Erfan Aref-Eshghi

List of Publications by Year in Descending Order

41 papers	912 citations	18 h-index	29 g-index
44 ext. papers	1,364 ext. citations	5.6 avg, IF	3.98 L-index

#	Paper	IF	Citations
41	Advanced approach for comprehensive mtDNA genome testing in mitochondrial disease.. *Molecular Genetics and Metabolism*, **2021**, 135, 93-93	3.7	1
40	The oncogenic roles of NTRK fusions and methods of molecular diagnosis. *Cancer Genetics*, **2021**, 258-259, 110-119	2.3	0
39	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	4.3	1
38	Detection of a DNA Methylation Signature for the Intellectual Developmental Disorder, X-Linked, Syndromic, Armfield Type. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
37	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. *Genetics in Medicine*, **2021**, 23, 1065-1074	8.1	10
36	Diagnostic Utility of Genome-Wide DNA Methylation Analysis in Mendelian Neurodevelopmental Disorders. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	7
35	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	4.3	5
34	Glucose-induced, duration-dependent genome-wide DNA methylation changes in human endothelial cells. *American Journal of Physiology - Cell Physiology*, **2020**, 319, C268-C276	5.4	5
33	Functional annotation of genomic variation: DNA methylation episignatures in neurodevelopmental Mendelian disorders. *Human Molecular Genetics*, **2020**, 29, R27-R32	5.6	9
32	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. *American Journal of Human Genetics*, **2020**, 106, 356-370	11	51
31	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	2.2	
30	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. *Clinical Epigenetics*, **2020**, 12, 7	7.7	23
29	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome. *Genetics in Medicine*, **2020**, 22, 1838-1850	8.1	8
28	Implementation of an NGS-based sequencing and gene fusion panel for clinical screening of patients with suspected hematologic malignancies. *European Journal of Haematology*, **2019**, 103, 178-189	3.8	16
27	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. *Clinical Epigenetics*, **2019**, 11, 64	7.7	29
26	DNA methylation signatures in mendelian developmental disorders as a diagnostic bridge between genotype and phenotype. *Epigenomics*, **2019**, 11, 563-575	4.4	21
25	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	11	57

24	Screening for genes that accelerate the epigenetic aging clock in humans reveals a role for the H3K36 methyltransferase NSD1. *Genome Biology*, **2019**, 20, 146	18.3	36
23	Genome-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	4.7	12
22	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. *American Journal of Human Genetics*, **2018**, 102, 156-174	11	75
21	Peripheral blood epi-signature of Claes-Jensen syndrome enables sensitive and specific identification of patients and healthy carriers with pathogenic mutations in. *Clinical Epigenetics*, **2018**, 10, 21	7.7	37
20	Genomic DNA Methylation-Derived Algorithm Enables Accurate Detection of Malignant Prostate Tissues. *Frontiers in Oncology*, **2018**, 8, 100	5.3	25
19	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	4.4	4
18	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	1
17	BAFopathiesTDNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin-Siris and Nicolaides-Baraitser syndromes. *Nature Communications*, **2018**, 9, 4885	17.4	48
16	Epigenomic Mechanisms of Human Developmental Disorders **2018**, 837-859		4
15	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	5.1	10
14	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, 1178222616685880	4.9	14
13	The defining DNA methylation signature of Kabuki syndrome enables functional assessment of genetic variants of unknown clinical significance. *Epigenetics*, **2017**, 12, 923-933	5.7	43
12	Clinical Validation of a Genome-Wide DNA Methylation Assay for Molecular Diagnosis of Imprinting Disorders. *Journal of Molecular Diagnostics*, **2017**, 19, 848-856	5.1	29
11	Clinical Validation of Copy Number Variant Detection from Targeted Next-Generation Sequencing Panels. *Journal of Molecular Diagnostics*, **2017**, 19, 905-920	5.1	75
10	SMAD3 Is Upregulated in Human Osteoarthritic Cartilage Independent of the Promoter DNA Methylation. *Journal of Rheumatology*, **2016**, 43, 388-94	4.1	8
9	Metabolomic analysis of human synovial fluid and plasma reveals that phosphatidylcholine metabolism is associated with both osteoarthritis and diabetes mellitus. *Metabolomics*, **2016**, 12, 1	4.7	30
8	Relationship between blood plasma and synovial fluid metabolite concentrations in patients with osteoarthritis. *Journal of Rheumatology*, **2015**, 42, 859-65	4.1	35
7	Overexpression of MMP13 in human osteoarthritic cartilage is associated with the SMAD-independent TGF-β signalling pathway. *Arthritis Research and Therapy*, **2015**, 17, 264	5.7	30

6	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	2.8	32
5	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	4.4	8
4	Single and mixed dyslipidaemia in Canadian primary care settings: findings from the Canadian primary care sentinel surveillance network database. *BMJ Open*, **2015**, 5, e007954	3	10
3	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	5.4	16
2	Classification of osteoarthritis phenotypes by metabolomics analysis. *BMJ Open*, **2014**, 4, e006286	3	67
1	SMAD3 is associated with the total burden of radiographic osteoarthritis: the Chingford study. *PLoS ONE*, **2014**, 9, e97786	3.7	17