Emily G Farrow

List of Publications by Year
in descending order

52 papers	2,626 citations	257450 24 h-index	233421 45 g-index
56 all docs	56 docs citations	56 times ranked	5832 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	AlstrÃ¶m Syndrome: Mutation Spectrum ofALMS1. Human Mutation, 2015, 36, 660-668.	2.5	117
6	Constellation: a tool for rapid, automated phenotype assignment of a highly polymorphic pharmacogene, CYP2D6, from whole-genome sequences. Npj Genomic Medicine, 2016, 1, 15007.	3.8	93
7	Erythropoietin stimulates murine and human fibroblast growth factor-23, revealing novel roles for bone and bone marrow. Haematologica, 2017, 102, e427-e430.	3.5	93
8	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. Frontiers in Immunology, 2016, 7, 466.	4.8	80
9	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
10	Diagnosis of mitochondrial disorders by concomitant next-generation sequencing of the exome and mitochondrial genome. Genomics, 2013, 102, 148-156.	2.9	68
11	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
12	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
13	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
14	Next-generation community genetics for low- and middle-income countries. Genome Medicine, 2012, 4, 25.	8.2	51
15	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
16	De novoframeshift mutation in ASXL3 in a patient with global developmental delay, microcephaly, and craniofacial anomalies. BMC Medical Genomics, 2013, 6, 32.	1.5	43
17	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
18	GPR37L1 modulates seizure susceptibility: Evidence from mouse studies and analyses of a human GPR37L1 variant. Neurobiology of Disease, 2017, 106, 181-190.	4.4	38

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19	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
20	Neonatal progeriod syndrome associated with biallelic truncating variants in POLR3A. American Journal of Medical Genetics, Part A, 2016, 170, 3343-3346.	1.2	37
21	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
22	Genomic answers for children: Dynamic analyses of >1000 pediatric rare disease genomes. Genetics in Medicine, 2022, 24, 1336-1348.	2.4	37
23	Clinical genome sequencing in an unbiased pediatric cohort. Genetics in Medicine, 2019, 21, 303-310.	2.4	36
24	Whole exome sequencing reveals HSPA1L as a genetic risk factor for spontaneous preterm birth. PLoS Genetics, 2018, 14, e1007394.	3.5	35
25	Clinical detection of deletion structural variants in whole-genome sequences. Npj Genomic Medicine, 2016, 1, 16026.	3.8	29
26	Autoimmune hyperphosphatemic tumoral calcinosis in a patient with FGF23 autoantibodies. Journal of Clinical Investigation, 2018, 128, 5368-5373.	8.2	27
27	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
28	CYP2C8, CYP2C9, and CYP2C19 Characterization Using Next-Generation Sequencing and Haplotype Analysis. Journal of Molecular Diagnostics, 2022, 24, 337-350.	2.8	23
29	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
30	Landscape of Somatic Mutations and Gene Expression Changes in Relapsed Infant MLL-Rearranged Acute Lymphoblastic Leukemia. Blood, 2016, 128, 1735-1735.	1.4	22
31	A novel nonsense substitution identified in the <scp>AMIGO</scp>2 gene in an OculoâEAuriculoâEVertebral spectrum patient. Orthodontics and Craniofacial Research, 2019, 22, 163-167.	2.8	20
32	POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e1-e1.	7.6	17
33	Hypotonia and intellectual disability without dysmorphic features in a patient with PIGN-related disease. BMC Medical Genetics, 2017, 18, 124.	2.1	15
34	Challenges in genetic testing: clinician variant interpretation processes and the impact on clinical care. Genetics in Medicine, 2021, 23, 2289-2299.	2.4	15
35	Rare Genetic Variants in Immune Genes and Neonatal Herpes Simplex Viral Infections. Pediatrics, 2021, 147, .	2.1	15
36	Factors Affecting Migration to GRCh38 in Laboratories Performing Clinical Next-Generation Sequencing. Journal of Molecular Diagnostics, 2021, 23, 651-657.	2.8	13

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37	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
38	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
39	Pathogenic variants in KPTN gene identified by clinical whole-genome sequencing. Journal of Physical Education and Sports Management, 2020, 6, a003970.	1.2	9
40	High Molecular Diagnosis Rate in Undermasculinized Males with Differences in Sex Development Using a Stepwise Approach. Endocrinology, 2020, 161, .	2.8	7
41	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
42	In vivo characterization of CYP2D6*12, *29 and *84 using dextromethorphan as a probe drug: a case report. Pharmacogenomics, 2017, 18, 427-431.	1.3	5
43	Phenotypic expansion and variable expressivity in individuals with JARID2â€related intellectual disability: A case series. Clinical Genetics, 2022, 102, 136-141.	2.0	3
44	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
45	An Unusual Presentation of Congenital Dyserythropoietic Anemia Type II (CDAII) Associated with Severe Anemia in a Patient with a Novel Mutation of the SEC23B Gene. Blood, 2012, 120, 990-990.	1.4	1
46	P-199â€ƒPathogenic CFTR Mutation in CrohnÊ¼s Disease in the Absence of Other CFTR-Related Manifestations. Inflammatory Bowel Diseases, 2016, 22, S69.	1.9	0
47	Coming up to Speed on Whole Genome Sequencing in Critically Ill Children. Advances in Molecular Pathology, 2018, 1, 1-8.	0.4	0
48	Using dried blood spots for variant analysis for patients with haemophilia. Haemophilia, 2019, 25, e339-e341.	2.1	0
49	Use of Dried Blood Spots for High through-Put, Rapid Turnaround Mutational Analysis in Patients with Hemophilia. Blood, 2014, 124, 5034-5034.	1.4	0
50	Single Cell Sequencing Reveals Heterogeneity of Gene Expression in KMT2A Rearranged Infant ALL at Relapse Compared to Diagnosis. Blood, 2019, 134, 2756-2756.	1.4	0
51	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	2.4	0
52	Germline Variants Associated with Cancer Predisposition and Bone Marrow Failure Are Common in KMT2A-r Infant Acute Lymphoblastic Leukemia Patients. Blood, 2020, 136, 41-41.	1.4	0