

# Sebastien Levesque

## List of Publications by Year in descending order

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

Version: 2024-02-01

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