Joep de Ligt

List of Publications by Year in descending order

56 papers	9,883 citations	101384 36 h-index	155451 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. New England Journal of Medicine, 2012, 367, 1921-1929.	13.9	1,367
2	A Living Biobank of Breast Cancer Organoids Captures Disease Heterogeneity. Cell, 2018, 172, 373-386.e10.	13.5	1,201
3	Long-Term Culture of Genome-Stable Bipotent Stem Cells from Adult Human Liver. Cell, 2015, 160, 299-312.	13.5	1,166
4	Tissue-specific mutation accumulation in human adult stem cells during life. Nature, 2016, 538, 260-264.	13.7	759
5	A de novo paradigm for mental retardation. Nature Genetics, 2010, 42, 1109-1112.	9.4	751
6	Longâ€term expanding human airway organoids for disease modeling. EMBO Journal, 2019, 38, .	3.5	619
7	Use of CRISPR-modified human stem cell organoids to study the origin of mutational signatures in cancer. Science, 2017, 358, 234-238.	6.0	337
8	A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214.	13.5	322
9	Mapping and phasing of structural variation in patient genomes using nanopore sequencing. Nature Communications, 2017, 8, 1326.	5.8	315
10	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	1.1	258
11	Genetic dissection of colorectal cancer progression by orthotopic transplantation of engineered cancer organoids. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E2357-E2364.	3.3	198
12	Mutations inDYNC1H1cause severe intellectual disability with neuronal migration defects. Journal of Medical Genetics, 2012, 49, 179-183.	1.5	151
13	Tracking the international spread of SARS-CoV-2 lineages B.1.1.7 and B.1.351/501Y-V2 with grinch. Wellcome Open Research, 2021, 6, 121.	0.9	129
14	Novel genetic causes for cerebral visual impairment. European Journal of Human Genetics, 2016, 24, 660-665.	1.4	127
15	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	2.6	125
16	An emergent clade of SARS-CoV-2 linked to returned travellers from Iran. Virus Evolution, 2020, 6, veaa027.	2.2	119
17	Characteristics of de novo structural changes in the human genome. Genome Research, 2015, 25, 792-801.	2.4	115
18	Tracking the international spread of SARS-CoV-2 lineages B.1.1.7 and B.1.351/501Y-V2. Wellcome Open Research, 2021, 6, 121.	0.9	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. Expert Opinion on Biological Therapy, 2012, 12, S19-S26.	1.4	111
20	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. Human Mutation, 2013, 34, 1439-1448.	1.1	105
21	Genomic epidemiology reveals transmission patterns and dynamics of SARS-CoV-2 in Aotearoa New Zealand. Nature Communications, 2020, 11, 6351.	5.8	100
22	A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. Nature Communications, 2016, 7, 12989.	5.8	99
23	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. Journal of Medical Genetics, 2013, 50, 802-811.	1.5	93
24	Mobster: accurate detection of mobile element insertions in next generation sequencing data. Genome Biology, 2014, 15, 488.	3.8	86
25	Genomic landscape of rat strain and substrain variation. BMC Genomics, 2015, 16, 357.	1.2	84
26	Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 during Border Quarantine and Air Travel, New Zealand (Aotearoa). Emerging Infectious Diseases, 2021, 27, 1274-1278.	2.0	68
27	Deficiency of nucleotide excision repair is associated with mutational signature observed in cancer. Genome Research, 2019, 29, 1067-1077.	2.4	66
28	GATAD2Bloss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth inDrosophila. Journal of Medical Genetics, 2013, 50, 507-514.	1.5	63
29	Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome. European Journal of Human Genetics, 2015, 23, 317-324.	1.4	61
30	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. American Journal of Human Genetics, 2014, 94, 649-661.	2.6	59
31	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	1.1	58
32	Cancer cells copy migratory behavior and exchange signaling networks via extracellular vesicles. EMBO Journal, 2018, 37, .	3.5	58
33	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. European Journal of Human Genetics, 2015, 23, 1142-1150.	1.4	56
34	Allelic Mutations of KITLG, Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. American Journal of Human Genetics, 2015, 97, 647-660.	2.6	55
35	Toward effective software solutions for big biology. Nature Biotechnology, 2015, 33, 686-687.	9.4	46
36	Point mutations as a source of de novo genetic disease. Current Opinion in Genetics and Development, 2013, 23, 257-263.	1.5	44

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37	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 1246-1252.	1.4	34
38	Use of Genomics to Track Coronavirus Disease Outbreaks, New Zealand. Emerging Infectious Diseases, 2021, 27, 1317-1322.	2.0	28
39	Real-Time Genomics for Tracking Severe Acute Respiratory Syndrome Coronavirus 2 Border Incursions after Virus Elimination, New Zealand. Emerging Infectious Diseases, 2021, 27, 2361-2368.	2.0	27
40	The molecular genetic make-up of male breast cancer. Endocrine-Related Cancer, 2019, 26, 779-794.	1.6	27
41	Molecular dissection of germline chromothripsis in a developmental context using patient-derived iPS cells. Genome Medicine, 2017, 9, 9.	3.6	25
42	Scalable Workflows and Reproducible Data Analysis for Genomics. Methods in Molecular Biology, 2019, 1910, 723-745.	0.4	25
43	Identification of novel human Wnt target genes using adult endodermal tissue-derived organoids. Developmental Biology, 2021, 474, 37-47.	0.9	23
44	Resolving the Breakpoints of the 17q21.31 Microdeletion Syndrome with Next-Generation Sequencing. American Journal of Human Genetics, 2012, 90, 599-613.	2.6	22
45	Airborne Transmission of SARS-CoV-2 Delta Variant within Tightly Monitored Isolation Facility, New Zealand (Aotearoa). Emerging Infectious Diseases, 2022, 28, 501-509.	2.0	21
46	Genomic epidemiology of Delta SARS-CoV-2 during transition from elimination to suppression in Aotearoa New Zealand. Nature Communications, 2022, 13, .	5.8	17
47	Reliable noninvasive prenatal testing by massively parallel sequencing of circulating cell-free DNA from maternal plasma processed up to 24h after venipuncture. Clinical Biochemistry, 2013, 46, 1783-1786.	0.8	15
48	Cerebral visual impairment and intellectual disability caused by PGAP1 variants. European Journal of Human Genetics, 2015, 23, 1689-1693.	1.4	15
49	COVID-19 vaccine strategies for Aotearoa New Zealand: a mathematical modelling study. The Lancet Regional Health - Western Pacific, 2021, 15, 100256.	1.3	15
50	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. Genomics Data, 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. Molecular and Cellular Proteomics, 2018, 17, 1892-1908.	2.5	13
52	Natural helix 9 mutants of PPARÎ³ differently affect its transcriptional activity. Molecular Metabolism, 2019, 20, 115-127.	3.0	12
53	A Single Complex Agpat2 Allele in a Patient With Partial Lipodystrophy. Frontiers in Physiology, 2018, 9, 1363.	1.3	7
54	The Genomic Scrapheap Challenge; Extracting Relevant Data from Unmapped Whole Genome Sequencing Reads, Including Strain Specific Genomic Segments, in Rats. PLoS ONE, 2016, 11, e0160036.	1.1	5

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55	Detecting fetal subchromosomal aberrations by MPS: an unexpected discrepancy between amniocyte DNA and ccffDNA. Prenatal Diagnosis, 2014, 34, 402-405.	1.1	2
56	Exome Sequencing of Late Recurrence T-Cell Acute Lymphoblastic Leukemia in Children Confirms Second Leukemia and Exposes Predisposition Candidate Genes. Blood, 2011, 118, 755-755.	0.6	0