Aniko Sabo

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7021676/publications.pdf

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40	26,557	257357	289141
papers	citations	24 h-index	40 g-index

41	41	41	50393
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
2	Somatic mutations affect key pathways in lung adenocarcinoma. Nature, 2008, 455, 1069-1075.	13.7	2,694
3	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	13.7	2,254
4	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	13.7	1,597
5	Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. Science, 2007, 316, 222-234.	6.0	1,283
6	The Genome Sequence of Taurine Cattle: A Window to Ruminant Biology and Evolution. Science, 2009, 324, 522-528.	6.0	1,038
7	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	9.4	943
8	Identification of genes subject to positive selection in uropathogenic strains of Escherichia coli: A comparative genomics approach. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 5977-5982.	3.3	509
9	Comparison of genome degradation in Paratyphi A and Typhi, human-restricted serovars of Salmonella enterica that cause typhoid. Nature Genetics, 2004, 36, 1268-1274.	9.4	367
10	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493.	3.8	265
11	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	3.8	242
12	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	1.7	163
13	Assessing structural variation in a personal genomeâ€"towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	1.2	153
14	Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorders. Human Molecular Genetics, 2011, 20, 3366-3375.	1.4	149
15	Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. PLoS Genetics, 2013, 9, e1003443.	1.5	133
16	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	2.6	112
17	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	15.2	103
18	Generation and annotation of the DNA sequences of human chromosomes 2 and 4. Nature, 2005, 434, 724-731.	13.7	85

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19	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	2.6	80
20	Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. Hepatology, 2019, 70, 899-910.	3.6	58
21	Germline Cancer Predisposition Variants inâ€, Pediatric Rhabdomyosarcoma: A Report From the Childrenâ€™s Oncology Group. Journal of the National Cancer Institute, 2021, 113, 875-883.	3.0	55
22	Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250.	2.6	36
23	Meta-analysis of 49â€…549 individuals imputed with the 1000 Genomes Project reveals an exonic damaging variant inANGPTL4determining fasting TG levels. Journal of Medical Genetics, 2016, 53, 441-449.	1.5	34
24	Combination of whole exome sequencing and animal modeling identifies TMPRSS9 as a candidate gene for autism spectrum disorder. Human Molecular Genetics, 2020, 29, 459-470.	1.4	32
25	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	2.6	30
26	Whole Exome Sequencing Identifies Novel Genes for Fetal Hemoglobin Response to Hydroxyurea in Children with Sickle Cell Anemia. PLoS ONE, 2014, 9, e110740.	1.1	28
27	EAnnot: A genome annotation tool using experimental evidence. Genome Research, 2004, 14, 2503-2509.	2.4	18
28	Xiaâ€“Gibbs syndrome in adulthood: a case report with insight into the natural history of the condition. Journal of Physical Education and Sports Management, 2019, 5, a003608.	0.5	15
29	PCR detection of nearly any dengue virus strain using a highly sensitive primer â€˜cocktailâ€™. FEBS Journal, 2011, 278, 1676-1687.	2.2	14
30	Phenotypic and protein localization heterogeneity associated with AHDC1 pathogenic proteinâ€truncating alleles in Xiaâ€“Gibbs syndrome. Human Mutation, 2021, 42, 577-591.	1.1	14
31	Genetic testing in ambulatory cardiology clinics reveals high rate of findings with clinical management implications. Genetics in Medicine, 2021, 23, 2404-2414.	1.1	14
32	Exome sequencing reveals novel genetic loci influencing obesityâ€related traits in Hispanic children. Obesity, 2017, 25, 1270-1276.	1.5	10
33	Fine mapping the CETP region reveals a common intronic insertion associated to HDL-C. Npj Aging and Mechanisms of Disease, 2015, 1, 15011.	4.5	8
34	Communityâ€based recruitment and exome sequencing indicates high diagnostic yield in adults with intellectual disability. Molecular Genetics & Genomic Medicine, 2020, 8, e1439.	0.6	6
35	Exome sequencing in children with clinically suspected <scp>maturityâ€onset</scp> diabetes of the young. Pediatric Diabetes, 2021, 22, 960-968.	1.2	6
36	Genetic Predictors of Hemoglobin F Response to Hydroxyurea in Sickle Cell Anemia. Blood, 2012, 120, 241-241.	0.6	5

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37	Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846.	1.1	3
38	A t(5;16) translocation is the likely driver of a syndrome with ambiguous genitalia, facial dysmorphism, intellectual disability, and speech delay. Journal of Physical Education and Sports Management, 2016, 2, a000703.	0.5	1
39	Sequencing of a central nervous system tumor demonstrates cancer transmission in an organ transplant. Life Science Alliance, 2021, 4, e202000941.	1.3	1
40	FOXO3 Variants Are Associated With Lower Fetal Hemoglobin Levels In Children With Sickle Cell Disease. Blood, 2013, 122, 778-778.	0.6	1