Anne H O'donnell-Luria

List of Publications by Year
in descending order

58 papers	20,868 citations	172207 29 h-index	123241 61 g-index
87 all docs	87 docs citations	87 times ranked	41445 citing authors

#	Article	IF	Citations
1	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	13.7	9,051
2	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
3	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	13.7	614
4	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	5.8	516
5	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	1.1	355
6	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. Nature, 2017, 544, 235-239.	13.7	292
7	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	5.8	289
8	Health and population effects of rare gene knockouts in adult humans with related parents. Science, 2016, 352, 474-477.	6.0	272
9	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	2.6	184
10	Variant interpretation using population databases: Lessons from gnomAD. Human Mutation, 2022, 43, 1012-1030.	1.1	184
11	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
12	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. Nature Genetics, 2017, 49, 806-810.	9.4	157
13	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	13.7	142
14	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	13.7	115
15	Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523.	5.8	99
16	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. Nature Communications, 2020, 11, 2539.	5.8	98
17	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.	2.6	79
18	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	3.6	65

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19	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	1.4	60
20	Inverting the model of genomics data sharing with the NHGRI Genomic Data Science Analysis, Visualization, and Informatics Lab-space. Cell Genomics, 2022, 2, 100085.	3.0	59
21	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
22	Mutations in ARID2 are associated with intellectual disabilities. Neurogenetics, 2015, 16, 307-314.	0.7	54
23	Pathogenic ASXL1 somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome. Human Mutation, 2017, 38, 517-523.	1.1	49
24	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. Nature Communications, 2021, 12, 3505.	5.8	49
25	Identification of pathogenic variant enriched regions across genes and gene families. Genome Research, 2020, 30, 62-71.	2.4	47
26	Neurogenetic fetal akinesia and arthrogryposis: genetics, expanding genotype-phenotypes and functional genomics. Journal of Medical Genetics, 2021, 58, 609-618.	1.5	46
27	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
28	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
29	A Clinicianâ€™s perspective on clinical exome sequencing. Human Genetics, 2016, 135, 643-654.	1.8	33
30	seqr : A webâ€based analysis and collaboration tool for rare disease genomics. Human Mutation, 2022, , .	1.1	31
31	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.	4.0	29
32	Comprehensive analysis of ADA2 genetic variants and estimation of carrier frequency driven by a function-based approach. Journal of Allergy and Clinical Immunology, 2022, 149, 379-387.	1.5	27
33	Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. Frontiers in Genetics, 2021, 12, 674295.	1.1	23
34	Mendelian etiologies identified with whole exome sequencing in cerebral palsy. Annals of Clinical and Translational Neurology, 2022, 9, 193-205.	1.7	23
35	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
36	matchbox: An open-source tool for patient matching via the Matchmaker Exchange. Human Mutation, 2018, 39, 1827-1834.	1.1	20

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37	Familial thrombocytopenia due to a complex structural variant resulting in a WAC-ANKRD26 fusion transcript. Journal of Experimental Medicine, 2021, 218, .	4.2	20
38	ClinVar data parsing. Wellcome Open Research, 2017, 2, 33.	0.9	19
39	Utility of rapid whole-exome sequencing in the diagnosis of Niemannâ€“Pick disease type C presenting with fetal hydrops and acute liver failure. Journal of Physical Education and Sports Management, 2017, 3, a002147.	0.5	18
40	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
41	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.	2.6	16
42	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. American Journal of Human Genetics, 2019, 104, 187-190.	2.6	15
43	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	2.6	15
44	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.	2.6	14
45	A gene-to-patient approach uplifts novel disease gene discovery and identifies 18 putative novel disease genes. Genetics in Medicine, 2022, 24, 1697-1707.	1.1	14
46	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.	1.4	13
47	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	2.6	13
48	Apcdd1 is a dual BMP/Wnt inhibitor in the developing nervous system and skin. Developmental Biology, 2020, 464, 71-87.	0.9	11
49	Brain MRS glutamine as a biomarker to guide therapy of hyperammonemic coma. Molecular Genetics and Metabolism, 2017, 121, 9-15.	0.5	8
50	Recessive variants in COL25A1 gene as novel cause of arthrogryposis multiplex congenita with ocular congenital cranial dysinnervation disorder. Human Mutation, 2022, 43, 487-498.	1.1	8
51	Improving the Understanding of Genetic Variants in Rare Disease With Large-scale Reference Populations. JAMA - Journal of the American Medical Association, 2019, 322, 1305.	3.8	7
52	Genome Sequencing Identifies the Pathogenic Variant Missed by Prior Testing in an Infant with Marfan Syndrome. Journal of Pediatrics, 2019, 213, 235-240.	0.9	6
53	Reply to â€˜Selective effects of heterozygous protein-truncating variantsâ€™. Nature Genetics, 2019, 51, 3-4.	9.4	6
54	Megaloblastic Anemia Progressing to Severe Thrombotic Microangiopathy in Patients with Disordered Vitamin B12 Metabolism: Case Reports and Literature Review. Journal of Pediatrics, 2018, 202, 315-319.e2.	0.9	5

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55	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	1.1	5
56	Oâ€™Donnell-Luria-Rodan syndrome: description of a second multinational cohort and refinement of the phenotypic spectrum. Journal of Medical Genetics, 2021, , jmedgenet-2020-107470.	1.5	4
57	Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. Molecular Genetics & Genomic Medicine, 2021, 9, e1809.	0.6	4
58	More than a fancy exome: unique capabilities of genome sequencing for pediatric rare disease diagnosis. Molecular Genetics and Metabolism, 2021, 132, S88.	0.5	0