# Sami S Amr

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7625379/publications.pdf

Version: 2024-02-01

| | | | |
|---|---|---|---|
| 41 papers | 1,506 citations | 471509 17 h-index | 345221 36 g-index |
| 47 all docs | 47 docs citations | 47 times ranked | 3112 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613. | 2.5 | 312 |
| 2 | A Homozygous Mutation in a Novel Zinc-Finger Protein, ERIS, Is Responsible for Wolfram Syndrome 2. American Journal of Human Genetics, 2007, 81, 673-683. | 6.2 | 233 |
| 3 | Association of Rare Pathogenic DNA Variants for Familial Hypercholesterolemia, Hereditary Breast and Ovarian Cancer Syndrome, and Lynch Syndrome With Disease Risk in Adults According to Family History. JAMA Network Open, 2020, 3, e203959. | 5.9 | 75 |
| 4 | ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247. | 2.4 | 67 |
| 5 | Improving hearing loss gene testing: a systematic review of gene evidence toward more efficient next-generation sequencingâ€“based diagnostic testing and interpretation. Genetics in Medicine, 2016, 18, 545-553. | 2.4 | 63 |
| 6 | A proposal for comprehensive newborn hearing screening to improve identification of deaf and hard-of-hearing children. Genetics in Medicine, 2019, 21, 2614-2630. | 2.4 | 63 |
| 7 | The eMERGE genotype set of 83,717 subjects imputed to ~40â€‰million variants genome wide and association with the herpes zoster medical record phenotype. Genetic Epidemiology, 2019, 43, 63-81. | 1.3 | 63 |
| 8 | VisCap: inference and visualization of germ-line copy-number variants from targeted clinical sequencing data. Genetics in Medicine, 2016, 18, 712-719. | 2.4 | 61 |
| 9 | Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. Genetics in Medicine, 2019, 21, 2442-2452. | 2.4 | 56 |
| 10 | Characteristics of Men Who Report Persistent Sexual Symptoms After Finasteride Use for Hair Loss. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 4669-4680. | 3.6 | 54 |
| 11 | Comprehensive Diagnostic Testing for Stereocilin. Journal of Molecular Diagnostics, 2014, 16, 639-647. | 2.8 | 53 |
| 12 | HIV Integration Site Analysis of Cellular Models of HIV Latency with a Probe-Enriched Next-Generation Sequencing Assay. Journal of Virology, 2016, 90, 4511-4519. | 3.4 | 47 |
| 13 | Targeted Droplet-Digital PCR as a Tool for Novel Deletion Discovery at the DFNB1 Locus. Human Mutation, 2016, 37, 119-126. | 2.5 | 37 |
| 14 | Next generation sequencingâ€based copy number analysis reveals low prevalence of deletions and duplications in 46 genes associated with genetic cardiomyopathies. Molecular Genetics & Genomic Medicine, 2016, 4, 143-151. | 1.2 | 29 |
| 15 | Empirical comparison of reduced representation bisulfite sequencing and Infinium BeadChip reproducibility and coverage of DNA methylation in humans. Npj Genomic Medicine, 2017, 2, 13. | 3.8 | 26 |
| 16 | Curating Clinically Relevant Transcripts for the Interpretation of Sequence Variants. Journal of Molecular Diagnostics, 2018, 20, 789-801. | 2.8 | 25 |
| 17 | Allele-Specific Droplet Digital PCR Combined with a Next-Generation Sequencing-Based Algorithm for Diagnostic Copy Number Analysis in Genes with High Homology: Proof of Concept Using Stereocilin. Clinical Chemistry, 2018, 64, 705-714. | 3.2 | 24 |
| 18 | Recurrent variants in OTOF are significant contributors to prelingual nonsydromic hearing loss in Saudi patients. Genetics in Medicine, 2018, 20, 536-544. | 2.4 | 18 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | COMPSRA: a COMprehensive Platform for Small RNA-Seq data Analysis. Scientific Reports, 2020, 10, 4552. | 3.3 | 18 |
| 20 | New Tmc1 Deafness Mutations Impact Mechanotransduction in Auditory Hair Cells. Journal of Neuroscience, 2021, 41, 4378-4391. | 3.6 | 18 |
| 21 | Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212. | 2.4 | 18 |
| 22 | Histopathology of the Human Inner Ear in a Patient With Sensorineural Hearing Loss Caused by a Variant in DFNA5. Otology and Neurotology, 2015, 36, 1616-1621. | 1.3 | 17 |
| 23 | SOX12 and NRSN2 are candidate genes for 20p13 subtelomeric deletions associated with developmental delay. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 832-840. | 1.7 | 15 |
| 24 | Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. Genetics in Medicine, 2017, 19, 496-504. | 2.4 | 15 |
| 25 | Genetic studies in Drosophila and humans support a model for the concerted function of CISD2, PPT1 and CLN3 in disease. Biology Open, 2014, 3, 342-352. | 1.2 | 10 |
| 26 | Molecular characterization of pathogenic OTOA gene conversions in hearing loss patients. Human Mutation, 2021, 42, 373-377. | 2.5 | 10 |
| 27 | Analysis of intragenic USH2A copy number variation unveils broad spectrum of unique and recurrent variants. European Journal of Medical Genetics, 2018, 61, 621-626. | 1.3 | 9 |
| 28 | The Translational Genomics Core at Partners Personalized Medicine: Facilitating the Transition of Research towards Personalized Medicine. Journal of Personalized Medicine, 2016, 6, 10. | 2.5 | 8 |
| 29 | Novel CERKL variant in consanguineous Jordanian pedigrees with inherited retinal dystrophies. Canadian Journal of Ophthalmology, 2019, 54, 51-59. | 0.7 | 8 |
| 30 | A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. European Journal of Human Genetics, 2021, 29, 988-997. | 2.8 | 8 |
| 31 | Commercially Available Blocking Oligonucleotides Effectively Suppress Unwanted Hemolysis-Related miRNAs in a Large Whole-Blood RNA Cohort. Journal of Molecular Diagnostics, 2021, 23, 671-682. | 2.8 | 8 |
| 32 | Automated Pharmacogenomic Reports for Clinical Genome Sequencing. Journal of Molecular Diagnostics, 2022, 24, 205-218. | 2.8 | 5 |
| 33 | Prenatal cytogenomic identification and molecular refinement of compound heterozygous STRC deletion breakpoints. Molecular Genetics & Genomic Medicine, 2019, 7, e806. | 1.2 | 4 |
| 34 | Novel frameshift variant in the IDUA gene underlies Mucopolysaccharidoses type I in a consanguineous Yemeni pedigree. Molecular Genetics and Metabolism Reports, 2017, 12, 76-79. | 1.1 | 3 |
| 35 | Extending the spectrum of CLRN1â€•and ABCA4â€associated inherited retinal dystrophies caused by novel and recurrent variants using exome sequencing. Molecular Genetics & Genomic Medicine, 2020, 8, e1123. | 1.2 | 3 |
| 36 | Unique Variant Spectrum in a Jordanian Cohort with Inherited Retinal Dystrophies. Genes, 2021, 12, 593. | 2.4 | 3 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Lung function, airway and peripheral basophils and eosinophils are associated with molecular pharmacogenomic endotypes of steroid response in severe asthma. Thorax, 2022, 77, 452-460. | 5.6 | 3 |
| 38 | Clinical heterogeneity in retinitis pigmentosa caused by variants in and in five extended consanguineous pedigrees. Molecular Vision, 2020, 26, 445-458. | 1.1 | 2 |
| 39 | The Burden and Benefits of Knowledge: Ethical Considerations Surrounding Population-Based Newborn Genome Screening for Hearing. International Journal of Neonatal Screening, 2022, 8, 36. | 3.2 | 2 |
| 40 | Targeted Hybrid Capture for Inherited Disease Panels. , 2015, , 251-269. | | 0 |
| 41 | On the Genetic Trail of Hearing Loss. ASHA Leader, 2015, 20, . | 0.1 | 0 |