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	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
2	Rare pathogenic variants in WNK3 cause X-linked intellectual disability. Genetics in Medicine, 2022, 24, 1941-1951.	2.4	5
3	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
4	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
5	Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. Frontiers in Genetics, 2021, 12, 674295.	2.3	23
6	Treatment of Multisystem Inflammatory Syndrome in Children. New England Journal of Medicine, 2021, 385, 11-22.	27.0	254
7	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	27.8	45
8	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
9	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
10	The Career Impact of the National Undergraduate Neuroanatomy Competition. World Neurosurgery, 2020, 133, e535-e539.	1.3	2
11	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
12	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
13	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	27.8	142
14	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	2.4	99
15	Identification of disease-associated loci using machine learning for genotype and network data integration. Bioinformatics, 2019, 35, 5182-5190.	4.1	7
16	The Efficacy of Frontline Nearâ€Peer Teaching in a Modern Medical Curriculum. Anatomical Sciences Education, 2019, 12, 236-244.	3.7	25
17	Inactivation of AMMECR1 is associated with growth, bone, and heart alterations. Human Mutation, 2018, 39, 281-291.	2.5	15
18	The benefits of being a near-peer teacher. Clinical Teacher, 2018, 15, 403-407.	0.8	57

#	Article	IF	Citations
19	Thrombotic microangiopathy following haematopoietic stem cell transplant. Pediatric Nephrology, 2018, 33, 1489-1500.	1.7	29
20	Autosomal dominant tubulointerstitial kidney disease-UMOD is the most frequent non polycystic genetic kidney disease. BMC Nephrology, 2018, 19, 301.	1.8	39
21	AMMECR1: a single point mutation causes developmental delay, midface hypoplasia and elliptocytosis. Journal of Medical Genetics, 2017, 54, 269-277.	3.2	12
22	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
23	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
24	Genome-wide Association Studies in Infectious Diseases. Pediatric Infectious Disease Journal, 2016, 35, 802-804.	2.0	1
25	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
26	Mutations specific to the Rac-GEF domain ofTRIOcause intellectual disability and microcephaly. Journal of Medical Genetics, 2016, 53, 735-742.	3.2	80
27	Progressive myoclonic epilepsy with Fanconi syndrome. JRSM Open, 2016, 7, 205427041562314.	0.5	3
28	Commercial chicken breeds exhibit highly divergent patterns of linkage disequilibrium. Heredity, 2016, 117, 375-382.	2.6	21
29	Deleterious coding variants in multi-case families with non-syndromic cleft lip and/or palate phenotypes. Scientific Reports, 2016, 6, 30457.	3.3	19
30	Exome sequencing explained: a practical guide to its clinical application. Briefings in Functional Genomics, 2016, 15, 374-384.	2.7	58
31	Collagen (COL4A) mutations are the most frequent mutations underlying adult focal segmental glomerulosclerosis. Nephrology Dialysis Transplantation, 2016, 31, 961-970.	0.7	199
32	Bullous Herpes Zoster. Journal of Pediatrics, 2014, 164, 667.	1.8	4