

Bekim Sadikovic

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Clinical findings and a DNA methylation signature in kindreds with alterations in ZNF711.. <i>European Journal of Human Genetics</i> , 2022 ,	5.3	1
97	Near complete deletion of KMT2D in a college student.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	
96	Serum erythropoietin levels in 696 patients investigated for erythrocytosis with JAK2 mutation analysis.. <i>American Journal of Hematology</i> , 2022 ,	7.1	1
95	Novel diagnostic DNA methylation epesignatures expand and refine the epigenetic landscapes of Mendelian disorders.. <i>Human Genetics and Genomics Advances</i> , 2022 , 3, 100075	0.8	1
94	Genome-wide DNA methylation profiling confirms a case of low-level mosaic Kabuki syndrome 1.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	0
93	Clinical Utility of Implementing a Frontline NGS-Based DNA and RNA Fusion Panel Test for Patients with Suspected Myeloid Malignancies.. <i>Molecular Diagnosis and Therapy</i> , 2022 , 1	4.5	0
92	Validation and clinical performance of a combined nuclear-mitochondrial next-generation sequencing and copy number variant analysis panel in a Canadian population. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 486-499	2.5	2
91	Deficiency of TET3 leads to a genome-wide DNA hypermethylation epesignature in human whole blood. <i>Npj Genomic Medicine</i> , 2021 , 6, 92	6.2	0
90	DNA methylation epesignature testing improves molecular diagnosis of Mendelian chromatinopathies.. <i>Genetics in Medicine</i> , 2021 ,	8.1	1
89	Investigating Erythrocytosis: Changing Practice Patterns in the Era of Molecular Diagnostics. <i>Blood</i> , 2021 , 138, 4630-4630	2.2	0
88	A Prediction Rule to Guide JAK2 Testing in Patients with Suspected Polycythemia Vera. <i>Blood</i> , 2021 , 138, 4635-4635	2.2	1
87	The Impact of Artificial Intelligence on Health Equity in Oncology: A Scoping Review. <i>Blood</i> , 2021 , 138, 4934-4934	2.2	0
86	Reducing cytogenetic testing in the era of next generation sequencing: Are we choosing wisely?. <i>International Journal of Laboratory Hematology</i> , 2021 ,	2.5	1
85	Consensus Recommendations for MRD Testing in Adult B-Cell Acute Lymphoblastic Leukemia in Ontario. <i>Current Oncology</i> , 2021 , 28, 1376-1387	2.8	0
84	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epesignature of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021 , 108, 502-516	11	12
83	Cross-oncopanel study reveals high sensitivity and accuracy with overall analytical performance depending on genomic regions. <i>Genome Biology</i> , 2021 , 22, 109	18.3	6
82	A case of congenital prothrombin deficiency with two concurrent mutations in the prothrombin gene. <i>Research and Practice in Thrombosis and Haemostasis</i> , 2021 , 5, e12510	5.1	

81	Comprehensive genetic sequence and copy number analysis for Charcot-Marie-Tooth disease in a Canadian cohort of 2517 patients. <i>Journal of Medical Genetics</i> , 2021 , 58, 284-288	5.8	2
80	Clinical value of next-generation sequencing compared to cytogenetics in patients with suspected myelodysplastic syndrome. <i>British Journal of Haematology</i> , 2021 , 192, 729-736	4.5	5
79	Genetic Testing in Children with Epilepsy: Report of a Single-Center Experience. <i>Canadian Journal of Neurological Sciences</i> , 2021 , 48, 233-244	1	1
78	Discovery of a novel CHD7 CHARGE syndrome variant by integrated omics analyses. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 544-548	2.5	1
77	Clinical and technical assessment of MedExome vs. NGS panels in patients with suspected genetic disorders in Southwestern Ontario. <i>Journal of Human Genetics</i> , 2021 , 66, 451-464	4.3	1
76	Detection of a DNA Methylation Signature for the Intellectual Developmental Disorder, X-Linked, Syndromic, Armfield Type. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	1
75	DNA methylation epi-signature is associated with two molecularly and phenotypically distinct clinical subtypes of Phelan-McDermid syndrome. <i>Clinical Epigenetics</i> , 2021 , 13, 2	7.7	4
74	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. <i>Genetics in Medicine</i> , 2021 , 23, 1065-1074	8.1	10
73	Incidental findings from cancer next generation sequencing panels. <i>Npj Genomic Medicine</i> , 2021 , 6, 63	6.2	1
72	Analysis of Sequence and Copy Number Variants in Canadian Patient Cohort With Familial Cancer Syndromes Using a Unique Next Generation Sequencing Based Approach. <i>Frontiers in Genetics</i> , 2021 , 12, 698595	4.5	0
71	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. <i>Genetics in Medicine</i> , 2021 , 23, 2122-2137	8.1	3
70	A Pan-Canadian Validation Study for the Detection of T790M Mutation Using Circulating Tumor DNA From Peripheral Blood. <i>JTO Clinical and Research Reports</i> , 2021 , 2, 100212	1.4	
69	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. <i>Clinical Epigenetics</i> , 2021 , 13, 157	7.7	0
68	Multisite verification of the accuracy of a multi-gene next generation sequencing panel for detection of mutations and copy number alterations in solid tumours. <i>PLoS ONE</i> , 2021 , 16, e0258188	3.7	3
67	Diagnostic Utility of Genome-Wide DNA Methylation Analysis in Mendelian Neurodevelopmental Disorders. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	7
66	Genetic and epigenetic profiling of BRCA1/2 in ovarian tumors reveals additive diagnostic yield and evidence of a genomic BRCA1/2 DNA methylation signature. <i>Journal of Human Genetics</i> , 2020 , 65, 865-873	4.3	5
65	Glucose-induced, duration-dependent genome-wide DNA methylation changes in human endothelial cells. <i>American Journal of Physiology - Cell Physiology</i> , 2020 , 319, C268-C276	5.4	5
64	Porphyria cutanea tarda associated with elevated serum ferritin, iron overload, and a bone morphogenetic protein 6 genetic variant. <i>Canadian Liver Journal</i> , 2020 , 3, 232-234	0.3	

63	Functional annotation of genomic variation: DNA methylation epesignatures in neurodevelopmental Mendelian disorders. <i>Human Molecular Genetics</i> , 2020 , 29, R27-R32	5.6	9
62	Evaluation of DNA Methylation Epesignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020 , 106, 356-370	11	51
61	Identifying Myeloid Mutations By NGS in Patients with Unexplained Erythrocytosis. <i>Blood</i> , 2020 , 136, 47-47	2.2	
60	Reducing Cytogenetic Testing in the Era of Next Generation Sequencing (NGS); Are We Choosing Wisely?. <i>Blood</i> , 2020 , 136, 12-13	2.2	
59	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.. <i>Journal of Clinical Oncology</i> , 2020 , 38, 20-20	2.2	
58	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. <i>Clinical Epigenetics</i> , 2020 , 12, 7	7.7	23
57	EpiSigns. <i>Advances in Molecular Pathology</i> , 2020 , 3, 29-39	0.3	2
56	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome. <i>Genetics in Medicine</i> , 2020 , 22, 1838-1850	8.1	8
55	Bone marrow-derived mitochondrial DNA has limited capacity for inter-tissue transfer in vivo. <i>FASEB Journal</i> , 2020 , 34, 9297-9306	0.9	2
54	Genomic data in prognostic models-what is lost in translation? The case of deletion 17p and mutant TP53 in chronic lymphocytic leukaemia. <i>British Journal of Haematology</i> , 2020 , 188, 652-660	4.5	2
53	Implementation of Epilepsy Multigene Panel Testing in Ontario, Canada. <i>Canadian Journal of Neurological Sciences</i> , 2020 , 47, 61-68	1	4
52	Genotypes of chronic progressive external ophthalmoplegia in a large adult-onset cohort. <i>Mitochondrion</i> , 2019 , 49, 227-231	4.9	13
51	Implementation of an NGS-based sequencing and gene fusion panel for clinical screening of patients with suspected hematologic malignancies. <i>European Journal of Haematology</i> , 2019 , 103, 178-189	3.8	16
50	Gene domain-specific DNA methylation epesignatures highlight distinct molecular entities of ADNP syndrome. <i>Clinical Epigenetics</i> , 2019 , 11, 64	7.7	29
49	DNA methylation signatures in mendelian developmental disorders as a diagnostic bridge between genotype and phenotype. <i>Epigenomics</i> , 2019 , 11, 563-575	4.4	21
48	Diagnostic Utility of Genome-wide DNA Methylation Testing in Genetically Unsolved Individuals with Suspected Hereditary Conditions. <i>American Journal of Human Genetics</i> , 2019 , 104, 685-700	11	57
47	Screening for genes that accelerate the epigenetic aging clock in humans reveals a role for the H3K36 methyltransferase NSD1. <i>Genome Biology</i> , 2019 , 20, 146	18.3	36
46	Genome-wide DNA methylation and RNA analyses enable reclassification of two variants of uncertain significance in a patient with clinical Kabuki syndrome. <i>Human Mutation</i> , 2019 , 40, 1684-1689	4.7	12

45	The Clinical Application of Oncomine Myeloid Next Generation Sequencing (NGS): Comparison of Cytogenetics and NGS in Patients with Suspected Myelodysplastic Syndrome. <i>Blood</i> , 2019 , 134, 3375-3375 ²		
44	Complete elimination of a pathogenic homoplasmic mtDNA mutation in one generation. <i>Mitochondrion</i> , 2019 , 45, 18-21	4.9	1
43	CPEO - Like mitochondrial myopathy associated with m.8340G>A mutation. <i>Mitochondrion</i> , 2019 , 46, 69-72	4.9	3
42	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. <i>American Journal of Human Genetics</i> , 2018 , 102, 156-174	11.4	75
41	Peripheral blood epi-signature of Claes-Jensen syndrome enables sensitive and specific identification of patients and healthy carriers with pathogenic mutations in. <i>Clinical Epigenetics</i> , 2018 , 10, 21	7.7	37
40	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). <i>Genetics in Medicine</i> , 2018 , 20, 294-302	8.1	20
39	Genomic DNA Methylation-Derived Algorithm Enables Accurate Detection of Malignant Prostate Tissues. <i>Frontiers in Oncology</i> , 2018 , 8, 100	5.3	25
38	BAFopathiesDNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin-Siris and Nicolaides-Baraitser syndromes. <i>Nature Communications</i> , 2018 , 9, 4885	17.4	48
37	The ClinGen Epilepsy Gene Curation Expert Panel-Bridging the divide between clinical domain knowledge and formal gene curation criteria. <i>Human Mutation</i> , 2018 , 39, 1476-1484	4.7	21
36	Epigenomic Mechanisms of Human Developmental Disorders 2018 , 837-859		4
35	Constitutional Epi/Genetic Conditions: Genetic, Epigenetic, and Environmental Factors. <i>Journal of Pediatric Genetics</i> , 2017 , 6, 30-41	0.7	13
34	The defining DNA methylation signature of Kabuki syndrome enables functional assessment of genetic variants of unknown clinical significance. <i>Epigenetics</i> , 2017 , 12, 923-933	5.7	43
33	Clinical Validation of a Genome-Wide DNA Methylation Assay for Molecular Diagnosis of Imprinting Disorders. <i>Journal of Molecular Diagnostics</i> , 2017 , 19, 848-856	5.1	29
32	Clinical Validation of Copy Number Variant Detection from Targeted Next-Generation Sequencing Panels. <i>Journal of Molecular Diagnostics</i> , 2017 , 19, 905-920	5.1	75
31	Identification of epigenetic signature associated with alpha thalassemia/mental retardation X-linked syndrome. <i>Epigenetics and Chromatin</i> , 2017 , 10, 10	5.8	39
30	Clinical evaluation of a hemochromatosis next-generation sequencing gene panel. <i>European Journal of Haematology</i> , 2017 , 98, 228-234	3.8	15
29	DNA methylation analysis in constitutional disorders: Clinical implications of the epigenome. <i>Critical Reviews in Clinical Laboratory Sciences</i> , 2016 , 53, 147-65	9.4	21
28	Clinical Next-Generation Sequencing Pipeline Outperforms a Combined Approach Using Sanger Sequencing and Multiplex Ligation-Dependent Probe Amplification in Targeted Gene Panel Analysis. <i>Journal of Molecular Diagnostics</i> , 2016 , 18, 657-667	5.1	36

27	Gender and BCR-ABL transcript type are correlated with molecular response to imatinib treatment in patients with chronic myeloid leukemia. <i>European Journal of Haematology</i> , 2016 , 96, 360-6	3.8	28
26	Movement Disorders Associated With Hemochromatosis. <i>Canadian Journal of Neurological Sciences</i> , 2016 , 43, 801-808	1	11
25	The defining DNA methylation signature of Floating-Harbor Syndrome. <i>Scientific Reports</i> , 2016 , 6, 38803	4.9	38
24	Clinical Validation of Fragile X Syndrome Screening by DNA Methylation Array. <i>Journal of Molecular Diagnostics</i> , 2016 , 18, 834-841	5.1	28
23	Identification of a methylation profile for DNMT1-associated autosomal dominant cerebellar ataxia, deafness, and narcolepsy. <i>Clinical Epigenetics</i> , 2016 , 8, 91	7.7	50
22	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. <i>Journal of Medical Genetics</i> , 2015 , 52, 431-7	5.8	137
21	A 42-year-old man with elevated ferritin. <i>Cmaj</i> , 2015 , 187, 820-821	3.5	1
20	NLRP7 affects trophoblast lineage differentiation, binds to overexpressed YY1 and alters CpG methylation. <i>Human Molecular Genetics</i> , 2014 , 23, 706-16	5.6	46
19	Genomics and epigenomics in pediatric oncology and clinical laboratory genetics. <i>Clinical Biochemistry</i> , 2014 , 47, 731-2	3.5	3
18	Mutation Update for UBE3A variants in Angelman syndrome. <i>Human Mutation</i> , 2014 , 35, 1407-17	4.7	38
17	Recurrent genomic alterations in sequential progressive leukoplakia and oral cancer: drivers of oral tumorigenesis?. <i>Human Molecular Genetics</i> , 2014 , 23, 2618-28	5.6	36
16	Retrospective Evaluation of Patients Referred for Hemochromatosis Genetic Testing. <i>Blood</i> , 2014 , 124, 4035-4035	2.2	
15	Array comparative genomic hybridization in osteosarcoma. <i>Methods in Molecular Biology</i> , 2013 , 973, 227-47	4.7	10
14	Epigenetic Mediation of Environmental Exposures to Polycyclic Aromatic Hydrocarbons 2012 , 111-127		1
13	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012 , 109, 7974-81	11.5	94
12	Chronic maternal protein deprivation in mice is associated with overexpression of the cohesin-mediator complex in liver of their offspring. <i>Journal of Nutrition</i> , 2011 , 141, 2106-12	4.1	8
11	Immunohistochemical expression and cluster analysis of mesenchymal and neural stem cell-associated proteins in pediatric soft tissue sarcomas. <i>Pediatric and Developmental Pathology</i> , 2011 , 14, 259-72	2.2	10
10	Sequence homology at the breakpoint and clinical phenotype of mitochondrial DNA deletion syndromes. <i>PLoS ONE</i> , 2010 , 5, e15687	3.7	43

9	Discovery of novel hypermethylated genes in prostate cancer using genomic CpG island microarrays. <i>PLoS ONE</i> , 2009 , 4, e4830	3.7	74
8	Identification of a microRNA signature associated with progression of leukoplakia to oral carcinoma. <i>Human Molecular Genetics</i> , 2009 , 18, 4818-29	5.6	187
7	Identification of interactive networks of gene expression associated with osteosarcoma oncogenesis by integrated molecular profiling. <i>Human Molecular Genetics</i> , 2009 , 18, 1962-75	5.6	100
6	In vitro analysis of integrated global high-resolution DNA methylation profiling with genomic imbalance and gene expression in osteosarcoma. <i>PLoS ONE</i> , 2008 , 3, e2834	3.7	66
5	Decitabine-induced demethylation of 5RCpG island in GADD45A leads to apoptosis in osteosarcoma cells. <i>Neoplasia</i> , 2008 , 10, 471-80	6.4	45
4	Genome-wide H3K9 histone acetylation profiles are altered in benzopyrene-treated MCF7 breast cancer cells. <i>Journal of Biological Chemistry</i> , 2008 , 283, 4051-60	5.4	60
3	DNA methylation analysis using CpG microarrays is impaired in benzopyrene exposed cells. <i>Toxicology and Applied Pharmacology</i> , 2007 , 225, 300-9	4.6	23
2	Benzopyrene exposure disrupts DNA methylation and growth dynamics in breast cancer cells. <i>Toxicology and Applied Pharmacology</i> , 2006 , 216, 458-68	4.6	101
1	Characterization of functional elements in the neurofibromatosis (NF1) proximal promoter region. <i>Oncogene</i> , 2004 , 23, 330-9	9.2	20