FranÃ§ois Lecoquierre

List of Publications by Year
in descending order

12 papers	134 citations	1307594 7 h-index	1372567 10 g-index
13 all docs	13 docs citations	13 times ranked	248 citing authors

#	Article	IF	Citations
1	Detection of copy-number variations from NGS data using read depth information: a diagnostic performance evaluation. European Journal of Human Genetics, 2021, 29, 99-109.	2.8	23
2	VariantÂrecurrence in neurodevelopmental disorders: the use of publicly available genomic data identifies clinically relevant pathogenic missense variants. Genetics in Medicine, 2019, 21, 2504-2511.	2.4	21
3	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. Human Genetics, 2022, 141, 65-80.	3.8	14
4	De novo variants in <scp>TCF7L2</scp> are associated with a syndromic neurodevelopmental disorder. American Journal of Medical Genetics, Part A, 2021, 185, 2384-2390.	1.2	13
5	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	3.2	13
6	Optimization of the diagnosis of inherited colorectal cancer using NGS and capture of exonic and intronic sequences of panel genes. European Journal of Human Genetics, 2018, 26, 1597-1602.	2.8	12
7	Confirmation and further delineation of the SMG9â€deficiency syndrome, a rare and severe developmental disorder. American Journal of Medical Genetics, Part A, 2019, 179, 2257-2262.	1.2	9
8	Haploinsufficiency of the Primary Familial Brain Calcification Gene <scp>SLC20A2</scp> Mediated by Disruption of a Regulatory Element. Movement Disorders, 2020, 35, 1336-1345.	3.9	9
9	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. Journal of Medical Genetics, 2020, 57, 466-474.	3.2	7
10	uORFâ€introducing variants in the 5â€²UTR of the NIPBL gene as a cause of Cornelia de Lange syndrome. Human Mutation, 2022, 43, 1239-1248.	2.5	6
11	Heterozygous variants in PRPF8 are associated with neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2022, 188, 2750-2759.	1.2	4
12	Patients with 10q22.3q23.1 recurrent deletion syndrome are at risk for juvenile polyposis. European Journal of Medical Genetics, 2020, 63, 103773.	1.3	3