Bekim Sadikovic

List of Publications by Citations

98 papers	2,128 citations	28 h-index	44 g-index
118 ext. papers	2,910 ext. citations	5.1 avg, IF	4.61 L-index

#	Paper	IF	Citations
98	Identification of a microRNA signature associated with progression of leukoplakia to oral carcinoma. *Human Molecular Genetics*, **2009**, 18, 4818-29	5.6	187
97	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. *Journal of Medical Genetics*, **2015**, 52, 431-7	5.8	137
96	Benzopyrene exposure disrupts DNA methylation and growth dynamics in breast cancer cells. *Toxicology and Applied Pharmacology*, **2006**, 216, 458-68	4.6	101
95	Identification of interactive networks of gene expression associated with osteosarcoma oncogenesis by integrated molecular profiling. *Human Molecular Genetics*, **2009**, 18, 1962-75	5.6	100
94	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 7974-81	11.5	94
93	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
92	Clinical Validation of Copy Number Variant Detection from Targeted Next-Generation Sequencing Panels. *Journal of Molecular Diagnostics*, **2017**, 19, 905-920	5.1	75
91	Discovery of novel hypermethylated genes in prostate cancer using genomic CpG island microarrays. *PLoS ONE*, **2009**, 4, e4830	3.7	74
90	In vitro analysis of integrated global high-resolution DNA methylation profiling with genomic imbalance and gene expression in osteosarcoma. *PLoS ONE*, **2008**, 3, e2834	3.7	66
89	Genome-wide H3K9 histone acetylation profiles are altered in benzopyrene-treated MCF7 breast cancer cells. *Journal of Biological Chemistry*, **2008**, 283, 4051-60	5.4	60
88	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
87	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
86	Identification of a methylation profile for DNMT1-associated autosomal dominant cerebellar ataxia, deafness, and narcolepsy. *Clinical Epigenetics*, **2016**, 8, 91	7.7	50
85	BAFopathiesRDNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin-Siris and Nicolaides-Baraitser syndromes. *Nature Communications*, **2018**, 9, 4885	17.4	48
84	NLRP7 affects trophoblast lineage differentiation, binds to overexpressed YY1 and alters CpG methylation. *Human Molecular Genetics*, **2014**, 23, 706-16	5.6	46
83	Decitabine-induced demethylation of 5RCpG island in GADD45A leads to apoptosis in osteosarcoma cells. *Neoplasia*, **2008**, 10, 471-80	6.4	45
82	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

81	Sequence homology at the breakpoint and clinical phenotype of mitochondrial DNA deletion syndromes. *PLoS ONE*, **2010**, 5, e15687	3.7	43
80	Identification of epigenetic signature associated with alpha thalassemia/mental retardation X-linked syndrome. *Epigenetics and Chromatin*, **2017**, 10, 10	5.8	39
79	Mutation Update for UBE3A variants in Angelman syndrome. *Human Mutation*, **2014**, 35, 1407-17	4.7	38
78	The defining DNA methylation signature of Floating-Harbor Syndrome. *Scientific Reports*, **2016**, 6, 38803	4.9	38
77	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
76	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
75	Recurrent genomic alterations in sequential progressive leukoplakia and oral cancer: drivers of oral tumorigenesis?. *Human Molecular Genetics*, **2014**, 23, 2618-28	5.6	36
74	Clinical Next-Generation Sequencing Pipeline Outperforms a Combined Approach Using Sanger Sequencing and Multiplex Ligation-Dependent Probe Amplification in Targeted Gene Panel Analysis. *Journal of Molecular Diagnostics*, **2016**, 18, 657-667	5.1	36
73	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. *Clinical Epigenetics*, **2019**, 11, 64	7.7	29
72	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
71	Gender and BCR-ABL transcript type are correlated with molecular response to imatinib treatment in patients with chronic myeloid leukemia. *European Journal of Haematology*, **2016**, 96, 360-6	3.8	28
70	Clinical Validation of Fragile X Syndrome Screening by DNA Methylation Array. *Journal of Molecular Diagnostics*, **2016**, 18, 834-841	5.1	28
69	Genomic DNA Methylation-Derived Algorithm Enables Accurate Detection of Malignant Prostate Tissues. *Frontiers in Oncology*, **2018**, 8, 100	5.3	25
68	DNA methylation analysis using CpG microarrays is impaired in benzopyrene exposed cells. *Toxicology and Applied Pharmacology*, **2007**, 225, 300-9	4.6	23
67	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. *Clinical Epigenetics*, **2020**, 12, 7	7.7	23
66	DNA methylation signatures in mendelian developmental disorders as a diagnostic bridge between genotype and phenotype. *Epigenomics*, **2019**, 11, 563-575	4.4	21
65	DNA methylation analysis in constitutional disorders: Clinical implications of the epigenome. *Critical Reviews in Clinical Laboratory Sciences*, **2016**, 53, 147-65	9.4	21
64	The ClinGen Epilepsy Gene Curation Expert Panel-Bridging the divide between clinical domain knowledge and formal gene curation criteria. *Human Mutation*, **2018**, 39, 1476-1484	4.7	21

63	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). *Genetics in Medicine*, **2018**, 20, 294-302	8.1	20
62	Characterization of functional elements in the neurofibromatosis (NF1) proximal promoter region. *Oncogene*, **2004**, 23, 330-9	9.2	20
61	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
60	Clinical evaluation of a hemochromatosis next-generation sequencing gene panel. *European Journal of Haematology*, **2017**, 98, 228-234	3.8	15
59	Constitutional Epi/Genetic Conditions: Genetic, Epigenetic, and Environmental Factors. *Journal of Pediatric Genetics*, **2017**, 6, 30-41	0.7	13
58	Genotypes of chronic progressive external ophthalmoplegia in a large adult-onset cohort. *Mitochondrion*, **2019**, 49, 227-231	4.9	13
57	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
56	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. *American Journal of Human Genetics*, **2021**, 108, 502-516	11	12
55	Movement Disorders Associated With Hemochromatosis. *Canadian Journal of Neurological Sciences*, **2016**, 43, 801-808	1	11
54	Array comparative genomic hybridization in osteosarcoma. *Methods in Molecular Biology*, **2013**, 973, 227-47	1.4	10
53	Immunohistochemical expression and cluster analysis of mesenchymal and neural stem cell-associated proteins in pediatric soft tissue sarcomas. *Pediatric and Developmental Pathology*, **2011**, 14, 259-72	2.2	10
52	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. *Genetics in Medicine*, **2021**, 23, 1065-1074	8.1	10
51	Functional annotation of genomic variation: DNA methylation episignatures in neurodevelopmental Mendelian disorders. *Human Molecular Genetics*, **2020**, 29, R27-R32	5.6	9
50	Chronic maternal protein deprivation in mice is associated with overexpression of the cohesin-mediator complex in liver of their offspring. *Journal of Nutrition*, **2011**, 141, 2106-12	4.1	8
49	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
48	Diagnostic Utility of Genome-Wide DNA Methylation Analysis in Mendelian Neurodevelopmental Disorders. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	7
47	Cross-oncopanel study reveals high sensitivity and accuracy with overall analytical performance depending on genomic regions. *Genome Biology*, **2021**, 22, 109	18.3	6
46	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

45	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
44	Clinical value of next-generation sequencing compared to cytogenetics in patients with suspected myelodysplastic syndrome. *British Journal of Haematology*, **2021**, 192, 729-736	4.5	5
43	Implementation of Epilepsy Multigene Panel Testing in Ontario, Canada. *Canadian Journal of Neurological Sciences*, **2020**, 47, 61-68	1	4
42	DNA methylation epi-signature is associated with two molecularly and phenotypically distinct clinical subtypes of Phelan-McDermid syndrome. *Clinical Epigenetics*, **2021**, 13, 2	7.7	4
41	Epigenomic Mechanisms of Human Developmental Disorders **2018**, 837-859		4
40	Genomics and epigenomics in pediatric oncology and clinical laboratory genetics. *Clinical Biochemistry*, **2014**, 47, 731-2	3.5	3
39	CPEO - Like mitochondrial myopathy associated with m.8340G>A mutation. *Mitochondrion*, **2019**, 46, 69-72	4.9	3
38	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. *Genetics in Medicine*, **2021**, 23, 2122-2137	8.1	3
37	Multisite verification of the accuracy of a multi-gene next generation sequencing panel for detection of mutations and copy number alterations in solid tumours. *PLoS ONE*, **2021**, 16, e0258188	3.7	3
36	Validation and clinical performance of a combined nuclear-mitochondrial next-generation sequencing and copy number variant analysis panel in a Canadian population. *American Journal of Medical Genetics, Part A*, **2021**, 185, 486-499	2.5	2
35	EpiSigns. *Advances in Molecular Pathology*, **2020**, 3, 29-39	0.3	2
34	Bone marrow-derived mitochondrial DNA has limited capacity for inter-tissue transfer in vivo. *FASEB Journal*, **2020**, 34, 9297-9306	0.9	2
33	Genomic data in prognostic models-what is lost in translation? The case of deletion 17p and mutant TP53 in chronic lymphocytic leukaemia. *British Journal of Haematology*, **2020**, 188, 652-660	4.5	2
32	Comprehensive genetic sequence and copy number analysis for Charcot-Marie-Tooth disease in a Canadian cohort of 2517 patients. *Journal of Medical Genetics*, **2021**, 58, 284-288	5.8	2
31	A 42-year-old man with elevated ferritin. *Cmaj*, **2015**, 187, 820-821	3.5	1
30	Epigenetic Mediation of Environmental Exposures to Polycyclic Aromatic Hydrocarbons **2012**, 111-127		1
29	Clinical findings and a DNA methylation signature in kindreds with alterations in ZNF711.. *European Journal of Human Genetics*, **2022**,	5.3	1
28	Serum erythropoietin levels in 696 patients investigated for erythrocytosis with JAK2 mutation analysis.. *American Journal of Hematology*, **2022**,	7.1	1

27	DNA methylation episignature testing improves molecular diagnosis of Mendelian chromatinopathies.. *Genetics in Medicine*, **2021**,	8.1	1
26	A Prediction Rule to Guide JAK2 Testing in Patients with Suspected Polycythemia Vera. *Blood*, **2021**, 138, 4635-4635	2.2	1
25	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders.. *Human Genetics and Genomics Advances*, **2022**, 3, 100075	0.8	1
24	Reducing cytogenetic testing in the era of next generation sequencing: Are we choosing wisely?. *International Journal of Laboratory Hematology*, **2021**,	2.5	1
23	Complete elimination of a pathogenic homoplasmic mtDNA mutation in one generation. *Mitochondrion*, **2019**, 45, 18-21	4.9	1
22	Genetic Testing in Children with Epilepsy: Report of a Single-Center Experience. *Canadian Journal of Neurological Sciences*, **2021**, 48, 233-244	1	1
21	Discovery of a novel CHD7 CHARGE syndrome variant by integrated omics analyses. *American Journal of Medical Genetics, Part A*, **2021**, 185, 544-548	2.5	1
20	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
19	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
18	Incidental findings from cancer next generation sequencing panels. *Npj Genomic Medicine*, **2021**, 6, 63	6.2	1
17	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. *Npj Genomic Medicine*, **2021**, 6, 92	6.2	0
16	Investigating Erythrocytosis: Changing Practice Patterns in the Era of Molecular Diagnostics. *Blood*, **2021**, 138, 4630-4630	2.2	0
15	The Impact of Artificial Intelligence on Health Equity in Oncology: A Scoping Review. *Blood*, **2021**, 138, 4934-4934	2.2	0
14	Consensus Recommendations for MRD Testing in Adult B-Cell Acute Lymphoblastic Leukemia in Ontario. *Current Oncology*, **2021**, 28, 1376-1387	2.8	0
13	Analysis of Sequence and Copy Number Variants in Canadian Patient Cohort With Familial Cancer Syndromes Using a Unique Next Generation Sequencing Based Approach. *Frontiers in Genetics*, **2021**, 12, 698595	4.5	0
12	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. *Clinical Epigenetics*, **2021**, 13, 157	7.7	0
11	Genome-wide DNA methylation profiling confirms a case of low-level mosaic Kabuki syndrome 1.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
10	Clinical Utility of Implementing a Frontline NGS-Based DNA and RNA Fusion Panel Test for Patients with Suspected Myeloid Malignancies.. *Molecular Diagnosis and Therapy*, **2022**, 1	4.5	0

9	Porphyria cutanea tarda associated with elevated serum ferritin, iron overload, and a bone morphogenetic protein 6 genetic variant. *Canadian Liver Journal*, **2020**, 3, 232-234	0.3
8	Identifying Myeloid Mutations By NGS in Patients with Unexplained Erythrocytosis. *Blood*, **2020**, 136, 47-47	2.2
7	Reducing Cytogenetic Testing in the Era of Next Generation Sequencing (NGS); Are We Choosing Wisely?. *Blood*, **2020**, 136, 12-13	2.2
6	Near complete deletion of KMT2D in a college student.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5
5	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
4	The Clinical Application of Oncomine Myeloid Next Generation Sequencing (NGS): Comparison of Cytogenetics and NGS in Patients with Suspected Myelodysplastic Syndrome. *Blood*, **2019**, 134, 3375-3375	2.2
3	Retrospective Evaluation of Patients Referred for Hemochromatosis Genetic Testing. *Blood*, **2014**, 124, 4035-4035	2.2
2	A case of congenital prothrombin deficiency with two concurrent mutations in the prothrombin gene. *Research and Practice in Thrombosis and Haemostasis*, **2021**, 5, e12510	5.1
1	A Pan-Canadian Validation Study for the Detection of T790M Mutation Using Circulating Tumor DNA From Peripheral Blood. *JTO Clinical and Research Reports*, **2021**, 2, 100212	1.4