Jolanda H Schieving

List of Publications by Year
in descending order

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