# Erfan Aref-Eshghi

List of Publications by Year
in descending order



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|---|---|---|---|
| 42 papers | 1,686 citations | 331670 21 h-index | 302126 39 g-index |
| 44 all docs | 44 docs citations | 44 times ranked | 2385 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370. | 6.2 | 171 |
| 2 | Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. American Journal of Human Genetics, 2018, 102, 156-174. | 6.2 | 135 |
| 3 | Diagnostic Utility of Genome-wide DNA Methylation Testing in Genetically Unsolved Individuals with Suspected Hereditary Conditions. American Journal of Human Genetics, 2019, 104, 685-700. | 6.2 | 125 |
| 4 | Clinical Validation of Copy Number Variant Detection from Targeted Next-Generation Sequencing Panels. Journal of Molecular Diagnostics, 2017, 19, 905-920. | 2.8 | 104 |
| 5 | Classification of osteoarthritis phenotypes by metabolomics analysis. BMJ Open, 2014, 4, e006286. | 1.9 | 90 |
| 6 | Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. Genetics in Medicine, 2021, 23, 1065-1074. | 2.4 | 88 |
| 7 | BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885. | 12.8 | 83 |
| 8 | The defining DNA methylation signature of Kabuki syndrome enables functional assessment of genetic variants of unknown clinical significance. Epigenetics, 2017, 12, 923-933. | 2.7 | 79 |
| 9 | Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64. | 4.1 | 71 |
| 10 | Screening for genes that accelerate the epigenetic aging clock in humans reveals a role for the H3K36 methyltransferase NSD1. Genome Biology, 2019, 20, 146. | 8.8 | 66 |
| 11 | Peripheral blood epi-signature of Claes-Jensen syndrome enables sensitive and specific identification of patients and healthy carriers with pathogenic mutations in KDM5C. Clinical Epigenetics, 2018, 10, 21. | 4.1 | 58 |
| 12 | Relationship Between Blood Plasma and Synovial Fluid Metabolite Concentrations in Patients with Osteoarthritis. Journal of Rheumatology, 2015, 42, 859-865. | 2.0 | 45 |
| 13 | DNA methylation signatures in mendelian developmental disorders as a diagnostic bridge between genotype and phenotype. Epigenomics, 2019, 11, 563-575. | 2.1 | 42 |
| 14 | Genome-wide DNA methylation study of hip and knee cartilage reveals embryonic organ and skeletal system morphogenesis as major pathways involved in osteoarthritis. BMC Musculoskeletal Disorders, 2015, 16, 287. | 1.9 | 41 |
| 15 | Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7. | 4.1 | 40 |
| 16 | Clinical Validation of a Genome-Wide DNA Methylation Assay for Molecular Diagnosis of Imprinting Disorders. Journal of Molecular Diagnostics, 2017, 19, 848-856. | 2.8 | 39 |
| 17 | Genomic DNA Methylation-Derived Algorithm Enables Accurate Detection of Malignant Prostate Tissues. Frontiers in Oncology, 2018, 8, 100. | 2.8 | 38 |
| 18 | Overexpression of MMP13 in human osteoarthritic cartilage is associated with the SMAD-independent TGF-Î² signalling pathway. Arthritis Research and Therapy, 2015, 17, 264. | 3.5 | 37 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Metabolomic analysis of human synovial fluid and plasma reveals that phosphatidylcholine metabolism is associated with both osteoarthritis and diabetes mellitus. Metabolomics, 2016, 12, 1. | 3.0 | 37 |
| 20 | De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaidesâ€“Baraitser syndrome. Genetics in Medicine, 2020, 22, 1838-1850. | 2.4 | 31 |
| 21 | Genomeâ€wide DNA methylation and RNA analyses enable reclassification of two variants of uncertain significance in a patient with clinical Kabuki syndrome. Human Mutation, 2019, 40, 1684-1689. | 2.5 | 27 |
| 22 | Diagnostic Utility of Genome-Wide DNA Methylation Analysis in Mendelian Neurodevelopmental Disorders. International Journal of Molecular Sciences, 2020, 21, 9303. | 4.1 | 23 |
| 23 | Functional annotation of genomic variation: DNA methylation episignatures in neurodevelopmental Mendelian disorders. Human Molecular Genetics, 2020, 29, R27-R32. | 2.9 | 23 |
| 24 | Identification of Dyslipidemic Patients Attending Primary Care Clinics Using Electronic Medical Record (EMR) Data from the Canadian Primary Care Sentinel Surveillance Network (CPCSSN) Database. Journal of Medical Systems, 2017, 41, 45. | 3.6 | 22 |
| 25 | Does the Prevalence of Dyslipidemias Differ between Newfoundland and the Rest of Canada? Findings from the Electronic Medical Records of the Canadian Primary Care Sentinel Surveillance Network. Frontiers in Cardiovascular Medicine, 2015, 2, 1. | 2.4 | 21 |
| 26 | Implementation of an NGSâ€based sequencing and gene fusion panel for clinical screening of patients with suspected hematologic malignancies. European Journal of Haematology, 2019, 103, 178-189. | 2.2 | 21 |
| 27 | Using Electronic Medical Record to Identify Patients With Dyslipidemia in Primary Care Settings: International Classification of Disease Code Matters From One Region to a National Database. Biomedical Informatics Insights, 2017, 9, 117822261668588. | 4.6 | 18 |
| 28 | SMAD3 Is Associated with the Total Burden of Radiographic Osteoarthritis: The Chingford Study. PLoS ONE, 2014, 9, e97786. | 2.5 | 17 |
| 29 | Single and mixed dyslipidaemia in Canadian primary care settings: findings from the Canadian primary care sentinel surveillance network database. BMJ Open, 2015, 5, e007954. | 1.9 | 13 |
| 30 | Low density lipoprotein cholesterol control status among Canadians at risk for cardiovascular disease: findings from the Canadian Primary Care Sentinel Surveillance Network Database. Lipids in Health and Disease, 2015, 14, 60. | 3.0 | 11 |
| 31 | SMAD3 Is Upregulated in Human Osteoarthritic Cartilage Independent of the Promoter DNA Methylation. Journal of Rheumatology, 2016, 43, 388-394. | 2.0 | 10 |
| 32 | Genetic and epigenetic profiling of BRCA1/2 in ovarian tumors reveals additive diagnostic yield and evidence of a genomic BRCA1/2 DNA methylation signature. Journal of Human Genetics, 2020, 65, 865-873. | 2.3 | 10 |
| 33 | Glucose-induced, duration-dependent genome-wide DNA methylation changes in human endothelial cells. American Journal of Physiology - Cell Physiology, 2020, 319, C268-C276. | 4.6 | 10 |
| 34 | Detection of a DNA Methylation Signature for the Intellectual Developmental Disorder, X-Linked, Syndromic, Armfield Type. International Journal of Molecular Sciences, 2021, 22, 1111. | 4.1 | 10 |
| 35 | Six-year time-trend analysis of dyslipidemia among adults in Newfoundland and Labrador: findings from the laboratory information system between 2009 and 2014. Lipids in Health and Disease, 2018, 17, 99. | 3.0 | 5 |
| 36 | The oncogenic roles of NTRK fusions and methods of molecular diagnosis. Cancer Genetics, 2021, 258-259, 110-119. | 0.4 | 5 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Advanced approach for comprehensive mtDNA genome testing in mitochondrial disease. Molecular Genetics and Metabolism, 2022, 135, 93-101. | 1.1 | 5 |
| 38 | Epigenomic Mechanisms of Human Developmental Disorders. , 2018, , 837-859. | | 4 |
| 39 | Genetic associations in community context: a mixed model approach identifies a functional variant in the RBP4 gene associated with HDL-C dyslipidemia. BMC Medical Genetics, 2018, 19, 205. | 2.1 | 3 |
| 40 | Clinical and technical assessment of MedExome vs. NGS panels in patients with suspected genetic disorders in Southwestern Ontario. Journal of Human Genetics, 2021, 66, 451-464. | 2.3 | 2 |
| 41 | A Novel TP53 Tandem Duplication in a Child with Li-Fraumeni Syndrome. Journal of Physical Education and Sports Management, 2022, , mcs.a006181. | 1.2 | 2 |
| 42 | Multiparametric magnetic resonance imaging of multifocal prostate cancer to reveal intra-prostatic genomic heterogeneity and novel radio-genomic correlates: Results of the Smarter Prostate Interventions and Therapeutics (SPIRIT) study.. Journal of Clinical Oncology, 2020, 38, 20-20. | 1.6 | 0 |