Martin Chevarin

List of Publications by Year
in descending order

11 papers	280 citations	1684188 5 h-index	1281871 11 g-index
11 all docs	11 docs citations	11 times ranked	899 citing authors

#	Article	IF	Citations
1	Clinical whole-exome sequencing for the diagnosis of rare disorders with congenital anomalies and/or intellectual disability: substantial interest of prospective annual reanalysis. Genetics in Medicine, 2018, 20, 645-654.	2.4	146
2	Increased diagnostic and new genes identification outcome using research reanalysis of singleton exome sequencing. European Journal of Human Genetics, 2019, 27, 1519-1531.	2.8	43
3	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
4	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
5	Secondary actionable findings identified by exome sequencing: expected impact on the organisation of care from the study of 700 consecutive tests. European Journal of Human Genetics, 2019, 27, 1197-1214.	2.8	18
6	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911.	2.4	9
7	Neutralization of HSF1 in cells from PIK3CA-related overgrowth spectrum patients blocks abnormal proliferation. Biochemical and Biophysical Research Communications, 2020, 530, 520-526.	2.1	5
8	Interest of exome sequencing trioâ€like strategy based on pooled parental DNA for diagnosis and translational research in rare diseases. Molecular Genetics & Genomic Medicine, 2021, 9, e1836.	1.2	5
9	Primrose syndrome: a phenotypic comparison of patients with a ZBTB20 missense variant versus a 3q13.31 microdeletion including ZBTB20. European Journal of Human Genetics, 2020, 28, 1044-1055.	2.8	4
10	Mosaic NEK9 mutation, fibrous dysplasia and premature puberty in naevus comedonicus syndrome. British Journal of Dermatology, 2021, , .	1.5	1
11	Same performance of exome sequencing before and after fetal autopsy for congenital abnormalities: toward a paradigm shift in prenatal diagnosis?. European Journal of Human Genetics, 2022, , .	2.8	1