Isabelle Thiffault

List of Publications by Year
in descending order

67 papers	2,978 citations	201674 27 h-index	189892 50 g-index
71 all docs	71 docs citations	71 times ranked	5913 citing authors

#	Article	IF	Citations
1	Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders. Science Translational Medicine, 2014, 6, 265ra168.	12.4	440
2	Whole-genome sequencing for identification of Mendelian disorders in critically ill infants: a retrospective analysis of diagnostic and clinical findings. Lancet Respiratory Medicine,the, 2015, 3, 377-387.	10.7	322
3	A 26-hour system of highly sensitive whole genome sequencing for emergency management of genetic diseases. Genome Medicine, 2015, 7, 100.	8.2	237
4	The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. Npj Genomic Medicine, 2018, 3, 6.	3.8	156
5	Recessive mutations in POLR1C cause a leukodystrophy by impairing biogenesis of RNA polymerase III. Nature Communications, 2015, 6, 7623.	12.8	127
6	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. Frontiers in Immunology, 2016, 7, 466.	4.8	80
7	A novel epileptic encephalopathy mutation in KCNB1 disrupts Kv2.1 ion selectivity, expression, and localization. Journal of General Physiology, 2015, 146, 399-410.	1.9	79
8	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	9.0	79
9	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	6.2	66
10	The 3â€² addition of CCA to mitochondrial tRNASer(AGY) is specifically impaired in patients with mutations in the tRNA nucleotidyl transferase TRNT1. Human Molecular Genetics, 2015, 24, 2841-2847.	2.9	65
11	MMP21 is mutated in human heterotaxy and is required for normal left-right asymmetry in vertebrates. Nature Genetics, 2015, 47, 1260-1263.	21.4	65
12	CLPB Variants Associated with Autosomal-Recessive Mitochondrial Disorder with Cataract, Neutropenia, Epilepsy, and Methylglutaconic Aciduria. American Journal of Human Genetics, 2015, 96, 258-265.	6.2	58
13	Bi-allelic Mutations in EPRS, Encoding the Glutamyl-Prolyl-Aminoacyl-tRNA Synthetase, Cause a Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2018, 102, 676-684.	6.2	58
14	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
15	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. Science Advances, 2020, 6, eaax0021.	10.3	56
16	Spectrum of K_V2.1 Dysfunction in KCNB1â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2019, 86, 899-912.	5.3	52
17	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
18	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50

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19	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
20	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	6.2	42
21	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. Journal of Clinical Investigation, 2020, 130, 1431-1445.	8.2	40
22	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase (TRIT1) gene. Human Mutation, 2017, 38, 511-516.	2.5	39
23	PCDH19â€related epileptic encephalopathy in a male mosaic for a truncating variant. American Journal of Medical Genetics, Part A, 2016, 170, 1585-1589.	1.2	37
24	Neonatal progeriod syndrome associated with biallelic truncating variants in POLR3A. American Journal of Medical Genetics, Part A, 2016, 170, 3343-3346.	1.2	37
25	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
26	Genomic answers for children: Dynamic analyses of >1000 pediatric rare disease genomes. Genetics in Medicine, 2022, 24, 1336-1348.	2.4	37
27	Clinical genome sequencing in an unbiased pediatric cohort. Genetics in Medicine, 2019, 21, 303-310.	2.4	36
28	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. American Journal of Human Genetics, 2019, 104, 758-766.	6.2	34
29	BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. American Journal of Human Genetics, 2020, 107, 1096-1112.	6.2	32
30	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
31	Clinical detection of deletion structural variants in whole-genome sequences. Npj Genomic Medicine, 2016, 1, 16026.	3.8	29
32	A patient with polymerase E1 deficiency (POLE1): clinical features and overlap with DNA breakage/instability syndromes. BMC Medical Genetics, 2015, 16, 31.	2.1	26
33	MAGEL2â€related disorders: A study and case series. Clinical Genetics, 2019, 96, 493-505.	2.0	26
34	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
35	Phenotypic spectrum associated with SPECC1L pathogenic variants: new families and critical review of the nosology of Teebi, Opitz GBBB, and Baraitser-Winter syndromes. European Journal of Medical Genetics, 2019, 62, 103588.	1.3	24
36	Functional validation of novel compound heterozygous variants in B3GAT3 resulting in severe osteopenia and fractures: expanding the disease phenotype. BMC Medical Genetics, 2016, 17, 86.	2.1	22

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37	Estimating the relative frequency of leukodystrophies and recommendations for carrier screening in the era of nextâ€generation sequencing. American Journal of Medical Genetics, Part A, 2020, 182, 1906-1912.	1.2	22
38	The Challenge of Analyzing the Results of Next-Generation Sequencing in Children. Pediatrics, 2016, 137, S3-S7.	2.1	19
39	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	2.9	19
40	POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e1-e1.	7.6	17
41	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	2.4	16
42	Mutations in GRK2 cause Jeune syndrome by impairing Hedgehog and canonical Wnt signaling. EMBO Molecular Medicine, 2020, 12, e11739.	6.9	16
43	Hypotonia and intellectual disability without dysmorphic features in a patient with PIGN-related disease. BMC Medical Genetics, 2017, 18, 124.	2.1	15
44	Challenges in genetic testing: clinician variant interpretation processes and the impact on clinical care. Genetics in Medicine, 2021, 23, 2289-2299.	2.4	15
45	Factors Affecting Migration to GRCh38 in Laboratories Performing Clinical Next-Generation Sequencing. Journal of Molecular Diagnostics, 2021, 23, 651-657.	2.8	13
46	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	6.2	13
47	Targeted next-generation sequencing revealed MYD88 deficiency in a child with chronic yersiniosis and granulomatous lymphadenitis. Journal of Allergy and Clinical Immunology, 2016, 137, 1591-1595.e4.	2.9	12
48	Arthrogryposis and pterygia as lethal end manifestations of genetically defined congenital myopathies. American Journal of Medical Genetics, Part A, 2018, 176, 359-367.	1.2	12
49	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
50	On the verge of diagnosis: Detection, reporting, and investigation of de novo variants in novel genes identified by clinical sequencing. Human Mutation, 2018, 39, 1505-1516.	2.5	9
51	Biallelic Loss-of-Function Variants in AIMP1 Cause a Rare Neurodegenerative Disease. Journal of Child Neurology, 2019, 34, 74-80.	1.4	9
52	Pathogenic variants in KPTN gene identified by clinical whole-genome sequencing. Journal of Physical Education and Sports Management, 2020, 6, a003970.	1.2	9
53	De novo heterozygous missense and lossâ€ofâ€function variants in CDC42BPB are associated with a neurodevelopmental phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 962-973.	1.2	8
54	Syndromic neurodevelopmental disorder associated with de novo variants in <scp>DDX23</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 2863-2872.	1.2	8

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55	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	2.4	8
56	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	2.4	7
57	Novel heterozygous pathogenic variants in CHUK in a patient with AEC-like phenotype, immune deficiencies and 1q21.1 microdeletion syndrome: a case report. BMC Medical Genetics, 2018, 19, 41.	2.1	6
58	Novel biallelic variants in NRROS associated with a lethal microgliopathy, brain calcifications, and neurodegeneration. Neurogenetics, 2022, 23, 151-156.	1.4	5
59	ITSN1: a novel candidate gene involved in autosomal dominant neurodevelopmental disorder spectrum. European Journal of Human Genetics, 2022, 30, 111-116.	2.8	4
60	Functionally impaired RPL8 variants associated with Diamondâ€“Blackfan anemia and a Diamondâ€“Blackfan anemiaâ€like phenotype. Human Mutation, 2022, 43, 389-402.	2.5	4
61	Phenotypic expansion and variable expressivity in individuals with JARID2â€related intellectual disability: A case series. Clinical Genetics, 2022, 102, 136-141.	2.0	3
62	Oculodentodigital Dysplasia. Neurology, 2022, 98, 675-677.	1.1	2
63	Expert opinion and caution are imperative for interpretation of next generation sequencing data. European Journal of Medical Genetics, 2016, 59, 519-521.	1.3	1
64	Examination of rare genetic variants in dental enamel genes: The potential role of nextâ€generation sequencing in primary dental care. Orthodontics and Craniofacial Research, 2019, 22, 49-55.	2.8	1
65	Coming up to Speed on Whole Genome Sequencing in Critically Ill Children. Advances in Molecular Pathology, 2018, 1, 1-8.	0.4	0
66	Cardiac Arrest in a Child with Non-classic Lipoid Congenital Adrenal Hyperplasia Associated with a New STAR Gene Mutation. Journal of the Endocrine Society, 2021, 5, A122-A122.	0.2	0
67	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	2.4	0