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	ITSN1: a novel candidate gene involved in autosomal dominant neurodevelopmental disorder spectrum. European Journal of Human Genetics, 2022, 30, 111-116.	2.8	4
2	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	2.4	0
3	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
4	Novel biallelic variants in NRROS associated with a lethal microgliopathy, brain calcifications, and neurodegeneration. Neurogenetics, 2022, 23, 151-156.	1.4	5
5	Oculodentodigital Dysplasia. Neurology, 2022, 98, 675-677.	1.1	2
6	Genomic answers for children: Dynamic analyses of >1000 pediatric rare disease genomes. Genetics in Medicine, 2022, 24, 1336-1348.	2.4	37
7	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
8	Phenotypic expansion and variable expressivity in individuals with JARID2â€related intellectual disability: A case series. Clinical Genetics, 2022, 102, 136-141.	2.0	3
9	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
10	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
11	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
12	Factors Affecting Migration to GRCh38 in Laboratories Performing Clinical Next-Generation Sequencing. Journal of Molecular Diagnostics, 2021, 23, 651-657.	2.8	13
13	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
14	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
15	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	2.4	16
16	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	2.4	7
17	Challenges in genetic testing: clinician variant interpretation processes and the impact on clinical care. Genetics in Medicine, 2021, 23, 2289-2299.	2.4	15
18	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	2.4	8

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19	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
20	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
21	Pathogenic variants in KPTN gene identified by clinical whole-genome sequencing. Journal of Physical Education and Sports Management, 2020, 6, a003970.	1.2	9
22	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
23	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
24	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
25	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
26	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. Science Advances, 2020, 6, eaax0021.	10.3	56
27	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. Journal of Clinical Investigation, 2020, 130, 1431-1445.	8.2	40
28	Mutations in GRK2 cause Jeune syndrome by impairing Hedgehog and canonical Wnt signaling. EMBO Molecular Medicine, 2020, 12, e11739.	6.9	16
29	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	2.9	19
30	Clinical genome sequencing in an unbiased pediatric cohort. Genetics in Medicine, 2019, 21, 303-310.	2.4	36
31	MAGEL2â€related disorders: A study and case series. Clinical Genetics, 2019, 96, 493-505.	2.0	26
32	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	6.2	42
33	Spectrum of K_V2.1 Dysfunction in KCNB1â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2019, 86, 899-912.	5.3	52
34	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
35	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
36	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

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37	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
38	Biallelic Loss-of-Function Variants in AIMP1 Cause a Rare Neurodegenerative Disease. Journal of Child Neurology, 2019, 34, 74-80.	1.4	9
39	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
40	POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e1-e1.	7.6	17
41	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
42	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
43	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
44	Coming up to Speed on Whole Genome Sequencing in Critically Ill Children. Advances in Molecular Pathology, 2018, 1, 1-8.	0.4	0
45	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
46	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
47	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
48	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	9.0	79
49	Hypotonia and intellectual disability without dysmorphic features in a patient with PIGN-related disease. BMC Medical Genetics, 2017, 18, 124.	2.1	15
50	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. Frontiers in Immunology, 2016, 7, 466.	4.8	80
51	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
52	Clinical detection of deletion structural variants in whole-genome sequences. Npj Genomic Medicine, 2016, 1, 16026.	3.8	29
53	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
54	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

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55	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
56	Neonatal progeriod syndrome associated with biallelic truncating variants in POLR3A. American Journal of Medical Genetics, Part A, 2016, 170, 3343-3346.	1.2	37
57	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
58	The Challenge of Analyzing the Results of Next-Generation Sequencing in Children. Pediatrics, 2016, 137, S3-S7.	2.1	19
59	A 26-hour system of highly sensitive whole genome sequencing for emergency management of genetic diseases. Genome Medicine, 2015, 7, 100.	8.2	237
60	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
61	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
62	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
63	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
64	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
65	Recessive mutations in POLR1C cause a leukodystrophy by impairing biogenesis of RNA polymerase III. Nature Communications, 2015, 6, 7623.	12.8	127
66	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
67	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