Frances Lucy Raymond

List of Publications by Year
in descending order

70	4,100	159585 30	133252 59
papers	citations	h-index	g-index

76	76	76	8456
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	6.2	343
2	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
3	Quantification of Homozygosity in Consanguineous Individuals with Autosomal Recessive Disease. American Journal of Human Genetics, 2006, 78, 889-896.	6.2	225
4	Whole genome sequencing reveals that genetic conditions are frequent in intensively ill children. Intensive Care Medicine, 2019, 45, 627-636.	8.2	183
5	Targeted Nextâ€Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. Human Mutation, 2015, 36, 1197-1204.	2.5	161
6	Mutations in ZDHHC9, Which Encodes a Palmitoyltransferase of NRAS and HRAS, Cause X-Linked Mental Retardation Associated with a Marfanoid Habitus. American Journal of Human Genetics, 2007, 80, 982-987.	6.2	150
7	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	6.2	121
8	A gain-of-function variant in DIAPH1 causes dominant macrothrombocytopenia and hearing loss. Blood, 2016, 127, 2903-2914.	1.4	121
9	Mutations in USP9X Are Associated with X-Linked Intellectual Disability and Disrupt Neuronal Cell Migration and Growth. American Journal of Human Genetics, 2014, 94, 470-478.	6.2	117
10	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. Genome Medicine, 2018, 10, 95.	8.2	111
11	Phenotypic insights into ADCY5 â€associated disease. Movement Disorders, 2016, 31, 1033-1040.	3.9	106
12	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	7.6	98
13	De Novo Loss-of-Function Mutations in SETD5, Encoding a Methyltransferase in a 3p25 Microdeletion Syndrome Critical Region, Cause Intellectual Disability. American Journal of Human Genetics, 2014, 94, 618-624.	6.2	96
14	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	6.2	96
15	Psychiatric disorders in children with 16p11.2 deletion and duplication. Translational Psychiatry, 2019, 9, 8.	4.8	93
16	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. American Journal of Human Genetics, 2018, 103, 1045-1052.	6.2	89
17	A dominant gain-of-function mutation in universal tyrosine kinase SRC causes thrombocytopenia, myelofibrosis, bleeding, and bone pathologies. Science Translational Medicine, 2016, 8, 328ra30.	12.4	87
18	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87

#	Article	IF	Citations
19	Genotypeâ€“phenotype associations in children with copy number variants associated with high neuropsychiatric risk in the UK (IMAGINE-ID): a case-control cohort study. Lancet Psychiatry,the, 2019, 6, 493-505.	7.4	87
20	Clinical and molecular predictors of mortality in neurofibromatosis 2: a UK national analysis of 1192 patients. Journal of Medical Genetics, 2015, 52, 699-705.	3.2	78
21	Spinal muscular atrophy diagnosis and carrier screening from genome sequencing data. Genetics in Medicine, 2020, 22, 945-953.	2.4	78
22	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	6.2	75
23	The genetics of mental retardation. Human Molecular Genetics, 2006, 15, R110-R116.	2.9	69
24	Detailed Clinical Phenotype and Molecular Genetic Findings in CLN3-Associated Isolated Retinal Degeneration. JAMA Ophthalmology, 2017, 135, 749.	2.5	61
25	Molecular prenatal diagnosis: the impact of modern technologies. Prenatal Diagnosis, 2010, 30, 674-681.	2.3	58
26	Cloning, genomic organization, alternative splicing and expression analysis of the human gene WNK3 (PRKWNK3). Gene, 2004, 335, 109-119.	2.2	52
27	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
28	Eight further individuals with intellectual disability and epilepsy carrying bi-allelicCNTNAP2aberrations allow delineation of the mutational and phenotypic spectrum. Journal of Medical Genetics, 2016, 53, 820-827.	3.2	45
29	De novo DDX3X missense variants in males appear viable and contribute to syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 570-578.	1.2	42
30	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	6.2	41
31	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. American Journal of Human Genetics, 2020, 107, 1129-1148.	6.2	38
32	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
33	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
34	Specific Alleles of CLN7/MFSD8, a Protein That Localizes to Photoreceptor Synaptic Terminals, Cause a Spectrum of Nonsyndromic Retinal Dystrophy. , 2017, 58, 2906.		35
35	Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene IFT140. , 2016, 57, 1053.		33
36	Epilepsy, cognitive deficits and neuroanatomy in males with <scp>ZDHHC</scp>9 mutations. Annals of Clinical and Translational Neurology, 2015, 2, 559-569.	3.7	31

#	Article	IF	Citations
37	Long-range evolutionary constraints reveal cis-regulatory interactions on the human X chromosome. Nature Communications, 2015, 6, 6904.	12.8	31
38	Analysis of Multiple Families With Single Individuals Affected by Pseudohypoparathyroidism Type Ib (PHP1B) Reveals Only One Novel Maternally Inherited GNAS Deletion. Journal of Bone and Mineral Research, 2016, 31, 796-805.	2.8	31
39	Mutations in CACNA2D4 Cause Distinctive Retinal Dysfunction in Humans. Ophthalmology, 2016, 123, 668-671.e2.	5.2	29
40	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. Journal of Human Genetics, 2016, 61, 95-101.	2.3	29
41	Re-annotation of 191 developmental and epileptic encephalopathy-associated genes unmasks de novo variants in SCN1A. Npj Genomic Medicine, 2019, 4, 31.	3.8	27
42	Childhood intellectual disability and parents' mental health: integrating social, psychological and genetic influences. British Journal of Psychiatry, 2021, 218, 315-322.	2.8	27
43	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	6.2	26
44	Reevaluation of the Retinal Dystrophy Due to Recessive Alleles of RGR With the Discovery of a Cis-Acting Mutation in CDHR1. , 2016, 57, 4806.		25
45	Abundancy of polymorphic CGG repeats in the human genome suggest a broad involvement in neurological disease. Scientific Reports, 2021, 11, 2515.	3.3	25
46	Lessons learnt from large-scale exon re-sequencing of the X chromosome. Human Molecular Genetics, 2009, 18, R60-R64.	2.9	23
47	Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa. JAMA Ophthalmology, 2017, 135, 137.	2.5	23
48	Structural brain abnormalities in a single gene disorder associated with epilepsy, language impairment and intellectual disability. NeuroImage: Clinical, 2016, 12, 655-665.	2.7	22
49	Myoclonusâ€dystonia caused by GNB1 mutation responsive to deep brain stimulation. Movement Disorders, 2019, 34, 1079-1080.	3.9	22
50	<scp>DNAJC6</scp> Mutations Disrupt Dopamine Homeostasis in Juvenile <scp>Parkinsonismâ€Dystonia</scp>. Movement Disorders, 2020, 35, 1357-1368.	3.9	22
51	Mutations in AGBL5, Encoding Î±-Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
52	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	12.8	20
53	Global and Local Connectivity Differences Converge With Gene Expression in a Neurodevelopmental Disorder of Known Genetic Origin. Cerebral Cortex, 2017, 27, 3806-3817.	2.9	17
54	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14

#	Article	IF	Citations
55	MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. Nucleic Acids Research, 2021, 49, 9686-9695.	14.5	14
56	Elucidating the molecular mechanisms associated with TARS2-related mitochondrial disease. Human Molecular Genetics, 2022, 31, 523-534.	2.9	12
57	Structural analysis of pathogenic missense mutations in GABRA2 and identification of a novel de novo variant in the desensitization gate. Molecular Genetics & Genomic Medicine, 2020, 8, e1106.	1.2	9
58	Long-Read Sequencing Identifies the First Retrotransposon Insertion and Resolves Structural Variants Causing Antithrombin Deficiency. Thrombosis and Haemostasis, 2022, 122, 1369-1378.	3.4	9
59	Psychopathology and cognitive performance in individuals with membrane-associated guanylate kinase mutations: a functional network phenotyping study. Journal of Neurodevelopmental Disorders, 2015, 7, 8.	3.1	7
60	Rare Genetic Variation in 135 Families With Family History Suggestive of X-Linked Intellectual Disability. Frontiers in Genetics, 2019, 10, 578.	2.3	7
61	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. JAMA Neurology, 2022, 79, 405.	9.0	7
62	The psychiatric phenotypes of 1q21 distal deletion and duplication. Translational Psychiatry, 2021, 11, 105.	4.8	6
63	Refinements and considerations for trio whole-genome sequence analysis when investigating Mendelian diseases presenting in early childhood. Human Genetics and Genomics Advances, 2022, 3, 100113.	1.7	4
64	Novel KAT6B proximal familial variant expands genotypic and phenotypic spectrum. Clinical Genetics, 2019, 95, 334-335.	2.0	3
65	Clinical Genomics in Critically Ill Infants and Children. JAMA - Journal of the American Medical Association, 2020, 323, 2480.	7.4	3
66	Identification and functional modelling of plausibly causative cis-regulatory variants in a highly-selected cohort with X-linked intellectual disability. PLoS ONE, 2021, 16, e0256181.	2.5	3
67	A Novel ATRX Mutation Presenting with Intellectual Disability and Severe Kyphoscoliosis. Fetal and Pediatric Pathology, 2020, 39, 539-543.	0.7	2
68	Novel phosphopantothenoylcysteine synthetase (<scp> PPCS </scp>) mutations with prominent neuromuscular features: Expanding the phenotypical spectrum of <scp> PPCS </scp> â€related disorders. American Journal of Medical Genetics, Part A, 0, , .	1.2	1
69	A Chromosome Breakpoint Mapping Strategy to Identify Candidate Genes for Nonsyndromic X-linked Mental Retardation within Xp11.2. Clinical Science, 2003, 104, 39P-40P.	0.0	0
70	1465â€...Mitochondrial encephalomyopathy lactic acidosis and stroke-like episodes (MELAS) syndrome: the highly variable diagnostic journey. , 2021, , .		0