Eleanor G Seaby

List of Publications by Year
in descending order

32 papers	7,841 citations	430874 18 h-index	395702 33 g-index
37 all docs	37 docs citations	37 times ranked	18322 citing authors

#	Article	IF	Citations
1	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
2	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
3	Treatment of Multisystem Inflammatory Syndrome in Children. New England Journal of Medicine, 2021, 385, 11-22.	27.0	254
4	Collagen (COL4A) mutations are the most frequent mutations underlying adult focal segmental glomerulosclerosis. Nephrology Dialysis Transplantation, 2016, 31, 961-970.	0.7	199
5	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	27.8	142
6	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	2.4	99
7	Mutations specific to the Rac-GEF domain ofTRIOcause intellectual disability and microcephaly. Journal of Medical Genetics, 2016, 53, 735-742.	3.2	80
8	Exome sequencing explained: a practical guide to its clinical application. Briefings in Functional Genomics, 2016, 15, 374-384.	2.7	58
9	The benefits of being a near-peer teacher. Clinical Teacher, 2018, 15, 403-407.	0.8	57
10	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245.	6.2	56
11	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	27.8	45
12	Autosomal dominant tubulointerstitial kidney disease-UMOD is the most frequent non polycystic genetic kidney disease. BMC Nephrology, 2018, 19, 301.	1.8	39
13	Thrombotic microangiopathy following haematopoietic stem cell transplant. Pediatric Nephrology, 2018, 33, 1489-1500.	1.7	29
14	Challenges in the diagnosis and discovery of rare genetic disorders using contemporary sequencing technologies. Briefings in Functional Genomics, 2020, 19, 243-258.	2.7	27
15	Can medical students accurately predict their learning? A study comparing perceived and actual performance in neuroanatomy. Anatomical Sciences Education, 2016, 9, 488-495.	3.7	25
16	The Efficacy of Frontline Nearâ€Peer Teaching in a Modern Medical Curriculum. Anatomical Sciences Education, 2019, 12, 236-244.	3.7	25
17	Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. Frontiers in Genetics, 2021, 12, 674295.	2.3	23
18	Commercial chicken breeds exhibit highly divergent patterns of linkage disequilibrium. Heredity, 2016, 117, 375-382.	2.6	21

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19	Deleterious coding variants in multi-case families with non-syndromic cleft lip and/or palate phenotypes. Scientific Reports, 2016, 6, 30457.	3.3	19
20	Inactivation of AMMECR1 is associated with growth, bone, and heart alterations. Human Mutation, 2018, 39, 281-291.	2.5	15
21	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.	6.2	14
22	A gene-to-patient approach uplifts novel disease gene discovery and identifies 18 putative novel disease genes. Genetics in Medicine, 2022, 24, 1697-1707.	2.4	14
23	AMMECR1: a single point mutation causes developmental delay, midface hypoplasia and elliptocytosis. Journal of Medical Genetics, 2017, 54, 269-277.	3.2	12
24	Identification of novel locus associated with coronary artery aneurysms and validation of loci for susceptibility to Kawasaki disease. European Journal of Human Genetics, 2021, 29, 1734-1744.	2.8	10
25	Unexpected Findings in a Child with Atypical Hemolytic Uremic Syndrome: An Example of How Genomics Is Changing the Clinical Diagnostic Paradigm. Frontiers in Pediatrics, 2017, 5, 113.	1.9	9
26	Identification of disease-associated loci using machine learning for genotype and network data integration. Bioinformatics, 2019, 35, 5182-5190.	4.1	7
27	Rare pathogenic variants in WNK3 cause X-linked intellectual disability. Genetics in Medicine, 2022, 24, 1941-1951.	2.4	5
28	Bullous Herpes Zoster. Journal of Pediatrics, 2014, 164, 667.	1.8	4
29	Progressive myoclonic epilepsy with Fanconi syndrome. JRSM Open, 2016, 7, 205427041562314.	0.5	3
30	The Career Impact of the National Undergraduate Neuroanatomy Competition. World Neurosurgery, 2020, 133, e535-e539.	1.3	2
31	Sporadic Isolated Fanconi Syndrome due to a Mutation of EHHADH: A Case Report. Journal of Clinical Nephrology and Renal Care, 2017, 3, .	0.1	2
32	Genome-wide Association Studies in Infectious Diseases. Pediatric Infectious Disease Journal, 2016, 35, 802-804.	2.0	1