# Reuben J Pengelly

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/967889/publications.pdf

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| 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 | Exonic splicing code and protein binding sites for calcium. Nucleic Acids Research, 2022, 50, 5493-5512. | 6.5 | 3 |
| 2 | 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 |
| 3 | Restriction of an intron size en route to endothermy. Nucleic Acids Research, 2021, 49, 2460-2487. | 6.5 | 6 |
| 4 | Cold-induced urticarial autoinflammatory syndrome related to factor XII activation. Nature Communications, 2020, 11, 179. | 5.8 | 32 |
| 5 | Comparison of Mendeliome exome capture kits for use in clinical diagnostics. Scientific Reports, 2020, 10, 3235. | 1.6 | 14 |
| 6 | 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 |
| 7 | Linkage disequilibrium maps to guide contig ordering for genome assembly. Bioinformatics, 2019, 35, 541-545. | 1.8 | 5 |
| 8 | Sequencing era methods for identifying signatures of selection in the genome. Briefings in Bioinformatics, 2019, 20, 1997-2008. | 3.2 | 18 |
| 9 | Linkage disequilibrium maps for European and African populations constructed from whole genome sequence data. Scientific Data, 2019, 6, 208. | 2.4 | 11 |
| 10 | Gene-dense autosomal chromosomes show evidence for increased selection. Heredity, 2019, 123, 774-783. | 1.2 | 3 |
| 11 | 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 |
| 12 | Risk factors for situs defects and congenital heart disease in primary ciliary dyskinesia. Thorax, 2019, 74, 203-205. | 2.7 | 52 |
| 13 | Understanding the disease genome: gene essentiality and the interplay of selection, recombination and mutation. Briefings in Bioinformatics, 2019, 20, 267-273. | 3.2 | 11 |
| 14 | Anticytokine autoantibodies in a patient with a heterozygous NFKB2 mutation. Journal of Allergy and Clinical Immunology, 2018, 141, 1479-1482.e6. | 1.5 | 24 |
| 15 | Clinical efficacy of a nextâ€generation sequencing gene panel for primary immunodeficiency diagnostics. Clinical Genetics, 2018, 93, 647-655. | 1.0 | 63 |
| 16 | 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 |
| 17 | Autosomal dominant tubulointerstitial kidney disease-UMOD is the most frequent non polycystic genetic kidney disease. BMC Nephrology, 2018, 19, 301. | 0.8 | 39 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Long term follow-up of a family with GUCY2D dominant cone dystrophy. International Journal of Ophthalmology, 2018, 11, 1945-1950. | 0.5 | 5 |
| 20 | Evaluating phenotype-driven approaches for genetic diagnoses from exomes in a clinical setting. Scientific Reports, 2017, 7, 13509. | 1.6 | 26 |
| 21 | 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 |
| 22 | Autoimmunity/inflammation in a monogenic primary immunodeficiency cohort. Clinical and Translational Immunology, 2017, 6, e155. | 1.7 | 27 |
| 23 | 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 |
| 24 | Mutations specific to the Rac-GEF domain ofTRIOcause intellectual disability and microcephaly. Journal of Medical Genetics, 2016, 53, 735-742. | 1.5 | 80 |
| 25 | Progressive myoclonic epilepsy with Fanconi syndrome. JRSM Open, 2016, 7, 205427041562314. | 0.2 | 3 |
| 26 | 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 |
| 27 | Precision treatment with sirolimus in a case of activated phosphoinositide 3-kinase Î´ syndrome. Clinical Immunology, 2016, 171, 38-40. | 1.4 | 28 |
| 28 | Deleterious coding variants in multi-case families with non-syndromic cleft lip and/or palate phenotypes. Scientific Reports, 2016, 6, 30457. | 1.6 | 19 |
| 29 | Exome sequencing explained: a practical guide to its clinical application. Briefings in Functional Genomics, 2016, 15, 374-384. | 1.3 | 58 |
| 30 | Collagen (COL4A) mutations are the most frequent mutations underlying adult focal segmental glomerulosclerosis. Nephrology Dialysis Transplantation, 2016, 31, 961-970. | 0.4 | 199 |
| 31 | Aarskog-Scott syndrome: phenotypic and genetic heterogeneity. AIMS Genetics, 2016, 03, 049-059. | 1.9 | 4 |
| 32 | 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 |
| 33 | Whole genome sequences are required to fully resolve the linkage disequilibrium structure of human populations. BMC Genomics, 2015, 16, 666. | 1.2 | 14 |
| 34 | Resolving clinical diagnoses for syndromic cleft lip and/or palate phenotypes using wholeâ€exome sequencing. Clinical Genetics, 2015, 88, 441-449. | 1.0 | 14 |
| 35 | Exome sequence read depth methods for identifying copy number changes. Briefings in Bioinformatics, 2015, 16, 380-392. | 3.2 | 84 |
| 36 | Immuno-Genomic Profiling of Patients with Inflammatory Bowel Disease. Inflammatory Bowel Diseases, 2014, 20, 1813-1819. | 0.9 | 10 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Rapid identification ofSaccharomyces eubayanusand its hybrids. FEMS Yeast Research, 2013, 13, 156-161. | 1.1 | 35 |
| 38 | A SNP profiling panel for sample tracking in whole-exome sequencing studies. Genome Medicine, 2013, 5, 89. | 3.6 | 57 |
| 39 | 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 |