Elise Fiala

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3174102/publications.pdf

Version: 2024-02-01

7 papers	288 citations	1478505 6 h-index	1474206 9 g-index
9 all docs	9 docs citations	9 times ranked	978 citing authors

#	Article	IF	Citations
1	Reticular dysgenesis caused by an intronic pathogenic variant in AK2. Journal of Physical Education and Sports Management, 2020, 6, a005017.	1.2	4
2	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782.	2.8	27
3	11p15.5 epimutations in children with Wilms tumor and hepatoblastoma detected in peripheral blood. Cancer, 2020, 126, 3114-3121.	4.1	23
4	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
5	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
6	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048.	2.4	85
7	CEDNIK. Child Neurology Open, 2017, 4, 2329048X1773321.	1.1	16