

Laurent C Francioli

List of Publications by Year in descending order

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Version: 2024-02-01

30
papers

10,626
citations

257357

24
h-index

395590

33
g-index

39
all docs

39
docs citations

39
times ranked

23355
citing authors

#	ARTICLE	IF	CITATIONS
1	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	1.1	44
2	Implementation of hand hygiene in health-care facilities: results from the WHO Hand Hygiene Self-Assessment Framework global survey 2019. <i>Lancet Infectious Diseases</i> , The, 2022, 22, 835-844.	4.6	29
3	Rare coding variants in ten genes confer substantial risk for schizophrenia. <i>Nature</i> , 2022, 604, 509-516.	13.7	326
4	Mapping the human genetic architecture of COVID-19. <i>Nature</i> , 2021, 600, 472-477.	13.7	640
5	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2021, 597, E3-E4.	13.7	45
6	Recurrent <i>TTN</i> metatranscriptome-only c.39974A>G splice variant associated with autosomal recessive arthrogyposis multiplex congenita and myopathy. <i>Human Mutation</i> , 2020, 41, 403-411.	1.1	28
7	Evaluating drug targets through human loss-of-function genetic variation. <i>Nature</i> , 2020, 581, 459-464.	13.7	115
8	The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2020, 581, 434-443.	13.7	6,140
9	Characterising the loss-of-function impact of 5' UTR untranslated region variants in 15,708 individuals. <i>Nature Communications</i> , 2020, 11, 2523.	5.8	99
10	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. <i>Nature Communications</i> , 2020, 11, 2539.	5.8	98
11	A structural variation reference for medical and population genetics. <i>Nature</i> , 2020, 581, 444-451.	13.7	614
12	Transcript expression-aware annotation improves rare variant interpretation. <i>Nature</i> , 2020, 581, 452-458.	13.7	142
13	The effect of LRRK2 loss-of-function variants in humans. <i>Nature Medicine</i> , 2020, 26, 869-877.	15.2	79
14	Disparities in discovery of pathogenic variants for autosomal recessive non-syndromic hearing impairment by ancestry. <i>European Journal of Human Genetics</i> , 2019, 27, 1456-1465.	1.4	19
15	novoCaller: a Bayesian network approach for <i>de novo</i> variant calling from pedigree and population sequence data. <i>Bioinformatics</i> , 2019, 35, 1174-1180.	1.8	5
16	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. <i>Brain</i> , 2019, 142, 35-49.	3.7	44
17	Negative selection in humans and fruit flies involves synergistic epistasis. <i>Science</i> , 2017, 356, 539-542.	6.0	103
18	The role of <i>de novo</i> mutations in the development of amyotrophic lateral sclerosis. <i>Human Mutation</i> , 2017, 38, 1534-1541.	1.1	13

#	ARTICLE	IF	CITATIONS
19	Human genetic variation alters CRISPR-Cas9 on- and off-targeting specificity at therapeutically implicated loci. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E11257-E11266.	3.3	96
20	A framework for the detection of de novo mutations in family-based sequencing data. <i>European Journal of Human Genetics</i> , 2017, 25, 227-233.	1.4	29
21	A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. <i>Nature Communications</i> , 2016, 7, 12989.	5.8	99
22	Leveraging Distant Relatedness to Quantify Human Mutation and Gene-Conversion Rates. <i>American Journal of Human Genetics</i> , 2015, 97, 775-789.	2.6	77
23	Characteristics of de novo structural changes in the human genome. <i>Genome Research</i> , 2015, 25, 792-801.	2.4	115
24	Genome-wide patterns and properties of de novo mutations in humans. <i>Nature Genetics</i> , 2015, 47, 822-826.	9.4	384
25	Genome of the Netherlands population-specific imputations identify an ABCA6 variant associated with cholesterol levels. <i>Nature Communications</i> , 2015, 6, 6065.	5.8	45
26	Improved imputation quality of low-frequency and rare variants in European samples using the "Genome of The Netherlands"™. <i>European Journal of Human Genetics</i> , 2014, 22, 1321-1326.	1.4	92
27	A genome-wide association study identifies a functional ERAP2 haplotype associated with birdshot chorioretinopathy. <i>Human Molecular Genetics</i> , 2014, 23, 6081-6087.	1.4	115
28	The Genome of the Netherlands: design, and project goals. <i>European Journal of Human Genetics</i> , 2014, 22, 221-227.	1.4	246
29	Deleterious Alleles in the Human Genome Are on Average Younger Than Neutral Alleles of the Same Frequency. <i>PLoS Genetics</i> , 2013, 9, e1003301.	1.5	63
30	A Numeric Model to Simulate Solar Individual Ultraviolet Exposure. <i>Photochemistry and Photobiology</i> , 2011, 87, 721-728.	1.3	33