Helen Stewart

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2967696/publications.pdf

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9 papers	506 citations	1307594 7 h-index	1474206 9 g-index
9 all docs	9 docs citations	9 times ranked	1505 citing authors

#	Article	IF	Citations
1	Clinical whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis. Human Molecular Genetics, 2014, 23, 3200-3211.	2.9	222
2	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	6.2	171
3	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. Neuropediatrics, 2014, 45, 175-182.	0.6	41
4	Homozygous mutations in the SCN1A gene associated with genetic epilepsy with febrile seizures plus and Dravet syndrome in 2 families. European Journal of Paediatric Neurology, 2015, 19, 484-488.	1.6	21
5	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	2.8	17
6	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
7	Expanding the phenotype of Wiedemannâ€Steiner syndrome: Craniovertebral junction anomalies. American Journal of Medical Genetics, Part A, 2020, 182, 2877-2886.	1.2	9
8	Recurrent <scp>KCNT2</scp> missense variants affecting p.Arg190 result in a recognizable phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3083-3091.	1.2	7
9	Dissection of contiguous gene effects for deletions around ERF on chromosome 19. Human Mutation, 2021, 42, 811-817.	2.5	2