

Marie Vincent

List of Publications by Year in descending order

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43
papers

1,803
citations

304743

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h-index

302126

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45
docs citations

45
times ranked

4083
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	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
3	Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamond-like features. Journal of Clinical Investigation, 2017, 127, 4090-4103.	8.2	126
4	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
5	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
6	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
7	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
8	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	3.2	72
9	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
10	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
11	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
12	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. Genetics in Medicine, 2020, 22, 547-556.	2.4	63
13	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
14	Delineating FOXG1 syndrome. Neurology: Genetics, 2018, 4, e281.	1.9	51
15	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
16	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
17	Further corroboration of distinct functional features in SCN2A variants causing intellectual disability or epileptic phenotypes. Molecular Medicine, 2019, 25, 6.	4.4	42
18	De Novo Truncating Mutations in the Kinetochores-Microtubules Attachment Gene CHAMP1 Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	2.5	40

#	ARTICLE	IF	CITATIONS
19	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. <i>Genetics in Medicine</i> , 2019, 21, 2025-2035.	2.4	40
20	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. <i>Human Genetics</i> , 2019, 138, 1051-1069.	3.8	35
21	Major intra-familial phenotypic heterogeneity and incomplete penetrance due to a CACNA1A pathogenic variant. <i>European Journal of Medical Genetics</i> , 2019, 62, 103530.	1.3	34
22	Treatment of two infants with PIK3CA-related overgrowth spectrum by alpelisib. <i>Journal of Experimental Medicine</i> , 2022, 219, .	8.5	27
23	Haploinsufficiency of <i>CUX1</i> Causes Nonsyndromic Global Developmental Delay With Possible Catch-up Development. <i>Annals of Neurology</i> , 2018, 84, 200-207.	5.3	23
24	Large deletions encompassing the TCOF1 and CAMK2A genes are responsible for Treacher Collins syndrome with intellectual disability. <i>European Journal of Human Genetics</i> , 2014, 22, 52-56.	2.8	22
25	Evolutionary conserved NSL complex/BRD4 axis controls transcription activation via histone acetylation. <i>Nature Communications</i> , 2020, 11, 2243.	12.8	21
26	Sex chromosome aneuploidies and copy-number variants: a further explanation for neurodevelopmental prognosis variability?. <i>European Journal of Human Genetics</i> , 2017, 25, 930-934.	2.8	19
27	Autosomal recessive Treacher Collins syndrome due to <i>POLR1C</i> mutations: Report of a new family and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1390-1394.	1.2	19
28	Duplication 8q12: confirmation of a novel recognizable phenotype with duane retraction syndrome and developmental delay. <i>European Journal of Human Genetics</i> , 2012, 20, 580-583.	2.8	17
29	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. <i>European Journal of Human Genetics</i> , 2021, 29, 625-636.	2.8	17
30	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2022, 109, 601-617.	6.2	16
31	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. <i>American Journal of Human Genetics</i> , 2021, 108, 929-941.	6.2	15
32	Two novel variants in CNTNAP1 in two siblings presenting with congenital hypotonia and hypomyelinating neuropathy. <i>European Journal of Human Genetics</i> , 2017, 25, 150-152.	2.8	13
33	Searching for secondary findings: considering actionability and preserving the right not to know. <i>European Journal of Human Genetics</i> , 2019, 27, 1481-1484.	2.8	13
34	A de novo 2q37.2 deletion encompassing AGAP1 and SH3BP4 in a patient with autism and intellectual disability. <i>European Journal of Medical Genetics</i> , 2019, 62, 103586.	1.3	12
35	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. <i>European Journal of Human Genetics</i> , 2022, 30, 567-576.	2.8	12
36	Patients with isolated oligo/hypodontia caused by RUNX2 duplication. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1386-1390.	1.2	10

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37	Isolated and syndromic brachydactylies: Diagnostic value of hand X-rays. Diagnostic and Interventional Imaging, 2015, 96, 443-448.	3.2	10
38	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
39	New splicing pathogenic variant in EBP causing extreme familial variability of Conradi-Hallermann-Riedel Syndrome. European Journal of Human Genetics, 2018, 26, 1784-1790.	2.8	7
40	Three patients presenting with severe macrosomia and congenital hypertrophic cardiomyopathy: a case series. Journal of Medical Case Reports, 2017, 11, 78.	0.8	6
41	Robinow Syndrome in a Fetus: Contribution of 2D and 3D Dimensional Ultrasonography. Journal of Ultrasound in Medicine, 2018, 37, 531-534.	1.7	4
42	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
43	Treacher Collins Syndrome. Plastic and Reconstructive Surgery, 2016, 138, 374e-376e.	1.4	2