARTICLE	IF	CITATIONS
1	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	8.1	517
2	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	21.4	494
3	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	21.4	218
4	The distinct genetic pattern of ALS in Turkey and novel mutations. <i>Neurobiology of Aging</i> , 2015, 36, 1764.e9-1764.e18.	3.1	78
5	Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. <i>Cell Reports</i> , 2019, 26, 2298-2306.e5.	6.4	57
6	Association of Variants in the <i>SPTLC1</i> Gene With Juvenile Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2021, 78, 1236.	9.0	46
7	ATXN2 and Its Neighbouring Gene SH2B3 Are Associated with Increased ALS Risk in the Turkish Population. <i>PLoS ONE</i> , 2012, 7, e42956.	2.5	43
8	The genetic structure of the Turkish population reveals high levels of variation and admixture. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	7.1	42
9	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. <i>Science Translational Medicine</i> , 2022, 14, eabj0264.	12.4	38
10	Î²-THALASSEMIA IN TURKEY: A REVIEW OF THE CLINICAL, EPIDEMIOLOGICAL, MOLECULAR, AND EVOLUTIONARY ASPECTS. <i>Hemoglobin</i> , 2001, 25, 227-239.	0.8	36
11	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. <i>Brain</i> , 2021, 144, 769-780.	7.6	33
12	Comprehensive Research on Past and Future Therapeutic Strategies Devoted to Treatment of Amyotrophic Lateral Sclerosis. <i>International Journal of Molecular Sciences</i> , 2022, 23, 2400.	4.1	32
13	FBXO7 R498X mutation: Phenotypic variability from chorea to early onset parkinsonism within a family. <i>Parkinsonism and Related Disorders</i> , 2014, 20, 1253-1256.	2.2	29
14	Elevated Global DNA Methylation Is Not Exclusive to Amyotrophic Lateral Sclerosis and Is Also Observed in Spinocerebellar Ataxia Types 1 and 2. <i>Neurodegenerative Diseases</i> , 2018, 18, 38-48.	1.4	27
15	ERLIN1 mutations cause teenage-onset slowly progressive ALS in a large Turkish pedigree. <i>European Journal of Human Genetics</i> , 2018, 26, 745-748.	2.8	25
16	Cerebellar cognitive-affective syndrome preceding ataxia associated with complex extrapyramidal features in a Turkish SCA48 family. <i>Neurogenetics</i> , 2020, 21, 51-58.	1.4	22
17	PRENATAL DIAGNOSIS OF Î²-THALASSAEMIA AND SICKLE CELL ANAEMIA IN TURKEY. , 1996, 16, 252-258.		21
18	Homozygous <i>CAPN1</i> mutations causing a spastic-ataxia phenotype in 2 families. <i>Neurology: Genetics</i> , 2018, 4, e218.	1.9	19

#	ARTICLE	IF	CITATIONS
19	Both Ubiquitin Ligases FBXW8 and PARK2 Are Sequestered into Insolubility by ATXN2 PolyQ Expansions, but Only FBXW8 Expression Is Dysregulated. PLoS ONE, 2015, 10, e0121089.	2.5	18
20	A novel PNPLA6 mutation in a Turkish family with intractable Holmes tremor and spastic ataxia. Neurological Sciences, 2021, 42, 1535-1539.	1.9	13
21	The first biallelic missense mutation in the FXN gene in a consanguineous Turkish family with Charcot-Marie-Tooth-like phenotype. Neurogenetics, 2020, 21, 73-78.	1.4	11
22	Turkish families with juvenile motor neuron disease broaden the phenotypic spectrum of <i>SPG11</i>. Neurology: Genetics, 2015, 1, e25.	1.9	10
23	Revisiting the complex architecture of ALS in Turkey: Expanding genotypes, shared phenotypes, molecular networks, and a public variant database. Human Mutation, 2020, 41, e7-e45.	2.5	10
24	The Effect of <sc><i>SMN</i></sc> Gene Dosage on <sc>ALS</sc> Risk and Disease Severity. Annals of Neurology, 2021, 89, 686-697.	5.3	10
25	A novel homozygous FBXO38 variant causes an early-onset distal hereditary motor neuronopathy type IID. Journal of Human Genetics, 2019, 64, 1141-1144.	2.3	9
26	The Complex Genetic Landscape of Hereditary Ataxias in Turkey and Implications in Clinical Practice. Movement Disorders, 2021, 36, 1676-1688.	3.9	9
27	De Novo and Dominantly Inherited <sc><i>SPTAN1</i></sc> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	3.9	9
28	Successful treatment of Juvenile parkinsonism with bilateral subthalamic deep brain stimulation in a 14-year-old patient with parkin gene mutation. Parkinsonism and Related Disorders, 2016, 24, 137-138.	2.2	8
29	A novel pathogenic variant in the 3Ê¹ end of the AGTPBP1 gene gives rise to neurodegeneration without cerebellar atrophy: an expansion of the disease phenotype?. Neurogenetics, 2021, 22, 127-132.	1.4	7
30	A Turkish Family with a Familial ALS-positive UBQLN2-S340I Mutation. Noropsikiyatri Arsivi, 2016, 53, 283-285.	0.7	5
31	Earlyâ€œOnset Parkinson's Disease: A Novel Deletion Comprising the <sc>DJ</sc>â€œ1 and <sc>TNFRSF9</sc> Genes. Movement Disorders, 2021, 36, 2973-2976.	3.9	5
32	Coexistence of autosomal recessive spastic ataxia of Charlevoix Saguenay and spondyloepiphyseal dysplasia in a Turkish patient. Journal of the Neurological Sciences, 2015, 357, 290-291.	0.6	4
33	Atypical Features in a Large Turkish Family Affected with Friedreich Ataxia. Case Reports in Neurological Medicine, 2016, 2016, 1-7.	0.4	4
34	The genetic basis of asymptomatic codon 8 frameâ€œshift (<i><sc>HBB</sc></i>:c25_26del<sc>AA</sc>) Î²⁰â€œthalassaemia homozygotes. British Journal of Haematology, 2016, 172, 958-965.	2.5	4
35	Braitâ€œFahnâ€œSchwarz disease: Parkinsonâ€œ™s disease and amyotrophic lateral sclerosis complex. Acta Neurologica Belgica, 2016, 116, 401-403.	1.1	4
36	Impact of Genetic Defects on Coronary Atherosclerosis among Turkish Cypriots. Heart Surgery Forum, 2017, 20, 223.	0.5	4

#	ARTICLE	IF	CITATIONS
37	Rare Î²-thalassemia mutation IVS-II-848 (C-A) first reported in a Turkish cyprriot family. , 1997, 54, 338-339.		3
38	Impact of the Amyotrophic Lateral Sclerosis Disease on the Biomechanical Properties and Oxidative Stress Metabolism of the Lung Tissue Correlated With the Human Mutant SOD1G93A Protein Accumulation. <i>Frontiers in Bioengineering and Biotechnology</i> , 2022, 10, 810243.	4.1	3
39	A rare case of juvenile amyotrophic lateral sclerosis. <i>Turkish Journal of Pediatrics</i> , 2021, 63, 495.	0.6	2
40	Ataxia telangiectasia like disorder: Another dopa-responsive disorder look-alike?. <i>Parkinsonism and Related Disorders</i> , 2020, 74, 22-24.	2.2	1
41	Vestibulo-ocular reflex impairment in SPG7 hereditary spastic paraplegia. <i>Clinical Neurophysiology</i> , 2021, 132, 77-79.	1.5	1
42	Two cases of early-onset autosomal recessive spastic ataxia of Charlevoix-Saguenay diagnosed in adulthood. <i>Clinical Neurology and Neurosurgery</i> , 2021, 201, 106423.	1.4	1
43	A combined clinical and computational approach to understand the SOD1A4T-mediated pathogenesis of rapidly progressive familial amyotrophic lateral sclerosis. <i>Acta Neurologica Belgica</i> , 2021, , 1.	1.1	1
44	Varied phenotypic spectrum presenting of paroxysmal exerciseâ€“induced dyskinesia: a Turkish family with SLC2A1 mutation. <i>Neurological Sciences</i> , 2021, 42, 4751-4754.	1.9	1
45	Phenotypic and Genotypic Analysis of Hereditary Ataxia Patients in Sakarya City, Turkey. <i>Noropsikiyatri Arsivi</i> , 2017, 56, 106-109.	0.3	0
46	Reply to letter to the editor by De Michele et al. <i>Neurogenetics</i> , 2020, 21, 147-147.	1.4	0