Grace Yoon

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/2593460/publications.pdf

Version: 2024-02-01

86 papers	2,872 citations	236925 25 h-index	206112 48 g-index
86 all docs	86 docs citations	86 times ranked	6044 citing authors

#	Article	IF	Citations
1	The Clinician-reported Genetic testing Utility InDEx (C-GUIDE): Preliminary evidence of validity and reliability. Genetics in Medicine, 2022, 24, 430-438.	2.4	8
2	Heterozygous De Novo <scp>KPNA3</scp> Mutations Cause Complex Hereditary Spastic Paraplegia. Annals of Neurology, 2022, 91, 730-732.	5.3	1
3	An integrated modelling methodology for estimating global incidence and prevalence of hereditary spastic paraplegia subtypes SPG4, SPG7, SPG11, and SPG15. BMC Neurology, 2022, 22, 115.	1.8	7
4	Clinical profile and multidisciplinary needs of patients with neuromuscular disorders transitioning from paediatric to adult care. Neuromuscular Disorders, 2022, 32, 206-212.	0.6	0
5	FXR1-related congenital myopathy: expansion of the clinical and genetic spectrum. Journal of Medical Genetics, 2022, 59, 1069-1074.	3.2	1
6	Genetic, structural and clinical analysis of spastic paraplegia 4. Parkinsonism and Related Disorders, 2022, 98, 62-69.	2.2	7
7	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	3.6	26
8	Evidence for Nonâ€Mendelian Inheritance in Spastic Paraplegia 7. Movement Disorders, 2021, 36, 1664-1675.	3.9	11
9	<scp>GCH1</scp> mutations in hereditary spastic paraplegia. Clinical Genetics, 2021, 100, 51-58.	2.0	5
10	International retrospective natural history study of LMNA-related congenital muscular dystrophy. Brain Communications, 2021, 3, fcab075.	3.3	17
11	Neurodegenerative VPS41 variants inhibit HOPS function and mTORC1â€dependent TFEB/TFE3 regulation. EMBO Molecular Medicine, 2021, 13, e13258.	6.9	26
12	Scoliosis in Friedreich's ataxia: longitudinal characterization in a large heterogeneous cohort. Annals of Clinical and Translational Neurology, 2021, 8, 1239-1250.	3.7	11
13	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
14	De novo variants in POLR3B cause ataxia, spasticity, and demyelinating neuropathy. American Journal of Human Genetics, 2021, 108, 186-193.	6.2	19
15	De Novo variants in EEF2 cause a neurodevelopmental disorder with benign external hydrocephalus. Human Molecular Genetics, 2021, 29, 3892-3899.	2.9	11
16	De novo pathogenic variant in SETX causes a rapidly progressive neurodegenerative disorder of early childhood-onset with severe axonal polyneuropathy. Acta Neuropathologica Communications, 2021, 9, 194.	5.2	5
17	Health related quality of life in Friedreich Ataxia in a large heterogeneous cohort. Journal of the Neurological Sciences, 2020, 410, 116642.	0.6	18
18	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	7.6	29

#	Article	IF	Citations
19	De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. American Journal of Human Genetics, 2020, 107, 352-363.	6.2	64
20	Heterozygous Variants in KDM4B Lead to Global Developmental Delay and Neuroanatomical Defects. American Journal of Human Genetics, 2020, 107, 1170-1177.	6.2	13
21	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	2.4	15
22	FLVCR1-related disease as a rare cause of retinitis pigmentosa and hereditary sensory autonomic neuropathy. European Journal of Medical Genetics, 2020, 63, 104037.	1.3	3
23	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. Movement Disorders Clinical Practice, 2020, 7, 940-949.	1.5	7
24	Hypolipidaemia among patients with PMM2-CDG is associated with low circulating PCSK9 levels: a case report followed by observational and experimental studies. Journal of Medical Genetics, 2020, 57, 11-17.	3.2	8
25	Pearls & Oy-sters: Fatal brain edema is a rare complication of severe CACNA1A-related disorder. Neurology, 2020, 94, 631-634.	1.1	7
26	Reply: IREB2-associated neurodegeneration. Brain, 2019, 142, e41-e41.	7.6	3
27	Absence of iron-responsive element-binding protein 2 causes a novel neurodegenerative syndrome. Brain, 2019, 142, 1195-1202.	7.6	38
28	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. Nature Communications, 2019, 10, 797.	12.8	24
29	Homozygous pathogenic variant in BRAT1 associated with nonprogressive cerebellar ataxia. Neurology: Genetics, 2019, 5, e359.	1.9	13
30	Severe cystic degeneration and intractable seizures in a newborn with molybdenum cofactor deficiency type B. Molecular Genetics and Metabolism Reports, 2019, 18, 11-13.	1.1	7
31	Bi-allelic mutations ofLONP1encoding the mitochondrial LonP1 protease cause pyruvate dehydrogenase deficiency and profound neurodegeneration with progressive cerebellar atrophy. Human Molecular Genetics, 2019, 28, 290-306.	2.9	27
32	Clinical spectrum of POLR3-related leukodystrophy caused by biallelic POLR1C pathogenic variants. Neurology: Genetics, 2019, 5, e369.	1.9	38
33	Impact of Mobility Device Use on Quality of Life in Children With Friedreich Ataxia. Journal of Child Neurology, 2018, 33, 397-404.	1.4	5
34	Periodic reanalysis of whole-genome sequencing data enhances the diagnostic advantage over standard clinical genetic testing. European Journal of Human Genetics, 2018, 26, 740-744.	2.8	88
35	Response to phenotypic hetergeneity of POMT2 variants. American Journal of Medical Genetics, Part A, 2018, 176, 746-747.	1.2	0
36	POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e1-e1.	7.6	17

#	Article	IF	Citations
37	Rationale for dopaâ€responsive CTNNB1/ÃŸâ€catenin deficient dystonia. Movement Disorders, 2018, 33, 656-657.	3.9	8
38	Triple A syndrome presenting as complicated hereditary spastic paraplegia. Molecular Genetics & Genomic Medicine, 2018, 6, 1134-1139.	1.2	11
39	Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. Orphanet Journal of Rare Diseases, 2018, 13, 86.	2.7	29
40	Nonsyndromic cerebellar ataxias associated with disorders of DNA single-strand break repair. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 155, 105-115.	1.8	36
41	Interferon-Stimulated Gene Expression as a Preferred Biomarker for Disease Activity in Aicardiâ€“GoutiÃ¨res Syndrome. Journal of Interferon and Cytokine Research, 2017, 37, 147-152.	1.2	8
42	Congenital myopathy with â€œcoronaâ€•fibres, selective muscle atrophy, and craniosynostosis associated with novel recessive mutations in SCN4A. Neuromuscular Disorders, 2017, 27, 574-580.	0.6	23
43	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
44	The genetic landscape of familial congenital hydrocephalus. Annals of Neurology, 2017, 81, 890-897.	5.3	108
45	Severe neurodegeneration, progressive cerebral volume loss and diffuse hypomyelination associated with a homozygous frameshift mutation in CSTB. European Journal of Human Genetics, 2017, 25, 775-778.	2.8	24
46	<scp>KCNA2</scp> mutations are rare in hereditary spastic paraplegia. Annals of Neurology, 2017, 81, 325-326.	5.3	0
47	Clinical and genetic study of hereditary spastic paraplegia in Canada. Neurology: Genetics, 2017, 3, e122.	1.9	82
48	XRCC1 mutation is associated with PARP1 hyperactivation and cerebellar ataxia. Nature, 2017, 541, 87-91.	27.8	209
49	Complex genomic rearrangement in SPG11 due to a DNA replicationâ€based mechanism. Movement Disorders, 2017, 32, 1792-1794.	3.9	1
50	Noncompaction cardiomyopathy in an infant with Walkerâ€“Warburg syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3082-3086.	1.2	8
51	Subacute demyelinating peripheral neuropathy as a novel presentation of late infantile metachromatic leukodystrophy. Muscle and Nerve, 2017, 56, E41-E44.	2.2	5
52	Impact of diabetes in the Friedreich ataxia clinical outcome measures study. Annals of Clinical and Translational Neurology, 2017, 4, 622-631.	3.7	16
53	Andermann Syndrome in a Pakistani Family Caused by a Novel Mutation in SLC12A6. Journal of Pediatric Neurology, 2017, 15, 090-094.	0.2	2
54	A novel KCNA1 mutation in a family with episodic ataxia and malignant hyperthermia. Neurogenetics, 2016, 17, 245-249.	1.4	21

#	Article	IF	Citations
55	Triplication of 16p12.1p12.3 associated with developmental and growth delay and distinctive facial features. American Journal of Medical Genetics, Part A, 2016, 170, 712-716.	1.2	2
56	High Frequency of Pathogenic Rearrangements in SPG11 and Extensive Contribution of Mutational Hotspots and Founder Alleles. Human Mutation, 2016, 37, 703-709.	2.5	12
57	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
58	Comorbid Medical Conditions in Friedreich Ataxia. Journal of Child Neurology, 2016, 31, 1161-1165.	1.4	6
59	Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2016, 98, 1038-1046.	6.2	96
60	The Pediatric Cerebellum in Inherited Neurodegenerative Disorders. Neuroimaging Clinics of North America, 2016, 26, 373-416.	1.0	17
61	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	3.3	35
62	Reply: Autosomal recessive cerebellar ataxia caused by a homozygous mutation inPMPCA. Brain, 2016, 139, e20-e20.	7.6	3
63	Lethal Disorder of Mitochondrial Fission Caused by Mutations in DNM1L. Journal of Pediatrics, 2016, 171, 313-316.e2.	1.8	67
64	Neuromuscular conditions associated with malignant hyperthermia in paediatric patients: A 25-year retrospective study. Neuromuscular Disorders, 2016, 26, 201-206.	0.6	24
65	Biallelic Mutations in UNC80 Cause Persistent Hypotonia, Encephalopathy, Growth Retardation, and Severe Intellectual Disability. American Journal of Human Genetics, 2016, 98, 202-209.	6.2	45
66	X-linked Joubert syndrome: Neuroimaging and clinical features associated with a novel mutation in oral-facial-digital syndrome type 1. Journal of Pediatric Neurology, 2015, 12, 157-160.	0.2	1
67	Frataxin levels in peripheral tissue in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2015, 2, 831-842.	3.7	55
68	Further Insights into the Allan-Herndon-Dudley Syndrome: Clinical and Functional Characterization of a Novel MCT8 Mutation. PLoS ONE, 2015, 10, e0139343.	2.5	23
69	Massive CAG Repeat Expansion and Somatic Instability in Maternally Transmitted Infantile Spinocerebellar Ataxia Type 7. JAMA Neurology, 2015, 72, 219.	9.0	22
70	Cognitive and Neurobehavioral Profile in Boys With Duchenne Muscular Dystrophy. Journal of Child Neurology, 2015, 30, 1472-1482.	1.4	134
71	Prevalence of inherited neurotransmitter disorders in patients with movement disorders and epilepsy: a retrospective cohort study. Orphanet Journal of Rare Diseases, 2015, 10, 12.	2.7	18
72	â€œBoth Sides of the Wheelchairâ€: The Views of Individuals with, and Parents of Individuals with Friedreich Ataxia Regarding Preâ€symptomatic Testing of Minors. Journal of Genetic Counseling, 2015, 24, 732-743.	1.6	3

#	Article	IF	Citations
73	PMPCAmutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. Brain, 2015, 138, 1505-1517.	7.6	58
74	Complete APTX deletion in a patient with ataxia with oculomotor apraxia type 1. BMC Medical Genetics, 2015, 16, 61.	2.1	4
75	Recessive mutations in POLR1C cause a leukodystrophy by impairing biogenesis of RNA polymerase III. Nature Communications, 2015, 6, 7623.	12.8	127
76	Exome Sequencing as a Diagnostic Tool for Pediatricâ€Onset Ataxia. Human Mutation, 2014, 35, 45-49.	2.5	91
77	Prospective study of activities of daily living outcomes in children with cerebellar atrophy. Developmental Medicine and Child Neurology, 2014, 56, 460-467.	2.1	3
78	Mosaic microdeletion of 17p11.2â€“p12 and duplication of 17q22â€“q24 in a girl with Smithâ€–Magenis phenotype and peripheral neuropathy. American Journal of Medical Genetics, Part A, 2014, 164, 748-752.	1.2	6
79	CHD2 haploinsufficiency is associated with developmental delay, intellectual disability, epilepsy and neurobehavioural problems. Journal of Neurodevelopmental Disorders, 2014, 6, 9.	3.1	71
80	Cardiac Transplantation in Friedreich Ataxia. Journal of Child Neurology, 2012, 27, 1193-1196.	1.4	11
81	Diagnostic Approach to Childhood-Onset Cerebellar Atrophy. Journal of Child Neurology, 2012, 27, 1121-1132.	1.4	42
82	Cockayne syndrome caused by paternally inherited 5Mb deletion of 10q11.2 and a frameshift mutation of ERCC6. European Journal of Medical Genetics, 2011, 54, 272-276.	1.3	136
83	Mitochondrial mimicry of multiple system atrophy of the cerebellar subtype. Movement Disorders, 2011, 26, 753-755.	3.9	17
84	2q23 de novo microdeletion involving the MBD5 gene in a patient with developmental delay, postnatal microcephaly and distinct facial features. American Journal of Medical Genetics, Part A, 2011, 155, 424-429.	1.2	27
85	Friedreich ataxia presenting as sudden cardiac death in childhood: Clinical, genetic and pathological correlation, with implications for genetic testing and counselling. Neuromuscular Disorders, 2010, 20, 340-342.	0.6	20
86	Neurological complications of cardioâ€facioâ€cutaneous syndrome. Developmental Medicine and Child Neurology, 2007, 49, 894-899.	2.1	106