

# Lynn Pais

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9722338/publications.pdf>

Version: 2024-02-01

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	<i>seqr</i> : A web-based analysis and collaboration tool for rare disease genomics. Human Mutation, 2022, , .	2.5	31
2	A mutation affecting laminin alpha 5 polymerisation gives rise to a syndromic developmental disorder. Development (Cambridge), 2020, 147, .	2.5	28
3	Bi-allelic Variants in DYNC112 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. American Journal of Human Genetics, 2019, 104, 1073-1087.	6.2	19
4	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
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	Diagnostic capabilities of nanopore long-read sequencing in muscular dystrophy. Annals of Clinical and Translational Neurology, 2022, 9, 1302-1309.	3.7	10
7	Recessive variants in <i>COL25A1</i> gene as novel cause of arthrogryposis multiplex congenita with ocular congenital cranial dysinnervation disorder. Human Mutation, 2022, 43, 487-498.	2.5	8
8	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
9	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