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List of Publications by Year
in descending order

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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	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	3.9	9
2	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. Frontiers in Bioengineering and Biotechnology, 2022, 10, 810243.	4.1	3
3	Comprehensive Research on Past and Future Therapeutic Strategies Devoted to Treatment of Amyotrophic Lateral Sclerosis. International Journal of Molecular Sciences, 2022, 23, 2400.	4.1	32
4	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. Science Translational Medicine, 2022, 14, eabj0264.	12.4	38
5	Vestibulo-ocular reflex impairment in SPG7 hereditary spastic paraplegia. Clinical Neurophysiology, 2021, 132, 77-79.	1.5	1
6	A novel PNPLA6 mutation in a Turkish family with intractable Holmes tremor and spastic ataxia. Neurological Sciences, 2021, 42, 1535-1539.	1.9	13
7	Two cases of early-onset autosomal recessive spastic ataxia of Charlevoix-Saguenay diagnosed in adulthood. Clinical Neurology and Neurosurgery, 2021, 201, 106423.	1.4	1
8	A rare case of juvenile amyotrophic lateral sclerosis. Turkish Journal of Pediatrics, 2021, 63, 495.	0.6	2
9	The Effect of <scp>SMN</scp> Gene Dosage on <scp>ALS</scp> Risk and Disease Severity. Annals of Neurology, 2021, 89, 686-697.	5.3	10
10	A combined clinical and computational approach to understand the SOD1A4T-mediated pathogenesis of rapidly progressive familial amyotrophic lateral sclerosis. Acta Neurologica Belgica, 2021, , 1.	1.1	1
11	The Complex Genetic Landscape of Hereditary Ataxias in Turkey and Implications in Clinical Practice. Movement Disorders, 2021, 36, 1676-1688.	3.9	9
12	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
13	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
14	Varied phenotypic spectrum presenting of paroxysmal exerciseâ€“induced dyskinesia: a Turkish family with SLC2A1 mutation. Neurological Sciences, 2021, 42, 4751-4754.	1.9	1
15	The genetic structure of the Turkish population reveals high levels of variation and admixture. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	42
16	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
17	Earlyâ€Onset Parkinson's Disease: A Novel Deletion Comprising the <scp>DJ</scp>â€1 and <scp>TNFRSF9</scp> Genes. Movement Disorders, 2021, 36, 2973-2976.	3.9	5
18	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

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19	Cerebellar cognitive-affective syndrome preceding ataxia associated with complex extrapyramidal features in a Turkish SCA48 family. Neurogenetics, 2020, 21, 51-58.	1.4	22
20	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
21	Reply to letter to the editor by De Michele et al. Neurogenetics, 2020, 21, 147-147.	1.4	0
22	Ataxia telangiectasia like disorder: Another dopa-responsive disorder look-alike?. Parkinsonism and Related Disorders, 2020, 74, 22-24.	2.2	1
23	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
24	Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. Cell Reports, 2019, 26, 2298-2306.e5.	6.4	57
25	ERLIN1 mutations cause teenage-onset slowly progressive ALS in a large Turkish pedigree. European Journal of Human Genetics, 2018, 26, 745-748.	2.8	25
26	Homozygous CAPN1 mutations causing a spastic-ataxia phenotype in 2 families. Neurology: Genetics, 2018, 4, e218.	1.9	19
27	Elevated Global DNA Methylation Is Not Exclusive to Amyotrophic Lateral Sclerosis and Is Also Observed in Spinocerebellar Ataxia Types 1 and 2. Neurodegenerative Diseases, 2018, 18, 38-48.	1.4	27
28	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
29	Phenotypic and Genotypic Analysis of Hereditary Ataxia Patients in Sakarya City, Turkey. Noropsikiyatri Arsivi, 2017, 56, 106-109.	0.3	0
30	Impact of Genetic Defects on Coronary Atherosclerosis among Turkish Cypriots. Heart Surgery Forum, 2017, 20, 223.	0.5	4
31	Atypical Features in a Large Turkish Family Affected with Friedreich Ataxia. Case Reports in Neurological Medicine, 2016, 2016, 1-7.	0.4	4
32	The genetic basis of asymptomatic codon 8 frameâ€shift (<scp>HBB</scp>:c25_26del<scp>AA</scp>) Î²⁰â€thalassaemia homozygotes. British Journal of Haematology, 2016, 172, 958-965.	2.5	4
33	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	21.4	494
34	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
35	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
36	Braitâ€“Fahnâ€“Schwarz disease: Parkinsonâ€™s disease and amyotrophic lateral sclerosis complex. Acta Neurologica Belgica, 2016, 116, 401-403.	1.1	4

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37	A Turkish Family with a Familial ALS-positive UBQLN2-S340I Mutation. Noropsikiyatri Arsivi, 2016, 53, 283-285.	0.7	5
38	Both Ubiquitin Ligases FBXW8 and PARK2 Are Sequestrated into Insolubility by ATXN2 PolyQ Expansions, but Only FBXW8 Expression Is Dysregulated. PLoS ONE, 2015, 10, e0121089.	2.5	18
39	Turkish families with juvenile motor neuron disease broaden the phenotypic spectrum of SPG11. Neurology: Genetics, 2015, 1, e25.	1.9	10
40	The distinct genetic pattern of ALS in Turkey and novel mutations. Neurobiology of Aging, 2015, 36, 1764.e9-1764.e18.	3.1	78
41	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
42	FBXO7â€“R498X mutation: Phenotypic variability from chorea to early onset parkinsonism within a family. Parkinsonism and Related Disorders, 2014, 20, 1253-1256.	2.2	29
43	ATXN2 and Its Neighbouring Gene SH2B3 Are Associated with Increased ALS Risk in the Turkish Population. PLoS ONE, 2012, 7, e42956.	2.5	43
44	Î²-THALASSEMIA IN TURKEY: A REVIEW OF THE CLINICAL, EPIDEMIOLOGICAL, MOLECULAR, AND EVOLUTIONARY ASPECTS. Hemoglobin, 2001, 25, 227-239.	0.8	36
45	Rare Î²-thalassemia mutation IVS-II-848 (C-A) first reported in a Turkish cypriot family. , 1997, 54, 338-339.		3
46	PRENATAL DIAGNOSIS OF Î²-THALASSAEMIA AND SICKLE CELL ANAEMIA IN TURKEY. , 1996, 16, 252-258.		21