Marie Vincent

List of Publications by Year
in descending order

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43 papers	1,803 citations	304743 22 h-index	302126 39 g-index
45 all docs	45 docs citations	45 times ranked	4083 citing authors

#	Article	IF	Citations
1	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. European Journal of Human Genetics, 2022, 30, 567-576.	2.8	12
2	Treatment of two infants with PIK3CA-related overgrowth spectrum by alpelisib. Journal of Experimental Medicine, 2022, 219, .	8.5	27
3	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
4	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	2.8	17
5	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
6	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. Genetics in Medicine, 2020, 22, 547-556.	2.4	63
7	Evolutionary conserved NSL complex/BRD4 axis controls transcription activation via histone acetylation. Nature Communications, 2020, 11, 2243.	12.8	21
8	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
9	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. Human Genetics, 2019, 138, 1051-1069.	3.8	35
10	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
11	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. Genetics in Medicine, 2019, 21, 2025-2035.	2.4	40
12	Clinical, Histopathological, and Molecular Diagnostics in Lethal Lung Developmental Disorders. American Journal of Respiratory and Critical Care Medicine, 2019, 200, 1093-1101.	5.6	47
13	Searching for secondary findings: considering actionability and preserving the right not to know. European Journal of Human Genetics, 2019, 27, 1481-1484.	2.8	13
14	Further corroboration of distinct functional features in SCN2A variants causing intellectual disability or epileptic phenotypes. Molecular Medicine, 2019, 25, 6.	4.4	42
15	Autosomal recessive Treacher Collins syndrome due to POLR1C mutations: Report of a new family and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1390-1394.	1.2	19
16	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	2.4	127
17	A de novo 2q37.2 deletion encompassing AGAP1 and SH3BP4 in a patient with autism and intellectual disability. European Journal of Medical Genetics, 2019, 62, 103586.	1.3	12
18	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90

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19	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
20	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
21	<scp>R</scp>ubinsteinâ€<scp>T</scp>aybi Syndrome in a Fetus: Contribution of 2â€•and 3â€Dimensional Ultrasonography. Journal of Ultrasound in Medicine, 2018, 37, 531-534.	1.7	4
22	Delineating FOXG1 syndrome. Neurology: Genetics, 2018, 4, e281.	1.9	51
23	Haploinsufficiency of CUX1 Causes Nonsyndromic Global Developmental Delay With Possible Catchâ€up Development. Annals of Neurology, 2018, 84, 200-207.	5.3	23
24	New splicing pathogenic variant in EBP causing extreme familial variability of Conradiâ€“HÃ¼nermannâ€“Happle Syndrome. European Journal of Human Genetics, 2018, 26, 1784-1790.	2.8	7
25	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	2.4	90
26	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
27	Sex chromosome aneuploidies and copy-number variants: a further explanation for neurodevelopmental prognosis variability?. European Journal of Human Genetics, 2017, 25, 930-934.	2.8	19
28	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	3.8	66
29	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
30	Three patients presenting with severe macrosomia and congenital hypertrophic cardiomyopathy: a case series. Journal of Medical Case Reports, 2017, 11, 78.	0.8	6
31	Two novel variants in CNTNAP1 in two siblings presenting with congenital hypotonia and hypomyelinating neuropathy. European Journal of Human Genetics, 2017, 25, 150-152.	2.8	13
32	Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamondâ€“like features. Journal of Clinical Investigation, 2017, 127, 4090-4103.	8.2	126
33	Treacher Collins Syndrome. Plastic and Reconstructive Surgery, 2016, 138, 374e-376e.	1.4	2
34	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. Nature Genetics, 2016, 48, 877-887.	21.4	67
35	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	6.2	45
36	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	2.5	40

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37	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	2.4	125
38	Patients with isolated oligo/hypodontia caused by RUNX2 duplication. American Journal of Medical Genetics, Part A, 2015, 167, 1386-1390.	1.2	10
39	Isolated and syndromic brachydactylies: Diagnostic value of hand X-rays. Diagnostic and Interventional Imaging, 2015, 96, 443-448.	3.2	10
40	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	3.2	72
41	Large deletions encompassing the TCOF1 and CAMK2A genes are responsible for Treacher Collins syndrome with intellectual disability. European Journal of Human Genetics, 2014, 22, 52-56.	2.8	22
42	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. Genetics in Medicine, 2014, 16, 720-724.	2.4	63
43	Duplication 8q12: confirmation of a novel recognizable phenotype with duane retraction syndrome and developmental delay. European Journal of Human Genetics, 2012, 20, 580-583.	2.8	17