Lynn Pais

List of Publications by Year
in descending order

9 papers	135 citations	1307594 7 h-index	1474206 9 g-index
10 all docs	10 docs citations	10 times ranked	209 citing authors

#	Article	IF	Citations
1	Recessive variants in COL25A1 gene as novel cause of arthrogryposis multiplex congenita with ocular congenital cranial dysinnervation disorder. Human Mutation, 2022, 43, 487-498.	2.5	8
2	seqr : A webâ€based analysis and collaboration tool for rare disease genomics. Human Mutation, 2022, , .	2.5	31
3	Monoallelic and biallelic variants in LEF1 are associated with a new syndrome combining ectodermal dysplasia and limb malformations caused by altered WNT signaling. Genetics in Medicine, 2022, 24, 1708-1721.	2.4	4
4	Diagnostic capabilities of nanopore longâ€read sequencing in muscular dystrophy. Annals of Clinical and Translational Neurology, 2022, 9, 1302-1309.	3.7	10
5	Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014.	1.7	10
6	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
7	Biallelic loss-of-function variants in WDR11 are associated with microcephaly and intellectual disability. European Journal of Human Genetics, 2021, 29, 1663-1668.	2.8	7
8	A mutation affecting laminin alpha 5 polymerisation gives rise to a syndromic developmental disorder. Development (Cambridge), 2020, 147, .	2.5	28
9	Bi-allelic Variants in DYNC1I2 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. American Journal of Human Genetics, 2019, 104, 1073-1087.	6.2	19