Laurent C Francioli

List of Publications by Year
in descending order

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30 papers	10,626 citations	257450 24 h-index	395702 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. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
2	Implementation of hand hygiene in health-care facilities: results from the WHO Hand Hygiene Self-Assessment Framework global survey 2019. Lancet Infectious Diseases, The, 2022, 22, 835-844.	9.1	29
3	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	27.8	326
4	Mapping the human genetic architecture of COVID-19. Nature, 2021, 600, 472-477.	27.8	640
5	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	27.8	45
6	Recurrent TTN metatranscriptâ€onlyc.39974â€“11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. Human Mutation, 2020, 41, 403-411.	2.5	28
7	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	27.8	115
8	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
9	Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523.	12.8	99
10	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. Nature Communications, 2020, 11, 2539.	12.8	98
11	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	27.8	614
12	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	27.8	142
13	The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877.	30.7	79
14	Disparities in discovery of pathogenic variants for autosomal recessive non-syndromic hearing impairment by ancestry. European Journal of Human Genetics, 2019, 27, 1456-1465.	2.8	19
15	novoCaller: a Bayesian network approach for de novo variant calling from pedigree and population sequence data. Bioinformatics, 2019, 35, 1174-1180.	4.1	5
16	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. Brain, 2019, 142, 35-49.	7.6	44
17	Negative selection in humans and fruit flies involves synergistic epistasis. Science, 2017, 356, 539-542.	12.6	103
18	The role of de novo mutations in the development of amyotrophic lateral sclerosis. Human Mutation, 2017, 38, 1534-1541.	2.5	13

#	Article	IF	Citations
19	Human genetic variation alters CRISPR-Cas9 on- and off-targeting specificity at therapeutically implicated loci. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E11257-E11266.	7.1	96
20	A framework for the detection of de novo mutations in family-based sequencing data. European Journal of Human Genetics, 2017, 25, 227-233.	2.8	29
21	A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. Nature Communications, 2016, 7, 12989.	12.8	99
22	Leveraging Distant Relatedness to Quantify Human Mutation and Gene-Conversion Rates. American Journal of Human Genetics, 2015, 97, 775-789.	6.2	77
23	Characteristics of de novo structural changes in the human genome. Genome Research, 2015, 25, 792-801.	5.5	115
24	Genome-wide patterns and properties of de novo mutations in humans. Nature Genetics, 2015, 47, 822-826.	21.4	384
25	Genome of the Netherlands population-specific imputations identify an ABCA6 variant associated with cholesterol levels. Nature Communications, 2015, 6, 6065.	12.8	45
26	Improved imputation quality of low-frequency and rare variants in European samples using the â€˜Genome of The Netherlandsâ€™. European Journal of Human Genetics, 2014, 22, 1321-1326.	2.8	92
27	A genome-wide association study identifies a functional ERAP2 haplotype associated with birdshot chorioretinopathy. Human Molecular Genetics, 2014, 23, 6081-6087.	2.9	115
28	The Genome of the Netherlands: design, and project goals. European Journal of Human Genetics, 2014, 22, 221-227.	2.8	246
29	Deleterious Alleles in the Human Genome Are on Average Younger Than Neutral Alleles of the Same Frequency. PLoS Genetics, 2013, 9, e1003301.	3.5	63
30	A Numeric Model to Simulate Solar Individual Ultraviolet Exposure. Photochemistry and Photobiology, 2011, 87, 721-728.	2.5	33