# Aniko Sabo

List of Publications by Year
in descending order

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

Version: 2024-02-01

| 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 | 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 |
| 2 | Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250. | 2.6 | 36 |
| 3 | Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846. | 1.1 | 3 |
| 4 | Sequencing of a central nervous system tumor demonstrates cancer transmission in an organ transplant. Life Science Alliance, 2021, 4, e202000941. | 1.3 | 1 |
| 5 | Exome sequencing in children with clinically suspected <scp>maturityâ€onset</scp> diabetes of the young. Pediatric Diabetes, 2021, 22, 960-968. | 1.2 | 6 |
| 6 | 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 |
| 7 | 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 |
| 8 | 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 |
| 9 | 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 |
| 10 | Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19. | 1.7 | 163 |
| 11 | A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 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. Hepatology, 2019, 70, 899-910. | 3.6 | 58 |
| 13 | 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 |
| 14 | SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493. | 3.8 | 265 |
| 15 | Exome sequencing reveals novel genetic loci influencing obesityâ€related traits in Hispanic children. Obesity, 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. Journal of Physical Education and Sports Management, 2016, 2, a000703. | 0.5 | 1 |
| 17 | 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 |
| 18 | Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336. | 2.6 | 112 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 20 | A global reference for human genetic variation. Nature, 2015, 526, 68-74. | 13.7 | 13,998 |
| 21 | Assessing structural variation in a personal genomeâ€"towards a human reference diploid genome. BMC Genomics, 2015, 16, 286. | 1.2 | 153 |
| 22 | Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215. | 13.7 | 2,254 |
| 23 | A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950. | 9.4 | 943 |
| 24 | 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 |
| 25 | 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 |
| 26 | Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242. | 3.8 | 242 |
| 27 | Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. PLoS Genetics, 2013, 9, e1003443. | 1.5 | 133 |
| 28 | FOXO3 Variants Are Associated With Lower Fetal Hemoglobin Levels In Children With Sickle Cell Disease. Blood, 2013, 122, 778-778. | 0.6 | 1 |
| 29 | Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428. | 15.2 | 103 |
| 30 | Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245. | 13.7 | 1,597 |
| 31 | Genetic Predictors of Hemoglobin F Response to Hydroxyurea in Sickle Cell Anemia. Blood, 2012, 120, 241-241. | 0.6 | 5 |
| 32 | PCR detection of nearly any dengue virus strain using a highly sensitive primer â€˜cocktailâ€™. FEBS Journal, 2011, 278, 1676-1687. | 2.2 | 14 |
| 33 | Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorders. Human Molecular Genetics, 2011, 20, 3366-3375. | 1.4 | 149 |
| 34 | The Genome Sequence of Taurine Cattle: A Window to Ruminant Biology and Evolution. Science, 2009, 324, 522-528. | 6.0 | 1,038 |
| 35 | Somatic mutations affect key pathways in lung adenocarcinoma. Nature, 2008, 455, 1069-1075. | 13.7 | 2,694 |
| 36 | Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. Science, 2007, 316, 222-234. | 6.0 | 1,283 |

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|---|---|---|---|
| 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 |