Thibaud Jouan

List of Publications by Year
in descending order

5 papers	268 citations	1684188 5 h-index	2053705 5 g-index
5 all docs	5 docs citations	5 times ranked	983 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 SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaidesâ€“Baraitser syndrome. Genetics in Medicine, 2020, 22, 1838-1850.	2.4	31
4	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La RÃ©union Island, in patients with Fryns syndrome. European Journal of Human Genetics, 2018, 26, 340-349.	2.8	27
5	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