

# Aniko Sabo

## List of Publications by Year in descending order

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Version: 2024-02-01

40  
papers

26,557  
citations

257101

24  
h-index

288905

40  
g-index

41  
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41  
docs citations

41  
times ranked

50393  
citing authors

#	ARTICLE	IF	CITATIONS
1	Phenotypic and protein localization heterogeneity associated with <i>AHDC1</i> pathogenic protein-truncating alleles in Xia-Gibbs syndrome. <i>Human Mutation</i> , 2021, 42, 577-591.	1.1	14
2	Exome variant discrepancies due to reference-genome differences. <i>American Journal of Human Genetics</i> , 2021, 108, 1239-1250.	2.6	36
3	Neptune: an environment for the delivery of genomic medicine. <i>Genetics in Medicine</i> , 2021, 23, 1838-1846.	1.1	3
4	Sequencing of a central nervous system tumor demonstrates cancer transmission in an organ transplant. <i>Life Science Alliance</i> , 2021, 4, e202000941.	1.3	1
5	Exome sequencing in children with clinically suspected maturity-onset diabetes of the young. <i>Pediatric Diabetes</i> , 2021, 22, 960-968.	1.2	6
6	Genetic testing in ambulatory cardiology clinics reveals high rate of findings with clinical management implications. <i>Genetics in Medicine</i> , 2021, 23, 2404-2414.	1.1	14
7	Germline Cancer Predisposition Variants in Pediatric Rhabdomyosarcoma: A Report From the Children's Oncology Group. <i>Journal of the National Cancer Institute</i> , 2021, 113, 875-883.	3.0	55
8	Community-based recruitment and exome sequencing indicates high diagnostic yield in adults with intellectual disability. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1439.	0.6	6
9	Combination of whole exome sequencing and animal modeling identifies <i>TMPRSS9</i> as a candidate gene for autism spectrum disorder. <i>Human Molecular Genetics</i> , 2020, 29, 459-470.	1.4	32
10	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. <i>Npj Genomic Medicine</i> , 2019, 4, 19.	1.7	163
11	A Genocentric Approach to Discovery of Mendelian Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 974-986.	2.6	30
12	Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. <i>Hepatology</i> , 2019, 70, 899-910.	3.6	58
13	Xia-Gibbs syndrome in adulthood: a case report with insight into the natural history of the condition. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003608.	0.5	15
14	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. <i>Neuron</i> , 2018, 97, 488-493.	3.8	265
15	Exome sequencing reveals novel genetic loci influencing obesity-related traits in Hispanic children. <i>Obesity</i> , 2017, 25, 1270-1276.	1.5	10
16	A t(5;16) translocation is the likely driver of a syndrome with ambiguous genitalia, facial dysmorphism, intellectual disability, and speech delay. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a000703.	0.5	1
17	Meta-analysis of 49,549 individuals imputed with the 1000 Genomes Project reveals an exonic damaging variant in <i>ANGPTL4</i> determining fasting TG levels. <i>Journal of Medical Genetics</i> , 2016, 53, 441-449.	1.5	34
18	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. <i>American Journal of Human Genetics</i> , 2016, 99, 318-336.	2.6	112

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19	Fine mapping the CETP region reveals a common intronic insertion associated to HDL-C. <i>Npj Aging and Mechanisms of Disease</i> , 2015, 1, 15011.	4.5	8
20	A global reference for human genetic variation. <i>Nature</i> , 2015, 526, 68-74.	13.7	13,998
21	Assessing structural variation in a personal genome towards a human reference diploid genome. <i>BMC Genomics</i> , 2015, 16, 286.	1.2	153
22	Synaptic, transcriptional and chromatin genes disrupted in autism. <i>Nature</i> , 2014, 515, 209-215.	13.7	2,254
23	A framework for the interpretation of de novo mutation in human disease. <i>Nature Genetics</i> , 2014, 46, 944-950.	9.4	943
24	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 745-754.	2.6	80
25	Whole Exome Sequencing Identifies Novel Genes for Fetal Hemoglobin Response to Hydroxyurea in Children with Sickle Cell Anemia. <i>PLoS ONE</i> , 2014, 9, e110740.	1.1	28
26	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. <i>Neuron</i> , 2013, 77, 235-242.	3.8	242
27	Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. <i>PLoS Genetics</i> , 2013, 9, e1003443.	1.5	133
28	FOXO3 Variants Are Associated With Lower Fetal Hemoglobin Levels In Children With Sickle Cell Disease. <i>Blood</i> , 2013, 122, 778-778.	0.6	1
29	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. <i>Nature Medicine</i> , 2012, 18, 1423-1428.	15.2	103
30	Patterns and rates of exonic de novo mutations in autism spectrum disorders. <i>Nature</i> , 2012, 485, 242-245.	13.7	1,597
31	Genetic Predictors of Hemoglobin F Response to Hydroxyurea in Sickle Cell Anemia. <i>Blood</i> , 2012, 120, 241-241.	0.6	5
32	PCR detection of nearly any dengue virus strain using a highly sensitive primer "cocktail"™. <i>FEBS Journal</i> , 2011, 278, 1676-1687.	2.2	14
33	Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorders. <i>Human Molecular Genetics</i> , 2011, 20, 3366-3375.	1.4	149
34	The Genome Sequence of Taurine Cattle: A Window to Ruminant Biology and Evolution. <i>Science</i> , 2009, 324, 522-528.	6.0	1,038
35	Somatic mutations affect key pathways in lung adenocarcinoma. <i>Nature</i> , 2008, 455, 1069-1075.	13.7	2,694
36	Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. <i>Science</i> , 2007, 316, 222-234.	6.0	1,283

#	ARTICLE	IF	CITATIONS
37	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
38	Generation and annotation of the DNA sequences of human chromosomes 2 and 4. Nature, 2005, 434, 724-731.	13.7	85
39	EAnnot: A genome annotation tool using experimental evidence. Genome Research, 2004, 14, 2503-2509.	2.4	18
40	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