

Caroline Michot

List of Publications by Year in descending order

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Version: 2024-02-01

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	Enriching UMLS-Based Phenotyping of Rare Diseases Using Deep-Learning: Evaluation on Jeune Syndrome. <i>Studies in Health Technology and Informatics</i> , 2022, , .	0.3	1
2	Geleophysic and acromicric dysplasias: natural history, genotypeâ€“phenotype correlations, and management guidelines from 38 cases. <i>Genetics in Medicine</i> , 2021, 23, 331-340.	2.4	17
3	Sleepâ€“disordered breathing and its management in children with rare skeletal dysplasias. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2108-2118.	1.2	2
4	A mutation in SLC37A4 causes a dominantly inherited congenital disorder of glycosylation characterized by liver dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 1040-1052.	6.2	7
5	Sleepâ€“disordered breathing in children with pycnodysostosis. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 122-129.	1.2	7
6	Homozygous Lossâ€“ofâ€“Function Mutations in <sc><i>CCDC134</i></sc> Are Responsible for a Severe Form of Osteogenesis Imperfecta. <i>Journal of Bone and Mineral Research</i> , 2020, 35, 1470-1480.	2.8	29
7	TAR syndrome: Clinical and molecular characterization of a cohort of 26 patients and description of novel noncoding variants of <i>RBM8A</i>. <i>Human Mutation</i> , 2020, 41, 1220-1225.	2.5	17
8	<i>PAPSS2</i>-related brachyolmia: Clinical and radiological phenotype in 18 new cases. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1884-1894.	1.2	9
9	Sleepâ€“disordered breathing in children with mucopolipidosis. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1196-1204.	1.2	10
10	Chromosomal rearrangements in the 11p15 imprinted region: 17 new 11p15.5 duplications with associated phenotypes and putative functional consequences. <i>Journal of Medical Genetics</i> , 2018, 55, 205-213.	3.2	36
11	A retrospective study on sleepâ€“disordered breathing in Morquioâ€“A syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2595-2603.	1.2	3
12	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscaphodysplasia. <i>European Journal of Human Genetics</i> , 2018, 26, 1611-1622.	2.8	18
13	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. <i>Genetics in Medicine</i> , 2017, 19, 989-997.	2.4	90
14	Sleepâ€“disordered breathing and its management in children with achondroplasia. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 868-878.	1.2	59
15	Phenotypes and genotypes in individuals with <i>SMC1A</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2108-2125.	1.2	69
16	Gainâ€“ofâ€“function mutations in <i>SMAD4</i> cause a distinctive repertoire of cardiovascular phenotypes in patients with Myhre syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2617-2631.	1.2	53
17	Uncommon nucleotide excision repair phenotypes revealed by targeted high-throughput sequencing. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 26.	2.7	32
18	<i>RPL10</i> mutation segregating in a family with Xâ€“linked syndromic Intellectual Disability. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1908-1912.	1.2	27

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19	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. <i>Human Molecular Genetics</i> , 2014, 23, 5763-5773.	2.9	58
20	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. <i>European Journal of Human Genetics</i> , 2014, 22, 1272-1277.	2.8	38
21	Parental mosaicism is a pitfall in preimplantation genetic diagnosis of dominant disorders. <i>European Journal of Human Genetics</i> , 2014, 22, 711-712.	2.8	5
22	Combination of lipid metabolism alterations and their sensitivity to inflammatory cytokines in human lipin-1-deficient myoblasts. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2013, 1832, 2103-2114.	3.8	50
23	Clinical and Molecular Spectrum of Renal Malformations in Kabuki Syndrome. <i>Journal of Pediatrics</i> , 2013, 163, 742-746.	1.8	27
24	Finger creases lend a hand in Kabuki syndrome. <i>European Journal of Medical Genetics</i> , 2013, 56, 556-560.	1.3	11
25	Mutations of TSEN and CASK genes are prevalent in pontocerebellar hypoplasias type 2 and 4. <i>Brain</i> , 2012, 135, e199-e199.	7.6	18
26	Study of <i>LPIN1</i> , <i>LPIN2</i> and <i>LPIN3</i> in rhabdomyolysis and exercise-induced myalgia. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 1119-1128.	3.6	75
27	Exome Sequencing Identifies PDE4D Mutations as Another Cause of Acrodysostosis. <i>American Journal of Human Genetics</i> , 2012, 90, 740-745.	6.2	115
28	Fatal Rhabdomyolysis in 2 Children with LPIN1 Mutations. <i>Journal of Pediatrics</i> , 2012, 160, 1052-1054.	1.8	50
29	LPIN1 gene mutations: a major cause of severe rhabdomyolysis in early childhood. <i>Human Mutation</i> , 2010, 31, E1564-E1573.	2.5	112
30	Cardio-facio-cutaneous and Noonan syndromes due to mutations in the RAS/MAPK signalling pathway: genotype phenotype relationships and overlap with Costello syndrome. <i>Journal of Medical Genetics</i> , 2007, 44, 763-771.	3.2	221