Sebastien Levesque

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2399079/publications.pdf

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17 papers	332 citations	933447 10 h-index	996975 15 g-index
17 all docs	17 docs citations	17 times ranked	924 citing authors

#	Article	IF	Citations
1	Clinical, neuroradiological, and molecular characterization of patients with atypical Zellweger spectrum disorder caused by PEX16 mutations: a case series. Neurogenetics, 2022, 23, 115-127.	1.4	0
2	Molecular Diagnosis of Pompe Disease in the Genomic Era: Correlation with Acid Alpha-Glucosidase Activity in Dried Blood Spots. Journal of Clinical Medicine, 2021, 10, 3868.	2.4	4
3	FA2H Mutations in a Young Adult Presenting as an Isolated Cognitive Impairment Syndrome. Canadian Journal of Neurological Sciences, 2020, 47, 858-860.	0.5	0
4	Biallelic variants in the small optic lobe calpain CAPN15 are associated with congenital eye anomalies, deafness and other neurodevelopmental deficits. Human Molecular Genetics, 2020, 29, 3054-3063.	2.9	15
5	Molecular diagnosis of muscular diseases in outpatient clinics. Neurology: Genetics, 2020, 6, e408.	1.9	15
6	HSD10 mitochondrial disease: p.Leu122Val variant, mild clinical phenotype, and founder effect in Frenchâ€Canadian patients from Quebec. Molecular Genetics & Genomic Medicine, 2019, 7, e1000.	1.2	8
7	Wide Spectrum of DUOX2 Deficiency: From Life-Threatening Compressive Goiter in Infancy to Lifelong Euthyroidism. Thyroid, 2019, 29, 1018-1022.	4.5	16
8	Combined malonic and methylmalonic aciduria due to ACSF3 mutations: Benign clinical course in an unselected cohort. Journal of Inherited Metabolic Disease, 2019, 42, 107-116.	3.6	23
9	Combined malonic and methylmalonic aciduria due to ACSF3 mutations: benign clinical course in an unselected cohort. Journal of Inherited Metabolic Disease, 2019, 42, 107.	3.6	4
10	Clinical validity of phenotype-driven analysis software PhenoVar as a diagnostic aid for clinical geneticists in the interpretation of whole-exome sequencing data. Genetics in Medicine, 2018, 20, 942-949.	2.4	15
11	Online Module for Carrier Screening in Ashkenazi Jewish Individuals Compared with Inâ€Person Genetics Education: A Randomized Controlled Trial. Journal of Genetic Counseling, 2018, 27, 426-438.	1.6	9
12	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	2.4	94
13	Gain-of-Function Mutations inRARBCause Intellectual Disability with Progressive Motor Impairment. Human Mutation, 2016, 37, 786-793.	2.5	34
14	Methylmalonyl-coA epimerase deficiency: A new case, with an acute metabolic presentation and an intronic splicing mutation in the MCEE gene. Molecular Genetics and Metabolism Reports, 2016, 9, 19-24.	1.1	14
15	Diagnosis of late-onset Pompe disease and other muscle disorders by next-generation sequencing. Orphanet Journal of Rare Diseases, 2016, 11, 8.	2.7	42
16	Spastic Paraparesis and Marked Improvement of Leukoencephalopathy in Aicardiâ€“GoutiÃ¨res Syndrome. Neuropediatrics, 2014, 45, 406-410.	0.6	9
17	A founder mutation in the PEX6 gene is responsible for increased incidence of Zellweger syndrome in a French Canadian population. BMC Medical Genetics, 2012, 13, 72.	2.1	30