

# Thibaud Jouan

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6894863/publications.pdf>

Version: 2024-02-01

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	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides's Baraitser syndrome. <i>Genetics in Medicine</i> , 2020, 22, 1838-1850.	2.4	31
2	Increased diagnostic and new genes identification outcome using research reanalysis of singleton exome sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 1519-1531.	2.8	43
3	Variant recurrence in neurodevelopmental disorders: the use of publicly available genomic data identifies clinically relevant pathogenic missense variants. <i>Genetics in Medicine</i> , 2019, 21, 2504-2511.	2.4	21
4	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La Réunion Island, in patients with Fryns syndrome. <i>European Journal of Human Genetics</i> , 2018, 26, 340-349.	2.8	27
5	Clinical whole-exome sequencing for the diagnosis of rare disorders with congenital anomalies and/or intellectual disability: substantial interest of prospective annual reanalysis. <i>Genetics in Medicine</i> , 2018, 20, 645-654.	2.4	146