Erfan Aref-Eshghi

List of Publications by Citations

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	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
40	Clinical Validation of Copy Number Variant Detection from Targeted Next-Generation Sequencing Panels. *Journal of Molecular Diagnostics*, **2017**, 19, 905-920	5.1	75
39	Classification of osteoarthritis phenotypes by metabolomics analysis. *BMJ Open*, **2014**, 4, e006286	3	67
38	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
37	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
36	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
35	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
34	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
33	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
32	Relationship between blood plasma and synovial fluid metabolite concentrations in patients with osteoarthritis. *Journal of Rheumatology*, **2015**, 42, 859-65	4.1	35
31	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
30	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
29	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
28	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. *Clinical Epigenetics*, **2019**, 11, 64	7.7	29
27	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
26	Genomic DNA Methylation-Derived Algorithm Enables Accurate Detection of Malignant Prostate Tissues. *Frontiers in Oncology*, **2018**, 8, 100	5.3	25
25	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. *Clinical Epigenetics*, **2020**, 12, 7	7.7	23

24	DNA methylation signatures in mendelian developmental disorders as a diagnostic bridge between genotype and phenotype. *Epigenomics*, **2019**, 11, 563-575	4.4	21
23	SMAD3 is associated with the total burden of radiographic osteoarthritis: the Chingford study. *PLoS ONE*, **2014**, 9, e97786	3.7	17
22	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
21	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
20	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
19	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
18	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
17	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
16	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. *Genetics in Medicine*, **2021**, 23, 1065-1074	8.1	10
15	Functional annotation of genomic variation: DNA methylation episignatures in neurodevelopmental Mendelian disorders. *Human Molecular Genetics*, **2020**, 29, R27-R32	5.6	9
14	SMAD3 Is Upregulated in Human Osteoarthritic Cartilage Independent of the Promoter DNA Methylation. *Journal of Rheumatology*, **2016**, 43, 388-94	4.1	8
13	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
12	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
11	Diagnostic Utility of Genome-Wide DNA Methylation Analysis in Mendelian Neurodevelopmental Disorders. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	7
10	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
9	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
8	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
7	Epigenomic Mechanisms of Human Developmental Disorders **2018**, 837-859		4

6	Advanced approach for comprehensive mtDNA genome testing in mitochondrial disease.. *Molecular Genetics and Metabolism*, **2021**, 135, 93-93	3.7	1
5	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
4	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
3	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
2	The oncogenic roles of NTRK fusions and methods of molecular diagnosis. *Cancer Genetics*, **2021**, 258-259, 110-119	2.3	0
1	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	