

# Emily G Farrow

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. <i>Genetics in Medicine</i> , 2022, 24, 631-644.	2.4	0
2	CYP2C8, CYP2C9, and CYP2C19 Characterization Using Next-Generation Sequencing and Haplotype Analysis. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 337-350.	2.8	23
3	Genomic answers for children: Dynamic analyses of >1000 pediatric rare disease genomes. <i>Genetics in Medicine</i> , 2022, 24, 1336-1348.	2.4	37
4	Phenotypic expansion and variable expressivity in individuals with <i>JARID2</i> -related intellectual disability: A case series. <i>Clinical Genetics</i> , 2022, 102, 136-141.	2.0	3
5	Factors Affecting Migration to GRCh38 in Laboratories Performing Clinical Next-Generation Sequencing. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 651-657.	2.8	13
6	Challenges in genetic testing: clinician variant interpretation processes and the impact on clinical care. <i>Genetics in Medicine</i> , 2021, 23, 2289-2299.	2.4	15
7	Rare Genetic Variants in Immune Genes and Neonatal Herpes Simplex Viral Infections. <i>Pediatrics</i> , 2021, 147, .	2.1	15
8	Pathogenic variants in <i>KPTN</i> gene identified by clinical whole-genome sequencing. <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a003970.	1.2	9
9	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	6.2	37
10	High Molecular Diagnosis Rate in Undermasculinized Males with Differences in Sex Development Using a Stepwise Approach. <i>Endocrinology</i> , 2020, 161, .	2.8	7
11	Germline Variants Associated with Cancer Predisposition and Bone Marrow Failure Are Common in KMT2A-r Infant Acute Lymphoblastic Leukemia Patients. <i>Blood</i> , 2020, 136, 41-41.	1.4	0
12	Clinical genome sequencing in an unbiased pediatric cohort. <i>Genetics in Medicine</i> , 2019, 21, 303-310.	2.4	36
13	Using dried blood spots for variant analysis for patients with haemophilia. <i>Haemophilia</i> , 2019, 25, e339-e341.	2.1	0
14	Examination of rare genetic variants in dental enamel genes: The potential role of next-generation sequencing in primary dental care. <i>Orthodontics and Craniofacial Research</i> , 2019, 22, 49-55.	2.8	1
15	A novel nonsense substitution identified in the <i>AMIGO2</i> gene in an Occulo-Auriculo-Vertebral spectrum patient. <i>Orthodontics and Craniofacial Research</i> , 2019, 22, 163-167.	2.8	20
16	Single Cell Sequencing Reveals Heterogeneity of Gene Expression in KMT2A Rearranged Infant ALL at Relapse Compared to Diagnosis. <i>Blood</i> , 2019, 134, 2756-2756.	1.4	0
17	The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. <i>Npj Genomic Medicine</i> , 2018, 3, 6.	3.8	156
18	POLR3A variants in hereditary spastic paraplegia and ataxia. <i>Brain</i> , 2018, 141, e1-e1.	7.6	17

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19	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2018, 102, 744-759.	6.2	51
20	Coming up to Speed on Whole Genome Sequencing in Critically Ill Children. <i>Advances in Molecular Pathology</i> , 2018, 1, 1-8.	0.4	0
21	On the verge of diagnosis: Detection, reporting, and investigation of de novo variants in novel genes identified by clinical sequencing. <i>Human Mutation</i> , 2018, 39, 1505-1516.	2.5	9
22	Whole exome sequencing reveals HSPA1L as a genetic risk factor for spontaneous preterm birth. <i>PLoS Genetics</i> , 2018, 14, e1007394.	3.5	35
23	Novel heterozygous pathogenic variants in CHUK in a patient with AEC-like phenotype, immune deficiencies and 1q21.1 microdeletion syndrome: a case report. <i>BMC Medical Genetics</i> , 2018, 19, 41.	2.1	6
24	Autoimmune hyperphosphatemic tumoral calcinosis in a patient with FGF23 autoantibodies. <i>Journal of Clinical Investigation</i> , 2018, 128, 5368-5373.	8.2	27
25	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase ( <i>TRIT1</i> ) gene. <i>Human Mutation</i> , 2017, 38, 511-516.	2.5	39
26	In vivo characterization of CYP2D6*12, *29 and *84 using dextromethorphan as a probe drug: a case report. <i>Pharmacogenomics</i> , 2017, 18, 427-431.	1.3	5
27	Erythropoietin stimulates murine and human fibroblast growth factor-23, revealing novel roles for bone and bone marrow. <i>Haematologica</i> , 2017, 102, e427-e430.	3.5	93
28	GPR37L1 modulates seizure susceptibility: Evidence from mouse studies and analyses of a human GPR37L1 variant. <i>Neurobiology of Disease</i> , 2017, 106, 181-190.	4.4	38
29	Hypotonia and intellectual disability without dysmorphic features in a patient with PIGN-related disease. <i>BMC Medical Genetics</i> , 2017, 18, 124.	2.1	15
30	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. <i>Frontiers in Immunology</i> , 2016, 7, 466.	4.8	80
31	<i>PCDH19</i> -related epileptic encephalopathy in a male mosaic for a truncating variant. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1585-1589.	1.2	37
32	P-199 Pathogenic CFTR Mutation in Crohn's Disease in the Absence of Other CFTR-Related Manifestations. <i>Inflammatory Bowel Diseases</i> , 2016, 22, S69.	1.9	0
33	Constellation: a tool for rapid, automated phenotype assignment of a highly polymorphic pharmacogene, CYP2D6, from whole-genome sequences. <i>Npj Genomic Medicine</i> , 2016, 1, 15007.	3.8	93
34	Clinical detection of deletion structural variants in whole-genome sequences. <i>Npj Genomic Medicine</i> , 2016, 1, 16026.	3.8	29
35	Targeted next-generation sequencing revealed MYD88 deficiency in a child with chronic yersiniosis and granulomatous lymphadenitis. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 137, 1591-1595.e4.	2.9	12
36	Biallelic Mutations in TBCD, Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 99, 962-973.	6.2	66

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37	Neonatal progeroid syndrome associated with biallelic truncating variants in <i>POLR3A</i> . American Journal of Medical Genetics, Part A, 2016, 170, 3343-3346.	1.2	37
38	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
39	Landscape of Somatic Mutations and Gene Expression Changes in Relapsed Infant MLL-Rearranged Acute Lymphoblastic Leukemia. Blood, 2016, 128, 1735-1735.	1.4	22
40	Alström Syndrome: Mutation Spectrum of <i>ALMS1</i> . Human Mutation, 2015, 36, 660-668.	2.5	117
41	A 26-hour system of highly sensitive whole genome sequencing for emergency management of genetic diseases. Genome Medicine, 2015, 7, 100.	8.2	237
42	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
43	Whole-genome sequencing for identification of Mendelian disorders in critically ill infants: a retrospective analysis of diagnostic and clinical findings. Lancet Respiratory Medicine, 2015, 3, 377-387.	10.7	322
44	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
45	A novel epileptic encephalopathy mutation in <i>KCNB1</i> disrupts Kv2.1 ion selectivity, expression, and localization. Journal of General Physiology, 2015, 146, 399-410.	1.9	79
46	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
47	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
48	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
49	Diagnosis of mitochondrial disorders by concomitant next-generation sequencing of the exome and mitochondrial genome. Genomics, 2013, 102, 148-156.	2.9	68
50	De novo frameshift mutation in ASXL3 in a patient with global developmental delay, microcephaly, and craniofacial anomalies. BMC Medical Genomics, 2013, 6, 32.	1.5	43
51	Next-generation community genetics for low- and middle-income countries. Genome Medicine, 2012, 4, 25.	8.2	51
52	An Unusual Presentation of Congenital Dyserythropoietic Anemia Type II (CDAIL) Associated with Severe Anemia in a Patient with a Novel Mutation of the SEC23B Gene. Blood, 2012, 120, 990-990.	1.4	1