Lifeng Tian

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/8081558/publications.pdf

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		516710	265206
47	1,931	16	42
papers	citations	h-index	g-index

51	51	51	5034
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Low concordance of multiple variant-calling pipelines: practical implications for exome and genome sequencing. Genome Medicine, 2013, 5, 28.	8.2	381
2	Analysis of 589,306 genomes identifies individuals resilient to severe Mendelian childhood diseases. Nature Biotechnology, 2016, 34, 531-538.	17.5	273
3	Genetic predisposition to neuroblastoma mediated by a LMO1 super-enhancer polymorphism. Nature, 2015, 528, 418-421.	27.8	263
4	GRIN2D Recurrent De Novo Dominant Mutation Causes a Severe Epileptic Encephalopathy Treatable with NMDA Receptor Channel Blockers. American Journal of Human Genetics, 2016, 99, 802-816.	6.2	138
5	ARAF recurrent mutation causes central conducting lymphatic anomaly treatable with a MEK inhibitor. Nature Medicine, 2019, 25, 1116-1122.	30.7	136
6	Inherited bone marrow failure associated with germline mutation of ACD, the gene encoding telomere protein TPP1. Blood, 2014, 124, 2767-2774.	1.4	97
7	Pathogenic variant in EPHB4 results in central conducting lymphatic anomaly. Human Molecular Genetics, 2018, 27, 3233-3245.	2.9	73
8	Transcriptome of the human retina, retinal pigmented epithelium and choroid. Genomics, 2015, 105, 253-264.	2.9	71
9	Mutation in IRF2BP2 is responsible for a familial form of common variable immunodeficiency disorder. Journal of Allergy and Clinical Immunology, 2016, 138, 544-550.e4.	2.9	54
10	Exome Sequencing Reveals Mutations in AIRE as a Cause of Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1726-1733.	3.6	35
11	Expanding the phenotype of PRPS1 syndromes in females: neuropathy, hearing loss and retinopathy. Orphanet Journal of Rare Diseases, 2014, 9, 190.	2.7	31
12	Combining targeted panel-based resequencing and copy-number variation analysis for the diagnosis of inherited syndromic retinopathies and associated ciliopathies. Scientific Reports, 2018, 8, 5285.	3.3	28
13	Expression Pattern of the SARS-CoV-2 Entry Genes ACE2 and TMPRSS2 in the Respiratory Tract. Viruses, 2020, 12, 1174.	3.3	27
14	Expanding the SPECC1L mutation phenotypic spectrum to include Teebi hypertelorism syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2497-2502.	1.2	26
15	A genome-wide association study of anorexia nervosa suggests a risk locus implicated in dysregulated leptin signaling. Scientific Reports, 2017, 7, 3847.	3.3	23
16	Application of Whole Exome Sequencing in Six Families with an Initial Diagnosis of Autosomal Dominant Retinitis Pigmentosa: Lessons Learned. PLoS ONE, 2015, 10, e0133624.	2.5	19
17	An integrative approach to investigate the respective roles of single-nucleotide variants and copy-number variants in Attention-Deficit/Hyperactivity Disorder. Scientific Reports, 2016, 6, 22851.	3.3	18
18	Bayesian analysis of genome-wide inflammatory bowel disease data sets reveals new risk loci. European Journal of Human Genetics, 2018, 26, 265-274.	2.8	17

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19	Delayed diagnosis of congenital myasthenia due to associated mitochondrial enzyme defect. Neuromuscular Disorders, 2015, 25, 257-261.	0.6	16
20	Serum levels of the IgA isotype switch factor TGFâ€î²1 are elevated in patients with COVIDâ€19. FEBS Letters, 2021, 595, 1819-1824.	2.8	16
21	Association of Rare Recurrent Copy Number Variants With Congenital Heart Defects Based on Next-Generation Sequencing Data From Family Trios. Frontiers in Genetics, 2019, 10, 819.	2.3	15
22	Application of deep learning algorithm on whole genome sequencing data uncovers structural variants associated with multiple mental disorders in African American patients. Molecular Psychiatry, 2022, 27, 1469-1478.	7.9	13
23	Advantage of Whole Exome Sequencing over Allele-Specific and Targeted Segment Sequencing in Detection of NovelTULP1Mutation in Leber Congenital Amaurosis. Ophthalmic Genetics, 2015, 36, 333-338.	1.2	12
24	An atypical form of AOA2 with myoclonus associated with mutations in SETX and AFG3L2. BMC Medical Genetics, 2015, 16, 16.	2.1	12
25	Association of a rare NOTCH4 coding variant with systemic sclerosis: a family-based whole exome sequencing study. BMC Musculoskeletal Disorders, 2016, 17, 462.	1.9	12
26	Application of ACMG criteria to classify variants in the human gene mutation database. Journal of Human Genetics, 2019, 64, 1091-1095.	2.3	10
27	Rare Recurrent Variants in Noncoding Regions Impact Attention-Deficit Hyperactivity Disorder (ADHD) Gene Networks in Children of both African American and European American Ancestry. Genes, 2021, 12, 310.	2.4	10
28	Expanding the Genetic Landscape of Usher-Like Phenotypes. , 2019, 60, 4701.		9
29	Increasing diagnostic yield by RNA-Sequencing in rare diseaseâ€"bypass hurdles of interpreting intronic or splice-altering variants. Annals of Translational Medicine, 2018, 6, 126-126.	1.7	9
30	Expansion of Schizophrenia Gene Network Knowledge Using Machine Learning Selected Signals From Dorsolateral Prefrontal Cortex and Amygdala RNA-seq Data. Frontiers in Psychiatry, 2022, 13, 797329.	2.6	9
31	Loss-of-Function Mutations in KIF15 Underlying a Braddock-Carey Genocopy. Human Mutation, 2017, 38, 507-510.	2.5	8
32	COVID-19: Look to the Future, Learn from the Past. Viruses, 2020, 12, 1226.	3.3	8
33	Deep learning prediction of attention-deficit hyperactivity disorder in African Americans by copy number variation. Experimental Biology and Medicine, 2021, 246, 2317-2323.	2.4	8
34	Insights into non-autoimmune type 1 diabetes with 13 novel loci in low polygenic risk score patients. Scientific Reports, 2021, 11, 16013.	3.3	7
35	Early Infantile Epileptic Encephalopathy in anSTXBP1Patient with Lactic Acidemia and Normal Mitochondrial Respiratory Chain Function. Case Reports in Genetics, 2016, 2016, 1-5.	0.2	6
36	Heterozygous Deletion Impacting SMARCAD1 in the Original Kindred with Absent Dermatoglyphs and Associated Features (Baird, 1964). Journal of Pediatrics, 2018, 194, 248-252.e2.	1.8	6

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37	Association of DLL1 with type 1 diabetes in patients characterized by low polygenic risk score. Metabolism: Clinical and Experimental, 2021, 114, 154418.	3.4	6
38	<i>FLNC</i> and <i>MYLK2</i> Gene Mutations in a Chinese Family with Different Phenotypes of Cardiomyopathy. International Heart Journal, 2021, 62, 127-134.	1.0	6
39	Non-coding structural variation differentially impacts attention-deficit hyperactivity disorder (ADHD) gene networks in African American vs Caucasian children. Scientific Reports, 2020, 10, 15252.	3.3	5
40	Machine Learning Reduced Gene/Non-Coding RNA Features That Classify Schizophrenia Patients Accurately and Highlight Insightful Gene Clusters. International Journal of Molecular Sciences, 2021, 22, 3364.	4.1	4
41	Mutation burden analysis of six common mental disorders in African Americans by whole genome sequencing. Human Molecular Genetics, 2022, 31, 3769-3776.	2.9	4
42	Role of the ADCY9 gene in cardiac abnormalities of the Rubinstein-Taybi syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 101.	2.7	2
43	The Infection Rate of COVID-19 in Wuhan, China: Combined Analysis of Population Samples. Journal of Medical Internet Research, 2020, 22, e20914.	4.3	2
44	Interpretation of Maturity-Onset Diabetes of the Young Genetic Variants Based on American College of Medical Genetics and Genomics Criteria: Machine-Learning Model Development. JMIR Biomedical Engineering, 2020, 5, e20506.	1.2	2
45	High prevalence of elevated serum liver enzymes in Chinese children suggests metabolic syndrome as a common risk factor. Journal of Paediatrics and Child Health, 2020, 56, 1590-1596.	0.8	1
46	Burden of rare coding variants reveals genetic heterogeneity between obese and non-obese asthma patients in the African American population. Respiratory Research, 2022, 23, 116.	3.6	1
47	An Acquired Mutation in Deubiquitinating Enzyme USP11 Associated with Clonal Hematopoiesis in Diamond Blackfan Anemia. Blood, 2014, 124, 1596-1596.	1.4	0