Laurent C Francioli

List of Publications by Year
in descending order

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	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
2	Mapping the human genetic architecture of COVID-19. Nature, 2021, 600, 472-477.	13.7	640
3	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	13.7	614
4	Genome-wide patterns and properties of de novo mutations in humans. Nature Genetics, 2015, 47, 822-826.	9.4	384
5	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	13.7	326
6	The Genome of the Netherlands: design, and project goals. European Journal of Human Genetics, 2014, 22, 221-227.	1.4	246
7	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	13.7	142
8	A genome-wide association study identifies a functional ERAP2 haplotype associated with birdshot chorioretinopathy. Human Molecular Genetics, 2014, 23, 6081-6087.	1.4	115
9	Characteristics of de novo structural changes in the human genome. Genome Research, 2015, 25, 792-801.	2.4	115
10	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	13.7	115
11	Negative selection in humans and fruit flies involves synergistic epistasis. Science, 2017, 356, 539-542.	6.0	103
12	A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. Nature Communications, 2016, 7, 12989.	5.8	99
13	Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523.	5.8	99
14	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. Nature Communications, 2020, 11, 2539.	5.8	98
15	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.	3.3	96
16	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.	1.4	92
17	The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877.	15.2	79
18	Leveraging Distant Relatedness to Quantify Human Mutation and Gene-Conversion Rates. American Journal of Human Genetics, 2015, 97, 775-789.	2.6	77

#	Article	IF	Citations
19	Deleterious Alleles in the Human Genome Are on Average Younger Than Neutral Alleles of the Same Frequency. PLoS Genetics, 2013, 9, e1003301.	1.5	63
20	Genome of the Netherlands population-specific imputations identify an ABCA6 variant associated with cholesterol levels. Nature Communications, 2015, 6, 6065.	5.8	45
21	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
22	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. Brain, 2019, 142, 35-49.	3.7	44
23	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
24	A Numeric Model to Simulate Solar Individual Ultraviolet Exposure. Photochemistry and Photobiology, 2011, 87, 721-728.	1.3	33
25	A framework for the detection of de novo mutations in family-based sequencing data. European Journal of Human Genetics, 2017, 25, 227-233.	1.4	29
26	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.	4.6	29
27	Recurrent TTN metatranscriptâ€•only c.39974â€“11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. Human Mutation, 2020, 41, 403-411.	1.1	28
28	Disparities in discovery of pathogenic variants for autosomal recessive non-syndromic hearing impairment by ancestry. European Journal of Human Genetics, 2019, 27, 1456-1465.	1.4	19
29	The role of de novo mutations in the development of amyotrophic lateral sclerosis. Human Mutation, 2017, 38, 1534-1541.	1.1	13
30	novoCaller: a Bayesian network approach for de novo variant calling from pedigree and population sequence data. Bioinformatics, 2019, 35, 1174-1180.	1.8	5