

Elke de Boer

List of Publications by Year in descending order

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Version: 2024-02-01

8
papers

505
citations

1307594

7
h-index

1474206

9
g-index

11
all docs

11
docs citations

11
times ranked

1711
citing authors

#	ARTICLE	IF	CITATIONS
1	Genome-wide variant calling in reanalysis of exome sequencing data uncovered a pathogenic TUBB3 variant. <i>European Journal of Medical Genetics</i> , 2022, 65, 104402.	1.3	2
2	Missense variants in ANKRD11 cause KBG syndrome by impairment of stability or transcriptional activity of the encoded protein. <i>Genetics in Medicine</i> , 2022, 24, 2051-2064.	2.4	12
3	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 346-356.	6.2	30
4	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismutation of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021, 108, 502-516.	6.2	48
5	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. <i>European Journal of Human Genetics</i> , 2021, 29, 1359-1368.	2.8	7
6	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. <i>American Journal of Human Genetics</i> , 2021, 108, 2112-2129.	6.2	23
7	Evidence for 28 genetic disorders discovered by combining healthcare and research data. <i>Nature</i> , 2020, 586, 757-762.	27.8	343
8	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. <i>American Journal of Human Genetics</i> , 2020, 107, 164-172.	6.2	37