Reuben J Pengelly

List of Publications by Year
in descending order

39	1,154	393982	414034
papers	citations	19 h-index	32 g-index

41	41	41	2893
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Collagen (COL4A) mutations are the most frequent mutations underlying adult focal segmental glomerulosclerosis. Nephrology Dialysis Transplantation, 2016, 31, 961-970.	0.4	199
2	Exome sequence read depth methods for identifying copy number changes. Briefings in Bioinformatics, 2015, 16, 380-392.	3.2	84
3	Mutations specific to the Rac-GEF domain ofTRIOcause intellectual disability and microcephaly. Journal of Medical Genetics, 2016, 53, 735-742.	1.5	80
4	Clinical efficacy of a nextâ€generation sequencing gene panel for primary immunodeficiency diagnostics. Clinical Genetics, 2018, 93, 647-655.	1.0	63
5	Exome sequencing explained: a practical guide to its clinical application. Briefings in Functional Genomics, 2016, 15, 374-384.	1.3	58
6	A SNP profiling panel for sample tracking in whole-exome sequencing studies. Genome Medicine, 2013, 5, 89.	3.6	57
7	Risk factors for situs defects and congenital heart disease in primary ciliary dyskinesia. Thorax, 2019, 74, 203-205.	2.7	52
8	Fragment Screening Using Capillary Electrophoresis (CEfrag) for Hit Identification of Heat Shock Protein 90 ATPase Inhibitors. Journal of Biomolecular Screening, 2012, 17, 868-876.	2.6	49
9	Identification of a functionally significant tri-allelic genotype in the Tyrosinase gene (TYR) causing hypomorphic oculocutaneous albinism (OCA1B). Scientific Reports, 2017, 7, 4415.	1.6	47
10	Autosomal dominant tubulointerstitial kidney disease-UMOD is the most frequent non polycystic genetic kidney disease. BMC Nephrology, 2018, 19, 301.	0.8	39
11	Rapid identification ofSaccharomyces eubayanusand its hybrids. FEMS Yeast Research, 2013, 13, 156-161.	1.1	35
12	Cold-induced urticarial autoinflammatory syndrome related to factor XII activation. Nature Communications, 2020, 11, 179.	5.8	32
13	Precision treatment with sirolimus in a case of activated phosphoinositide 3-kinase Î´ syndrome. Clinical Immunology, 2016, 171, 38-40.	1.4	28
14	Subclonal Evolution of Cancer-Related Gene Mutations in p53 Immunopositive Patches in Human Skin. Journal of Investigative Dermatology, 2018, 138, 189-198.	0.3	28
15	Autoimmunity/inflammation in a monogenic primary immunodeficiency cohort. Clinical and Translational Immunology, 2017, 6, e155.	1.7	27
16	Evaluating phenotype-driven approaches for genetic diagnoses from exomes in a clinical setting. Scientific Reports, 2017, 7, 13509.	1.6	26
17	Precision Molecular Diagnosis Defines Specific Therapy in Combined Immunodeficiency with Megaloblastic Anemia Secondary to MTHFD1 Deficiency. Journal of Allergy and Clinical Immunology: in Practice, 2016, 4, 1160-1166.e10.	2.0	25
18	Anticytokine autoantibodies in a patient with a heterozygous NFKB2 mutation. Journal of Allergy and Clinical Immunology, 2018, 141, 1479-1482.e6.	1.5	24

#	Article	IF	Citations
19	Adult-Onset Leukoencephalopathy with Axonal Spheroids and Pigmented Glia Caused by a Novel R782G Mutation in CSF1R. Scientific Reports, 2015, 5, 10042.	1.6	22
20	Deleterious coding variants in multi-case families with non-syndromic cleft lip and/or palate phenotypes. Scientific Reports, 2016, 6, 30457.	1.6	19
21	Analysis of Mutation and Loss of Heterozygosity by Whole-Exome Sequencing Yields Insights into Pseudomyxoma Peritonei. Journal of Molecular Diagnostics, 2018, 20, 635-642.	1.2	19
22	Sequencing era methods for identifying signatures of selection in the genome. Briefings in Bioinformatics, 2019, 20, 1997-2008.	3.2	18
23	Whole genome sequences are required to fully resolve the linkage disequilibrium structure of human populations. BMC Genomics, 2015, 16, 666.	1.2	14
24	Resolving clinical diagnoses for syndromic cleft lip and/or palate phenotypes using wholeâ€exome sequencing. Clinical Genetics, 2015, 88, 441-449.	1.0	14
25	Comparison of Mendeliome exome capture kits for use in clinical diagnostics. Scientific Reports, 2020, 10, 3235.	1.6	14
26	Linkage disequilibrium maps for European and African populations constructed from whole genome sequence data. Scientific Data, 2019, 6, 208.	2.4	11
27	Understanding the disease genome: gene essentiality and the interplay of selection, recombination and mutation. Briefings in Bioinformatics, 2019, 20, 267-273.	3.2	11
28	Immuno-Genomic Profiling of Patients with Inflammatory Bowel Disease. Inflammatory Bowel Diseases, 2014, 20, 1813-1819.	0.9	10
29	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.	0.9	9
30	A CRISPR and high-content imaging assay compliant with ACMG/AMP guidelines for clinical variant interpretation in ciliopathies. Human Genetics, 2021, 140, 593-607.	1.8	6
31	Restriction of an intron size en route to endothermy. Nucleic Acids Research, 2021, 49, 2460-2487.	6.5	6
32	Linkage disequilibrium maps to guide contig ordering for genome assembly. Bioinformatics, 2019, 35, 541-545.	1.8	5
33	Long term follow-up of a family with GUCY2D dominant cone dystrophy. International Journal of Ophthalmology, 2018, 11, 1945-1950.	0.5	5
34	Aarskog-Scott syndrome: phenotypic and genetic heterogeneity. AIMS Genetics, 2016, 03, 049-059.	1.9	4
35	Progressive myoclonic epilepsy with Fanconi syndrome. JRSM Open, 2016, 7, 205427041562314.	0.2	3
36	Gene-dense autosomal chromosomes show evidence for increased selection. Heredity, 2019, 123, 774-783.	1.2	3

#	Article	IF	Citations
37	Exonic splicing code and protein binding sites for calcium. Nucleic Acids Research, 2022, 50, 5493-5512.	6.5	3
38	Heterogeneity in the extent of linkage disequilibrium among exonic, intronic, non-coding RNA and intergenic chromosome regions. European Journal of Human Genetics, 2019, 27, 1436-1444.	1.4	2
39	zalpha: an R package for the identification of regions of the genome under selection. Journal of Open Source Software, 2020, 5, 2638.	2.0	0