

Joep de Ligt

List of Publications by Year in descending order

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Version: 2024-02-01

56
papers

9,883
citations

101543

36
h-index

155660

55
g-index

63
all docs

63
docs citations

63
times ranked

20396
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|--|------|-----------|
| 1 | Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. <i>New England Journal of Medicine</i> , 2012, 367, 1921-1929. | 27.0 | 1,367 |
| 2 | A Living Biobank of Breast Cancer Organoids Captures Disease Heterogeneity. <i>Cell</i> , 2018, 172, 373-386.e10. | 28.9 | 1,201 |
| 3 | Long-Term Culture of Genome-Stable Bipotent Stem Cells from Adult Human Liver. <i>Cell</i> , 2015, 160, 299-312. | 28.9 | 1,166 |
| 4 | Tissue-specific mutation accumulation in human adult stem cells during life. <i>Nature</i> , 2016, 538, 260-264. | 27.8 | 759 |
| 5 | A de novo paradigm for mental retardation. <i>Nature Genetics</i> , 2010, 42, 1109-1112. | 21.4 | 751 |
| 6 | Long-term expanding human airway organoids for disease modeling. <i>EMBO Journal</i> , 2019, 38, . | 7.8 | 619 |
| 7 | Use of CRISPR-modified human stem cell organoids to study the origin of mutational signatures in cancer. <i>Science</i> , 2017, 358, 234-238. | 12.6 | 337 |
| 8 | A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. <i>Cell</i> , 2014, 159, 200-214. | 28.9 | 322 |
| 9 | Mapping and phasing of structural variation in patient genomes using nanopore sequencing. <i>Nature Communications</i> , 2017, 8, 1326. | 12.8 | 315 |
| 10 | Next-generation genetic testing for retinitis pigmentosa. <i>Human Mutation</i> , 2012, 33, 963-972. | 2.5 | 258 |
| 11 | Genetic dissection of colorectal cancer progression by orthotopic transplantation of engineered cancer organoids. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E2357-E2364. | 7.1 | 198 |
| 12 | Mutations in <i>DYNC1H1</i> cause severe intellectual disability with neuronal migration defects. <i>Journal of Medical Genetics</i> , 2012, 49, 179-183. | 3.2 | 151 |
| 13 | Tracking the international spread of SARS-CoV-2 lineages B.1.1.7 and B.1.351/501Y-V2 with grinch. <i>Wellcome Open Research</i> , 2021, 6, 121. | 1.8 | 129 |
| 14 | Novel genetic causes for cerebral visual impairment. <i>European Journal of Human Genetics</i> , 2016, 24, 660-665. | 2.8 | 127 |
| 15 | NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 303-309. | 6.2 | 125 |
| 16 | An emergent clade of SARS-CoV-2 linked to returned travellers from Iran. <i>Virus Evolution</i> , 2020, 6, veaa027. | 4.9 | 119 |
| 17 | Characteristics of de novo structural changes in the human genome. <i>Genome Research</i> , 2015, 25, 792-801. | 5.5 | 115 |
| 18 | Tracking the international spread of SARS-CoV-2 lineages B.1.1.7 and B.1.351/501Y-V2. <i>Wellcome Open Research</i> , 2021, 6, 121. | 1.8 | 115 |

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|----|--|------|-----------|
| 19 | Non-invasive prenatal diagnosis of fetal aneuploidies using massively parallel sequencing-by-ligation and evidence that cell-free fetal DNA in the maternal plasma originates from cytotrophoblastic cells. <i>Expert Opinion on Biological Therapy</i> , 2012, 12, S19-S26. | 3.1 | 111 |
| 20 | Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. <i>Human Mutation</i> , 2013, 34, 1439-1448. | 2.5 | 105 |
| 21 | Genomic epidemiology reveals transmission patterns and dynamics of SARS-CoV-2 in Aotearoa New Zealand. <i>Nature Communications</i> , 2020, 11, 6351. | 12.8 | 100 |
| 22 | A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. <i>Nature Communications</i> , 2016, 7, 12989. | 12.8 | 99 |
| 23 | Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. <i>Journal of Medical Genetics</i> , 2013, 50, 802-811. | 3.2 | 93 |
| 24 | Mobster: accurate detection of mobile element insertions in next generation sequencing data. <i>Genome Biology</i> , 2014, 15, 488. | 8.8 | 86 |
| 25 | Genomic landscape of rat strain and substrain variation. <i>BMC Genomics</i> , 2015, 16, 357. | 2.8 | 84 |
| 26 | Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 during Border Quarantine and Air Travel, New Zealand (Aotearoa). <i>Emerging Infectious Diseases</i> , 2021, 27, 1274-1278. | 4.3 | 68 |
| 27 | Deficiency of nucleotide excision repair is associated with mutational signature observed in cancer. <i>Genome Research</i> , 2019, 29, 1067-1077. | 5.5 | 66 |
| 28 | <i>GATAD2B</i> loss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth in <i>Drosophila</i> . <i>Journal of Medical Genetics</i> , 2013, 50, 507-514. | 3.2 | 63 |
| 29 | Heterozygous germline mutations in <i>A2ML1</i> are associated with a disorder clinically related to Noonan syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 317-324. | 2.8 | 61 |
| 30 | Mutations Affecting the SAND Domain of <i>DEAF1</i> Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. <i>American Journal of Human Genetics</i> , 2014, 94, 649-661. | 6.2 | 59 |
| 31 | Novel mutations in <i>LRP6</i> highlight the role of WNT signaling in tooth agenesis. <i>Genetics in Medicine</i> , 2016, 18, 1158-1162. | 2.4 | 58 |
| 32 | Cancer cells copy migratory behavior and exchange signaling networks via extracellular vesicles. <i>EMBO Journal</i> , 2018, 37, . | 7.8 | 58 |
| 33 | Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. <i>European Journal of Human Genetics</i> , 2015, 23, 1142-1150. | 2.8 | 56 |
| 34 | Allelic Mutations of <i>KITLG</i> , Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. <i>American Journal of Human Genetics</i> , 2015, 97, 647-660. | 6.2 | 55 |
| 35 | Toward effective software solutions for big biology. <i>Nature Biotechnology</i> , 2015, 33, 686-687. | 17.5 | 46 |
| 36 | Point mutations as a source of de novo genetic disease. <i>Current Opinion in Genetics and Development</i> , 2013, 23, 257-263. | 3.3 | 44 |

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|----|--|------|-----------|
| 37 | Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2017, 25, 1246-1252. | 2.8 | 34 |
| 38 | Use of Genomics to Track Coronavirus Disease Outbreaks, New Zealand. <i>Emerging Infectious Diseases</i> , 2021, 27, 1317-1322. | 4.3 | 28 |
| 39 | Real-Time Genomics for Tracking Severe Acute Respiratory Syndrome Coronavirus 2 Border Incursions after Virus Elimination, New Zealand. <i>Emerging Infectious Diseases</i> , 2021, 27, 2361-2368. | 4.3 | 27 |
| 40 | The molecular genetic make-up of male breast cancer. <i>Endocrine-Related Cancer</i> , 2019, 26, 779-794. | 3.1 | 27 |
| 41 | Molecular dissection of germline chromothripsis in a developmental context using patient-derived iPSC cells. <i>Genome Medicine</i> , 2017, 9, 9. | 8.2 | 25 |
| 42 | Scalable Workflows and Reproducible Data Analysis for Genomics. <i>Methods in Molecular Biology</i> , 2019, 1910, 723-745. | 0.9 | 25 |
| 43 | Identification of novel human Wnt target genes using adult endodermal tissue-derived organoids. <i>Developmental Biology</i> , 2021, 474, 37-47. | 2.0 | 23 |
| 44 | Resolving the Breakpoints of the 17q21.31 Microdeletion Syndrome with Next-Generation Sequencing. <i>American Journal of Human Genetics</i> , 2012, 90, 599-613. | 6.2 | 22 |
| 45 | Airborne Transmission of SARS-CoV-2 Delta Variant within Tightly Monitored Isolation Facility, New Zealand (Aotearoa). <i>Emerging Infectious Diseases</i> , 2022, 28, 501-509. | 4.3 | 21 |
| 46 | Genomic epidemiology of Delta SARS-CoV-2 during transition from elimination to suppression in Aotearoa New Zealand. <i>Nature Communications</i> , 2022, 13, . | 12.8 | 17 |
| 47 | Reliable noninvasive prenatal testing by massively parallel sequencing of circulating cell-free DNA from maternal plasma processed up to 24h after venipuncture. <i>Clinical Biochemistry</i> , 2013, 46, 1783-1786. | 1.9 | 15 |
| 48 | Cerebral visual impairment and intellectual disability caused by PGAP1 variants. <i>European Journal of Human Genetics</i> , 2015, 23, 1689-1693. | 2.8 | 15 |
| 49 | COVID-19 vaccine strategies for Aotearoa New Zealand: a mathematical modelling study. <i>The Lancet Regional Health - Western Pacific</i> , 2021, 15, 100256. | 2.9 | 15 |
| 50 | Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. <i>Genomics Data</i> , 2014, 2, 144-146. | 1.3 | 13 |
| 51 | A System-wide Approach to Monitor Responses to Synergistic BRAF and EGFR Inhibition in Colorectal Cancer Cells. <i>Molecular and Cellular Proteomics</i> , 2018, 17, 1892-1908. | 3.8 | 13 |
| 52 | Natural helix 9 mutants of PPAR γ differently affect its transcriptional activity. <i>Molecular Metabolism</i> , 2019, 20, 115-127. | 6.5 | 12 |
| 53 | A Single Complex Agpat2 Allele in a Patient With Partial Lipodystrophy. <i>Frontiers in Physiology</i> , 2018, 9, 1363. | 2.8 | 7 |
| 54 | The Genomic Scrapheap Challenge; Extracting Relevant Data from Unmapped Whole Genome Sequencing Reads, Including Strain Specific Genomic Segments, in Rats. <i>PLoS ONE</i> , 2016, 11, e0160036. | 2.5 | 5 |

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|----|---|-----|-----------|
| 55 | Detecting fetal subchromosomal aberrations by MPS: an unexpected discrepancy between amniocyte DNA and ccffDNA. <i>Prenatal Diagnosis</i> , 2014, 34, 402-405. | 2.3 | 2 |
| 56 | Exome Sequencing of Late Recurrence T-Cell Acute Lymphoblastic Leukemia in Children Confirms Second Leukemia and Exposes Predisposition Candidate Genes. <i>Blood</i> , 2011, 118, 755-755. | 1.4 | 0 |