Anne H O donnell-Luria

List of Publications by Year in Descending Order

62 papers	12,077 citations	25 h-index	87 g-index
87 ext. papers	17,388 ext. citations	14.3 avg, IF	4.76 L-index

#	Paper	IF	Citations
62	Inverting the model of genomics data sharing with the NHGRI Genomic Data Science Analysis, Visualization, and Informatics Lab-space.. *Cell Genomics*, **2022**, 2, 100085-100085		6
61	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
60	Genes To Mental Health (G2MH): A Framework to Map the Combined Effects of Rare and Common Variants on Dimensions of Cognition and Psychopathology.. *American Journal of Psychiatry*, **2022**, 179, 189-203	11.9	1
59	seqr: a web-based analysis and collaboration tool for rare disease genomics.. *Human Mutation*, **2022** ,	4.7	1
58	Neurogenetic fetal akinesia and arthrogryposis: genetics, expanding genotype-phenotypes and functional genomics. *Journal of Medical Genetics*, **2021**, 58, 609-618	5.8	13
57	Novel variants in TUBA1A cause congenital fibrosis of the extraocular muscles with or without malformations of cortical brain development. *European Journal of Human Genetics*, **2021**, 29, 816-826	5.3	1
56	Familial thrombocytopenia due to a complex structural variant resulting in a WAC-ANKRD26 fusion transcript. *Journal of Experimental Medicine*, **2021**, 218,	16.6	5
55	Comprehensive analysis of ADA2 genetic variants and estimation of carrier frequency driven by a function-based approach. *Journal of Allergy and Clinical Immunology*, **2021**,	11.5	4
54	A form of muscular dystrophy associated with pathogenic variants in JAG2. *American Journal of Human Genetics*, **2021**, 108, 840-856	11	3
53	Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. *Frontiers in Genetics*, **2021**, 12, 674295	4.5	2
52	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. *Nature Communications*, **2021**, 12, 3505	17.4	5
51	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 119-133	2.5	6
50	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. *American Journal of Human Genetics*, **2021**, 108, 357-367	11	1
49	O'Donnell-Luria-Rodan syndrome: description of a second multinational cohort and refinement of the phenotypic spectrum. *Journal of Medical Genetics*, **2021**,	5.8	1
48	Unique variants in CLCN3, encoding an endosomal anion/proton exchanger, underlie a spectrum of neurodevelopmental disorders. *American Journal of Human Genetics*, **2021**, 108, 1450-1465	11	0
47	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2021**, 597, E3-E4	50.4	3
46	Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1809	2.3	1

45	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. *Genetics in Medicine*, **2020**, 22, 1215-1226	8.1	7
44	Evaluating drug targets through human loss-of-function genetic variation. *Nature*, **2020**, 581, 459-464	50.4	53
43	The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2020**, 581, 434-443	50.4	2278
42	Characterising the loss-of-function impact of 5' untranslated region variants in 15,708 individuals. *Nature Communications*, **2020**, 11, 2523	17.4	35
41	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. *Nature Communications*, **2020**, 11, 2539	17.4	51
40	A structural variation reference for medical and population genetics. *Nature*, **2020**, 581, 444-451	50.4	223
39	Transcript expression-aware annotation improves rare variant interpretation. *Nature*, **2020**, 581, 452-458	50.4	55
38	Apcdd1 is a dual BMP/Wnt inhibitor in the developing nervous system and skin. *Developmental Biology*, **2020**, 464, 71-87	3.1	2
37	Identification of pathogenic variant enriched regions across genes and gene families. *Genome Research*, **2020**, 30, 62-71	9.7	14
36	Improving the Understanding of Genetic Variants in Rare Disease With Large-scale Reference Populations. *JAMA - Journal of the American Medical Association*, **2019**, 322, 1305-1306	27.4	3
35	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
34	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. *European Journal of Human Genetics*, **2019**, 27, 1398-1405	5.3	25
33	Genome Sequencing Identifies the Pathogenic Variant Missed by Prior Testing in an Infant with Marfan Syndrome. *Journal of Pediatrics*, **2019**, 213, 235-240	3.6	2
32	Characterization of Prevalence and Health Consequences of Uniparental Disomy in Four Million Individuals from the General Population. *American Journal of Human Genetics*, **2019**, 105, 921-932	11	36
31	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. *American Journal of Human Genetics*, **2019**, 104, 187-190	11	8
30	Reply to 'Selective effects of heterozygous protein-truncating variants'. *Nature Genetics*, **2019**, 51, 3-4	36.3	1
29	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
28	Megaloblastic Anemia Progressing to Severe Thrombotic Microangiopathy in Patients with Disordered Vitamin B Metabolism: Case Reports and Literature Review. *Journal of Pediatrics*, **2018**, 202, 315-319.e2	3.6	4

27	The Genetic Landscape of Diamond-Blackfan Anemia. *American Journal of Human Genetics*, **2018**, 103, 930-947	11	108
26	matchbox: An open-source tool for patient matching via the Matchmaker Exchange. *Human Mutation*, **2018**, 39, 1827-1834	4.7	13
25	Pathogenic ASXL1 somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome. *Human Mutation*, **2017**, 38, 517-523	4.7	34
24	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. *Nature*, **2017**, 544, 235-239	50.4	208
23	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. *Science Translational Medicine*, **2017**, 9,	17.5	338
22	Using high-resolution variant frequencies to empower clinical genome interpretation. *Genetics in Medicine*, **2017**, 19, 1151-1158	8.1	208
21	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. *Nature Genetics*, **2017**, 49, 806-810	36.3	84
20	Brain MRS glutamine as a biomarker to guide therapy of hyperammonemic coma. *Molecular Genetics and Metabolism*, **2017**, 121, 9-15	3.7	8
19	Utility of rapid whole-exome sequencing in the diagnosis of Niemann-Pick disease type C presenting with fetal hydrops and acute liver failure. *Journal of Physical Education and Sports Management*, **2017**, 3,	2.8	12
18	ClinVar data parsing. *Wellcome Open Research*, **2017**, 2, 33	4.8	16
17	Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, **2016**, 536, 285-91	50.4	6940
16	Quantifying prion disease penetrance using large population control cohorts. *Science Translational Medicine*, **2016**, 8, 322ra9	17.5	205
15	Health and population effects of rare gene knockouts in adult humans with related parents. *Science* , **2016**, 352, 474-7	33.3	185
14	A Clinician's perspective on clinical exome sequencing. *Human Genetics*, **2016**, 135, 643-54	6.3	28
13	Mutations in ARID2 are associated with intellectual disabilities. *Neurogenetics*, **2015**, 16, 307-14	3	40
12	Systematic evaluation of genome sequencing as a first-tier diagnostic test for prenatal and pediatric disorders		2
11	seqr : a web-based analysis and collaboration tool for rare disease genomics		1
10	Analysis of protein-coding genetic variation in 60,706 humans		81

List of Publications

9	Using high-resolution variant frequencies to empower clinical genome interpretation	4
8	Regional missense constraint improves variant deleteriousness prediction	102
7	The mutational constraint spectrum quantified from variation in 141,456 humans	381
6	Characterising the loss-of-function impact of 5′untranslated region variants in whole genome sequence data from 15,708 individuals	5
5	Transcript expression-aware annotation improves rare variant discovery and interpretation	8
4	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes	3
3	An open resource of structural variation for medical and population genetics	33
2	Pathogenic ASXL1 somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome	1
1	Inverting the model of genomics data sharing with the NHGRI Genomic Data Science Analysis, Visualization, and Informatics Lab-space (AnVIL)	5