

Grace Yoon

List of Publications by Year in descending order

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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	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. <i>Npj Genomic Medicine</i> , 2016, 1, .	3.8	295
2	XRCC1 mutation is associated with PARP1 hyperactivation and cerebellar ataxia. <i>Nature</i> , 2017, 541, 87-91.	27.8	209
3	Cockayne syndrome caused by paternally inherited 5Mb deletion of 10q11.2 and a frameshift mutation of ERCC6. <i>European Journal of Medical Genetics</i> , 2011, 54, 272-276.	1.3	136
4	Cognitive and Neurobehavioral Profile in Boys With Duchenne Muscular Dystrophy. <i>Journal of Child Neurology</i> , 2015, 30, 1472-1482.	1.4	134
5	Recessive mutations in POLR1C cause a leukodystrophy by impairing biogenesis of RNA polymerase III. <i>Nature Communications</i> , 2015, 6, 7623.	12.8	127
6	The genetic landscape of familial congenital hydrocephalus. <i>Annals of Neurology</i> , 2017, 81, 890-897.	5.3	108
7	Neurological complications of cardio-facio-cutaneous syndrome. <i>Developmental Medicine and Child Neurology</i> , 2007, 49, 894-899.	2.1	106
8	Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2016, 98, 1038-1046.	6.2	96
9	Exome Sequencing as a Diagnostic Tool for Pediatric Onset Ataxia. <i>Human Mutation</i> , 2014, 35, 45-49.	2.5	91
10	Periodic reanalysis of whole-genome sequencing data enhances the diagnostic advantage over standard clinical genetic testing. <i>European Journal of Human Genetics</i> , 2018, 26, 740-744.	2.8	88
11	Clinical and genetic study of hereditary spastic paraplegia in Canada. <i>Neurology: Genetics</i> , 2017, 3, e122.	1.9	82
12	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. <i>Nature Genetics</i> , 2017, 49, 537-549.	21.4	81
13	CHD2 haploinsufficiency is associated with developmental delay, intellectual disability, epilepsy and neurobehavioural problems. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 9.	3.1	71
14	Lethal Disorder of Mitochondrial Fission Caused by Mutations in DNM1L. <i>Journal of Pediatrics</i> , 2016, 171, 313-316.e2.	1.8	67
15	De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. <i>American Journal of Human Genetics</i> , 2020, 107, 352-363.	6.2	64
16	<i>PMPCA</i> mutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. <i>Brain</i> , 2015, 138, 1505-1517.	7.6	58
17	Frataxin levels in peripheral tissue in Friedreich ataxia. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 831-842.	3.7	55
18	Biallelic Mutations in UNC80 Cause Persistent Hypotonia, Encephalopathy, Growth Retardation, and Severe Intellectual Disability. <i>American Journal of Human Genetics</i> , 2016, 98, 202-209.	6.2	45

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19	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. <i>Nature Genetics</i> , 2021, 53, 1006-1021.	21.4	44
20	Diagnostic Approach to Childhood-Onset Cerebellar Atrophy. <i>Journal of Child Neurology</i> , 2012, 27, 1121-1132.	1.4	42
21	Absence of iron-responsive element-binding protein 2 causes a novel neurodegenerative syndrome. <i>Brain</i> , 2019, 142, 1195-1202.	7.6	38
22	Clinical spectrum of POLR3-related leukodystrophy caused by biallelic <i>POLR1C</i> pathogenic variants. <i>Neurology: Genetics</i> , 2019, 5, e369.	1.9	38
23	Nonsyndromic cerebellar ataxias associated with disorders of DNA single-strand break repair. <i>Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn</i> , 2018, 155, 105-115.	1.8	36
24	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. <i>Scientific Reports</i> , 2016, 6, 28663.	3.3	35
25	Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 86.	2.7	29
26	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. <i>Brain</i> , 2020, 143, 2929-2944.	7.6	29
27	2q23 de novo microdeletion involving the <i>MBD5</i> gene in a patient with developmental delay, postnatal microcephaly and distinct facial features. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 424-429.	1.2	27
28	Bi-allelic mutations of <i>LONP1</i> encoding the mitochondrial LonP1 protease cause pyruvate dehydrogenase deficiency and profound neurodegeneration with progressive cerebellar atrophy. <i>Human Molecular Genetics</i> , 2019, 28, 290-306.	2.9	27
29	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in <i>POLR3A</i> , <i>POLR3B</i> , and <i>POLR1C</i> . <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e660-e674.	3.6	26
30	Neurodegenerative <i>VPS41</i> variants inhibit HOPS function and mTORC1-dependent TFEB/TFE3 regulation. <i>EMBO Molecular Medicine</i> , 2021, 13, e13258.	6.9	26
31	Neuromuscular conditions associated with malignant hyperthermia in paediatric patients: A 25-year retrospective study. <i>Neuromuscular Disorders</i> , 2016, 26, 201-206.	0.6	24
32	Severe neurodegeneration, progressive cerebral volume loss and diffuse hypomyelination associated with a homozygous frameshift mutation in CSTB. <i>European Journal of Human Genetics</i> , 2017, 25, 775-778.	2.8	24
33	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. <i>Nature Communications</i> , 2019, 10, 797.	12.8	24
34	Further Insights into the Allan-Herndon-Dudley Syndrome: Clinical and Functional Characterization of a Novel MCT8 Mutation. <i>PLoS ONE</i> , 2015, 10, e0139343.	2.5	23
35	Congenital myopathy with "corona" fibres, selective muscle atrophy, and craniosynostosis associated with novel recessive mutations in SCN4A. <i>Neuromuscular Disorders</i> , 2017, 27, 574-580.	0.6	23
36	Massive CAG Repeat Expansion and Somatic Instability in Maternally Transmitted Infantile Spinocerebellar Ataxia Type 7. <i>JAMA Neurology</i> , 2015, 72, 219.	9.0	22

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37	A novel KCNA1 mutation in a family with episodic ataxia and malignant hyperthermia. <i>Neurogenetics</i> , 2016, 17, 245-249.	1.4	21
38	Friedreich ataxia presenting as sudden cardiac death in childhood: Clinical, genetic and pathological correlation, with implications for genetic testing and counselling. <i>Neuromuscular Disorders</i> , 2010, 20, 340-342.	0.6	20
39	De novo variants in POLR3B cause ataxia, spasticity, and demyelinating neuropathy. <i>American Journal of Human Genetics</i> , 2021, 108, 186-193.	6.2	19
40	Prevalence of inherited neurotransmitter disorders in patients with movement disorders and epilepsy: a retrospective cohort study. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 12.	2.7	18
41	Health related quality of life in Friedreich Ataxia in a large heterogeneous cohort. <i>Journal of the Neurological Sciences</i> , 2020, 410, 116642.	0.6	18
42	Mitochondrial mimicry of multiple system atrophy of the cerebellar subtype. <i>Movement Disorders</i> , 2011, 26, 753-755.	3.9	17
43	The Pediatric Cerebellum in Inherited Neurodegenerative Disorders. <i>Neuroimaging Clinics of North America</i> , 2016, 26, 373-416.	1.0	17
44	POLR3A variants in hereditary spastic paraplegia and ataxia. <i>Brain</i> , 2018, 141, e1-e1.	7.6	17
45	International retrospective natural history study of <i>LMNA</i>-related congenital muscular dystrophy. <i>Brain Communications</i> , 2021, 3, fcab075.	3.3	17
46	Impact of diabetes in the Friedreich ataxia clinical outcome measures study. <i>Annals of Clinical and Translational Neurology</i> , 2017, 4, 622-631.	3.7	16
47	Assessing non-Mendelian inheritance in inherited axonopathies. <i>Genetics in Medicine</i> , 2020, 22, 2114-2119.	2.4	15
48	Homozygous pathogenic variant in <i>BRAT1</i> associated with nonprogressive cerebellar ataxia. <i>Neurology: Genetics</i> , 2019, 5, e359.	1.9	13
49	Heterozygous Variants in KDM4B Lead to Global Developmental Delay and Neuroanatomical Defects. <i>American Journal of Human Genetics</i> , 2020, 107, 1170-1177.	6.2	13
50	High Frequency of Pathogenic Rearrangements in <i>SPG11</i> and Extensive Contribution of Mutational Hotspots and Founder Alleles. <i>Human Mutation</i> , 2016, 37, 703-709.	2.5	12
51	Cardiac Transplantation in Friedreich Ataxia. <i>Journal of Child Neurology</i> , 2012, 27, 1193-1196.	1.4	11
52	Triple A syndrome presenting as complicated hereditary spastic paraplegia. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 1134-1139.	1.2	11
53	Evidence for Non-Mendelian Inheritance in Spastic Paraplegia 7. <i>Movement Disorders</i> , 2021, 36, 1664-1675.	3.9	11
54	Scoliosis in Friedreich's ataxia: longitudinal characterization in a large heterogeneous cohort. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1239-1250.	3.7	11

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55	<i>De Novo</i> variants in <i>EEF2</i> cause a neurodevelopmental disorder with benign external hydrocephalus. <i>Human Molecular Genetics</i> , 2021, 29, 3892-3899.	2.9	11
56	Interferon-Stimulated Gene Expression as a Preferred Biomarker for Disease Activity in Aicardi's Goutières Syndrome. <i>Journal of Interferon and Cytokine Research</i> , 2017, 37, 147-152.	1.2	8
57	Noncompaction cardiomyopathy in an infant with Walker-Warburg syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3082-3086.	1.2	8
58	Rationale for dopa-responsive <i>CTNNB1</i> catenin deficient dystonia. <i>Movement Disorders</i> , 2018, 33, 656-657.	3.9	8
59	Hypolipidaemia among patients with PMM2-CDG is associated with low circulating PCSK9 levels: a case report followed by observational and experimental studies. <i>Journal of Medical Genetics</i> , 2020, 57, 11-17.	3.2	8
60	The Clinician-reported Genetic testing Utility InDEx (C-GUIDE): Preliminary evidence of validity and reliability. <i>Genetics in Medicine</i> , 2022, 24, 430-438.	2.4	8
61	Severe cystic degeneration and intractable seizures in a newborn with molybdenum cofactor deficiency type B. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 18, 11-13.	1.1	7
62	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. <i>Movement Disorders Clinical Practice</i> , 2020, 7, 940-949.	1.5	7
63	Pearls & Oysters: Fatal brain edema is a rare complication of severe CACNA1A-related disorder. <i>Neurology</i> , 2020, 94, 631-634.	1.1	7
64	An integrated modelling methodology for estimating global incidence and prevalence of hereditary spastic paraplegia subtypes SPG4, SPG7, SPG11, and SPG15. <i>BMC Neurology</i> , 2022, 22, 115.	1.8	7
65	Genetic, structural and clinical analysis of spastic paraplegia 4. <i>Parkinsonism and Related Disorders</i> , 2022, 98, 62-69.	2.2	7
66	Mosaic microdeletion of 17p11.2 <p12 17q22q24="" <i="" a="" and="" duplication="" girl="" in="" neuropathy.="" of="" peripheral="" phenotype="" smith-magenis="" with="">American Journal of Medical Genetics, Part A, 2014, 164, 748-752.</p12>	1.2	6
67	Comorbid Medical Conditions in Friedreich Ataxia. <i>Journal of Child Neurology</i> , 2016, 31, 1161-1165.	1.4	6
68	Subacute demyelinating peripheral neuropathy as a novel presentation of late infantile metachromatic leukodystrophy. <i>Muscle and Nerve</i> , 2017, 56, E41-E44.	2.2	5
69	Impact of Mobility Device Use on Quality of Life in Children With Friedreich Ataxia. <i>Journal of Child Neurology</i> , 2018, 33, 397-404.	1.4	5
70	<i>GCH1</i> mutations in hereditary spastic paraplegia. <i>Clinical Genetics</i> , 2021, 100, 51-58.	2.0	5
71	De novo pathogenic variant in <i>SETX</i> causes a rapidly progressive neurodegenerative disorder of early childhood-onset with severe axonal polyneuropathy. <i>Acta Neuropathologica Communications</i> , 2021, 9, 194.	5.2	5
72	Complete <i>APTX</i> deletion in a patient with ataxia with oculomotor apraxia type 1. <i>BMC Medical Genetics</i> , 2015, 16, 61.	2.1	4

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73	Prospective study of activities of daily living outcomes in children with cerebellar atrophy. <i>Developmental Medicine and Child Neurology</i> , 2014, 56, 460-467.	2.1	3
74	“Both Sides of the Wheelchair” The Views of Individuals with, and Parents of Individuals with Friedreich Ataxia Regarding Pre-symptomatic Testing of Minors. <i>Journal of Genetic Counseling</i> , 2015, 24, 732-743.	1.6	3
75	Reply: Autosomal recessive cerebellar ataxia caused by a homozygous mutation in <i>PMP22</i> . <i>Brain</i> , 2016, 139, e20-e20.	7.6	3
76	Reply: IREB2-associated neurodegeneration. <i>Brain</i> , 2019, 142, e41-e41.	7.6	3
77	FLVCR1-related disease as a rare cause of retinitis pigmentosa and hereditary sensory autonomic neuropathy. <i>European Journal of Medical Genetics</i> , 2020, 63, 104037.	1.3	3
78	Triplication of 16p12.1p12.3 associated with developmental and growth delay and distinctive facial features. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 712-716.	1.2	2
79	Andermann Syndrome in a Pakistani Family Caused by a Novel Mutation in SLC12A6. <i>Journal of Pediatric Neurology</i> , 2017, 15, 090-094.	0.2	2
80	X-linked Joubert syndrome: Neuroimaging and clinical features associated with a novel mutation in oral-facial-digital syndrome type 1. <i>Journal of Pediatric Neurology</i> , 2015, 12, 157-160.	0.2	1
81	Complex genomic rearrangement in <i>SPG11</i> due to a DNA replication-based mechanism. <i>Movement Disorders</i> , 2017, 32, 1792-1794.	3.9	1
82	Heterozygous De Novo <i>KPNA3</i> Mutations Cause Complex Hereditary Spastic Paraplegia. <i>Annals of Neurology</i> , 2022, 91, 730-732.	5.3	1
83	<i>FXR1</i> -related congenital myopathy: expansion of the clinical and genetic spectrum. <i>Journal of Medical Genetics</i> , 2022, 59, 1069-1074.	3.2	1
84	<i>KCNA2</i> mutations are rare in hereditary spastic paraplegia. <i>Annals of Neurology</i> , 2017, 81, 325-326.	5.3	0
85	Response to phenotypic heterogeneity of POMT2 variants. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 746-747.	1.2	0
86	Clinical profile and multidisciplinary needs of patients with neuromuscular disorders transitioning from paediatric to adult care. <i>Neuromuscular Disorders</i> , 2022, 32, 206-212.	0.6	0