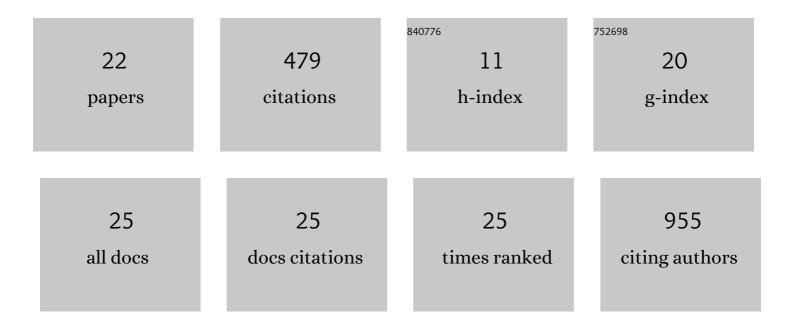
Aurelien Trimouille

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/4051090/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	Oculo-auriculo-vertebral spectrum: new genes and literature review on a complex disease. Journal of Medical Genetics, 2022, 59, 417-427.	3.2	20
2	<i>De novo</i> coding variants in the <i>AGO1</i> gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	3.2	13
3	A recurrent missense variant in EYA3 gene is associated with oculo-auriculo-vertebral spectrum. Human Genetics, 2021, 140, 933-944.	3.8	14
4	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	2.8	49
5	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. European Journal of Human Genetics, 2021, 29, 1359-1368.	2.8	7
6	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	2.8	34
7	Implication of folate deficiency in CYP2U1 loss of function. Journal of Experimental Medicine, 2021, 218, .	8.5	13
8	Hereditary Mucoepithelial Dysplasia Results from Heterozygous Variants at p.Arg557 Mutational Hotspot in SREBF1, Encoding a Transcription Factor Involved in Cholesterol Homeostasis. Journal of Investigative Dermatology, 2020, 140, 1289-1292.e2.	0.7	12
9	Description of a family with Xâ€linked oculoâ€auriculoâ€vertebral spectrum associated with polyalanine tract expansion in <scp><i>ZIC3</i></scp> . Clinical Genetics, 2020, 98, 384-389.	2.0	11
10	Functional and genetic analyses of <i>ZYG11B</i> provide evidences for its involvement in OAVS. Molecular Genetics & Genomic Medicine, 2020, 8, e1375.	1.2	21
11	Rare variants in the GABA _A receptor subunit ε identified in patients with a wide spectrum of epileptic phenotypes. Molecular Genetics & Genomic Medicine, 2020, 8, e1388.	1.2	8
12	BLOC1S5 pathogenic variants cause a new type of Hermansky–Pudlak syndrome. Genetics in Medicine, 2020, 22, 1613-1622.	2.4	44
13	Major intra-familial phenotypic heterogeneity and incomplete penetrance due to a CACNA1A pathogenic variant. European Journal of Medical Genetics, 2019, 62, 103530.	1.3	34
14	PADDAS syndrome associated with hair dysplasia caused by a de novo missense variant of PUM1. American Journal of Medical Genetics, Part A, 2019, 179, 1030-1033.	1.2	6
15	Deletion in 2q35 excluding the IHH gene leads to fetal severe limb anomalies and suggests a disruption of chromatin architecture. European Journal of Human Genetics, 2019, 27, 384-388.	2.8	3
16	An in-frame deletion in BICD2 associated with a non-progressive form of SMALED. Clinical Neurology and Neurosurgery, 2018, 166, 1-3.	1.4	6
17	Phenotype and genotype of 87 patients with Mowat–Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
18	Further delineation of the phenotype caused by biallelic variants in the <i><scp>WDR4</scp></i> gene. Clinical Genetics, 2018, 93, 374-377.	2.0	33

#	Article	IF	CITATIONS
19	19p13 microduplications encompassing NFIX are responsible for intellectual disability, short stature and small head circumference. European Journal of Human Genetics, 2018, 26, 85-93.	2.8	7
20	<i>IL11RAâ€</i> related Crouzonâ€like autosomal recessive craniosynostosis in 10 new patients: Resemblances and differences. Clinical Genetics, 2018, 94, 373-380.	2.0	29
21	Deletion of the transcription factor <i><scp>SOX4</scp></i> is implicated in syndromic nephroblastoma. Clinical Genetics, 2017, 92, 449-450.	2.0	2
22	Neuroimaging findings in Mowat–Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45