

Jolanda H Schieving

List of Publications by Year in descending order

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Version: 2024-02-01

15
papers

972
citations

687363

13
h-index

996975

15
g-index

15
all docs

15
docs citations

15
times ranked

2847
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	6.2	230
2	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. <i>Genetics in Medicine</i> , 2017, 19, 1055-1063.	2.4	220
3	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. <i>Nature Genetics</i> , 2016, 48, 877-887.	21.4	67
4	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 716-724.	6.2	66
5	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. <i>European Journal of Human Genetics</i> , 2020, 28, 40-49.	2.8	65
6	Primary Melanoma of the CNS in Children Is Driven by Congenital Expression of Oncogenic <i>NRAS</i> in Melanocytes. <i>Cancer Discovery</i> , 2013, 3, 458-469.	9.4	61
7	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	6.2	59
8	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. <i>Journal of Medical Genetics</i> , 2018, 55, 104-113.	3.2	59
9	De Novo Mutations Affecting the Catalytic C β Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2019, 104, 139-156.	6.2	39
10	The Genetic Homogeneity of CAPOS Syndrome: Four New Patients With the c.2452G>A (p.Glu818Lys) Mutation in the ATP1A3 Gene. <i>Pediatric Neurology</i> , 2016, 59, 71-75.e1.	2.1	35
11	Acute toxicity profile of craniospinal irradiation with intensity-modulated radiation therapy in children with medulloblastoma: A prospective analysis. <i>Radiation Oncology</i> , 2015, 10, 241.	2.7	24
12	De novo SPAST mutations may cause a complex SPG4 phenotype. <i>Brain</i> , 2019, 142, e31-e31.	7.6	21
13	Reanalysis of exome negative patients with rare disease: a pragmatic workflow for diagnostic applications. <i>Genome Medicine</i> , 2022, 14, .	8.2	17
14	Red flags for early recognition of adult patients with PTEN Hamartoma Tumour Syndrome. <i>European Journal of Medical Genetics</i> , 2021, 64, 104364.	1.3	7
15	Social competence in newly diagnosed pediatric brain tumor patients. <i>Pediatric Hematology and Oncology</i> , 2020, 37, 41-57.	0.8	2