

Stella A De Man

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4260836/publications.pdf>

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3
papers

31
citations

2258059

3
h-index

2550090

3
g-index

3
all docs

3
docs citations

3
times ranked

63
citing authors

#	ARTICLE	IF	CITATIONS
1	De novo truncating <i>NOVA2</i> variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes. <i>Human Mutation</i> , 2022, 43, 1299-1313.	2.5	6
2	Missense and truncating variants in <i>CHD5</i> in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , 2021, 140, 1109-1120.	3.8	18
3	Molecular analysis of the erythroid phenotype of a patient with <i>BCL11A</i> haploinsufficiency. <i>Blood Advances</i> , 2021, 5, 2339-2349.	5.2	7