Caroline Michot

List of Publications by Year
in descending order

30 papers	1,267 citations	430874 18 h-index	454955 30 g-index
32 all docs	32 docs citations	32 times ranked	2313 citing authors

#	Article	IF	Citations
1	Cardio-facio-cutaneous and Noonan syndromes due to mutations in the RAS/MAPK signalling pathway: genotype phenotype relationships and overlap with Costello syndrome. Journal of Medical Genetics, 2007, 44, 763-771.	3.2	221
2	Exome Sequencing Identifies PDE4D Mutations as Another Cause of Acrodysostosis. American Journal of Human Genetics, 2012, 90, 740-745.	6.2	115
3	LPIN1 gene mutations: a major cause of severe rhabdomyolysis in early childhood. Human Mutation, 2010, 31, E1564-E1573.	2.5	112
4	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
5	Study of LPIN1, LPIN2 and LPIN3 in rhabdomyolysis and exerciseâ€induced myalgia. Journal of Inherited Metabolic Disease, 2012, 35, 1119-1128.	3.6	75
6	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
7	Sleepâ€disordered breathing and its management in children with achondroplasia. American Journal of Medical Genetics, Part A, 2017, 173, 868-878.	1.2	59
8	Extensive investigation of the IGF2/H19 imprinting control region reveals novel OCT4/SOX2 binding site defects associated with specific methylation patterns in Beckwith-Wiedemann syndrome. Human Molecular Genetics, 2014, 23, 5763-5773.	2.9	58
9	Gainâ€ofâ€function mutations in SMAD4 cause a distinctive repertoire of cardiovascular phenotypes in patients with Myhre syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2617-2631.	1.2	53
10	Fatal Rhabdomyolysis in 2 Children with LPIN1 Mutations. Journal of Pediatrics, 2012, 160, 1052-1054.	1.8	50
11	Combination of lipid metabolism alterations and their sensitivity to inflammatory cytokines in human lipin-1-deficient myoblasts. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 2103-2114.	3.8	50
12	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	2.8	38
13	Chromosomal rearrangements in the 11p15 imprinted region: 17 new 11p15.5 duplications with associated phenotypes and putative functional consequences. Journal of Medical Genetics, 2018, 55, 205-213.	3.2	36
14	Uncommon nucleotide excision repair phenotypes revealed by targeted high-throughput sequencing. Orphanet Journal of Rare Diseases, 2016, 11, 26.	2.7	32
15	Homozygous Lossâ€ofâ€Function Mutations in <scp>CCDC134</scp> Are Responsible for a Severe Form of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2020, 35, 1470-1480.	2.8	29
16	Clinical and Molecular Spectrum of Renal Malformations in Kabuki Syndrome. Journal of Pediatrics, 2013, 163, 742-746.	1.8	27
17	RPL10 mutation segregating in a family with Xâ€linked syndromic Intellectual Disability. American Journal of Medical Genetics, Part A, 2015, 167, 1908-1912.	1.2	27
18	Mutations of TSEN and CASK genes are prevalent in pontocerebellar hypoplasias type 2 and 4. Brain, 2012, 135, e199-e199.	7.6	18

#	Article	IF	Citations
19	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscyphodysplasia. European Journal of Human Genetics, 2018, 26, 1611-1622.	2.8	18
20	TAR syndrome: Clinical and molecular characterization of a cohort of 26 patients and description of novel noncoding variants of RBM8A. Human Mutation, 2020, 41, 1220-1225.	2.5	17
21	Geleophysic and acromicric dysplasias: natural history, genotypeâ€“phenotype correlations, and management guidelines from 38 cases. Genetics in Medicine, 2021, 23, 331-340.	2.4	17
22	Finger creases lend a hand in Kabuki syndrome. European Journal of Medical Genetics, 2013, 56, 556-560.	1.3	11
23	Sleepâ€disordered breathing in children with mucolipidosis. American Journal of Medical Genetics, Part A, 2019, 179, 1196-1204.	1.2	10
24	PAPSS2â€related brachyolmia: Clinical and radiological phenotype in 18 new cases. American Journal of Medical Genetics, Part A, 2019, 179, 1884-1894.	1.2	9
25	Sleepâ€disordered breathing in children with pycnodysostosis. American Journal of Medical Genetics, Part A, 2020, 182, 122-129.	1.2	7
26	A mutation in SLC37A4 causes a dominantly inherited congenital disorder of glycosylation characterized by liver dysfunction. American Journal of Human Genetics, 2021, 108, 1040-1052.	6.2	7
27	Parental mosaicism is a pitfall in preimplantation genetic diagnosis of dominant disorders. European Journal of Human Genetics, 2014, 22, 711-712.	2.8	5
28	A retrospective study on sleepâ€disordered breathing in Morquioâ€A syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2595-2603.	1.2	3
29	Sleepâ€disordered breathing and its management in children with rare skeletal dysplasias. American Journal of Medical Genetics, Part A, 2021, 185, 2108-2118.	1.2	2
30	Enriching UMLS-Based Phenotyping of Rare Diseases Using Deep-Learning: Evaluation on Jeune Syndrome. Studies in Health Technology and Informatics, 2022, , .	0.3	1