Aurelien Trimouille

List of Publications by Year
in descending order

22	479	840776 11	752698 20
papers	citations	h-index	g-index

25	25	25	955
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
2	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
3	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45
4	BLOC1S5 pathogenic variants cause a new type of Hermanskyâ€“Pudlak syndrome. Genetics in Medicine, 2020, 22, 1613-1622.	2.4	44
5	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
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	Further delineation of the phenotype caused by biallelic variants in the <scp>WDR4</scp> gene. Clinical Genetics, 2018, 93, 374-377.	2.0	33
8	IL11RAâ€related Crouzonâ€like autosomal recessive craniosynostosis in 10 new patients: Resemblances and differences. Clinical Genetics, 2018, 94, 373-380.	2.0	29
9	Functional and genetic analyses of ZYG11B provide evidences for its involvement in OAVS. Molecular Genetics & Genomic Medicine, 2020, 8, e1375.	1.2	21
10	Oculo-auriculo-vertebral spectrum: new genes and literature review on a complex disease. Journal of Medical Genetics, 2022, 59, 417-427.	3.2	20
11	A recurrent missense variant in EYA3 gene is associated with oculo-auriculo-vertebral spectrum. Human Genetics, 2021, 140, 933-944.	3.8	14
12	Implication of folate deficiency in CYP2U1 loss of function. Journal of Experimental Medicine, 2021, 218, .	8.5	13
13	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
14	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
15	Description of a family with Xâ€linked oculoâ€auriculoâ€vertebral spectrum associated with polyalanine tract expansion in <scp>ZIC3</scp>. Clinical Genetics, 2020, 98, 384-389.	2.0	11
16	Rare variants in the GABA_A receptor subunit Îµ identified in patients with a wide spectrum of epileptic phenotypes. Molecular Genetics & Genomic Medicine, 2020, 8, e1388.	1.2	8
17	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
18	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

#	Article	IF	Citations
19	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
20	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
21	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
22	Deletion of the transcription factor <scp>SOX4</scp> is implicated in syndromic nephroblastoma. Clinical Genetics, 2017, 92, 449-450.	2.0	2