Bekim Sadikovic

List of Publications by Year
in descending order

110 papers	3,536 citations	117571 34 h-index	161767 54 g-index
118 all docs	118 docs citations	118 times ranked	5249 citing authors

#	Article	IF	Citations
1	Identification of a microRNA signature associated with progression of leukoplakia to oral carcinoma. Human Molecular Genetics, 2009, 18, 4818-4829.	1.4	223
2	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, 431-437.	1.5	187
3	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	2.6	171
4	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. American Journal of Human Genetics, 2018, 102, 156-174.	2.6	135
5	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.	2.6	125
6	Identification of interactive networks of gene expression associated with osteosarcoma oncogenesis by integrated molecular profiling. Human Molecular Genetics, 2009, 18, 1962-1975.	1.4	119
7	Benzopyrene exposure disrupts DNA methylation and growth dynamics in breast cancer cells. Toxicology and Applied Pharmacology, 2006, 216, 458-468.	1.3	118
8	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, 7974-7981.	3.3	118
9	Clinical Validation of Copy Number Variant Detection from Targeted Next-Generation Sequencing Panels. Journal of Molecular Diagnostics, 2017, 19, 905-920.	1.2	104
10	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. Genetics in Medicine, 2021, 23, 1065-1074.	1.1	88
11	BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885.	5.8	83
12	Discovery of Novel Hypermethylated Genes in Prostate Cancer Using Genomic CpG Island Microarrays. PLoS ONE, 2009, 4, e4830.	1.1	81
13	The defining DNA methylation signature of Kabuki syndrome enables functional assessment of genetic variants of unknown clinical significance. Epigenetics, 2017, 12, 923-933.	1.3	79
14	Genome-wide H3K9 Histone Acetylation Profiles Are Altered in Benzopyrene-treated MCF7 Breast Cancer Cells. Journal of Biological Chemistry, 2008, 283, 4051-4060.	1.6	72
15	In Vitro Analysis of Integrated Global High-Resolution DNA Methylation Profiling with Genomic Imbalance and Gene Expression in Osteosarcoma. PLoS ONE, 2008, 3, e2834.	1.1	71
16	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64.	1.8	71
17	Identification of a methylation profile for DNMT1-associated autosomal dominant cerebellar ataxia, deafness, and narcolepsy. Clinical Epigenetics, 2016, 8, 91.	1.8	66
18	Screening for genes that accelerate the epigenetic aging clock in humans reveals a role for the H3K36 methyltransferase NSD1. Genome Biology, 2019, 20, 146.	3.8	66

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19	Identification of epigenetic signature associated with alpha thalassemia/mental retardation X-linked syndrome. Epigenetics and Chromatin, 2017, 10, 10.	1.8	60
20	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.	1.8	58
21	Mutation Update for UBE3A Variants in Angelman Syndrome. Human Mutation, 2014, 35, 1407-1417.	1.1	56
22	The defining DNA methylation signature of Floating-Harbor Syndrome. Scientific Reports, 2016, 6, 38803.	1.6	55
23	Decitabine-Induced Demethylation of 5â€² CpG Island in GADD45A Leads to Apoptosis in Osteosarcoma Cells. Neoplasia, 2008, 10, 471-480.	2.3	54
24	NLRP7 affects trophoblast lineage differentiation, binds to overexpressed YY1 and alters CpG methylation. Human Molecular Genetics, 2014, 23, 706-716.	1.4	54
25	Sequence Homology at the Breakpoint and Clinical Phenotype of Mitochondrial DNA Deletion Syndromes. PLoS ONE, 2010, 5, e15687.	1.1	49
26	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.	2.6	48
27	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.	1.2	47
28	Recurrent genomic alterations in sequential progressive leukoplakia and oral cancer: drivers of oral tumorigenesis?. Human Molecular Genetics, 2014, 23, 2618-2628.	1.4	46
29	DNA methylation signatures in mendelian developmental disorders as a diagnostic bridge between genotype and phenotype. Epigenomics, 2019, 11, 563-575.	1.0	42
30	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
31	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	1.8	40
32	Clinical Validation of a Genome-Wide DNA Methylation Assay for Molecular Diagnosis of Imprinting Disorders. Journal of Molecular Diagnostics, 2017, 19, 848-856.	1.2	39
33	Genomic DNA Methylation-Derived Algorithm Enables Accurate Detection of Malignant Prostate Tissues. Frontiers in Oncology, 2018, 8, 100.	1.3	38
34	Clinical Validation of Fragile X Syndrome Screening by DNA Methylation Array. Journal of Molecular Diagnostics, 2016, 18, 834-841.	1.2	37
35	Gender and <scp>BCR</scp>â€<scp>ABL</scp> transcript type are correlated with molecular response to imatinib treatment in patients with chronic myeloid leukemia. European Journal of Haematology, 2016, 96, 360-366.	1.1	35
36	The ClinGen Epilepsy Gene Curation Expert Panelâ€”Bridging the divide between clinical domain knowledge and formal gene curation criteria. Human Mutation, 2018, 39, 1476-1484.	1.1	33

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37	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.	1.1	31
38	DNA methylation analysis in constitutional disorders: Clinical implications of the epigenome. Critical Reviews in Clinical Laboratory Sciences, 2016, 53, 147-165.	2.7	28
39	DNA methylation analysis using CpG microarrays is impaired in benzopyrene exposed cells. Toxicology and Applied Pharmacology, 2007, 225, 300-309.	1.3	27
40	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.	1.1	27
41	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.	1.1	27
42	DNA methylation epi-signature is associated with two molecularly and phenotypically distinct clinical subtypes of Phelan-McDermid syndrome. Clinical Epigenetics, 2021, 13, 2.	1.8	27
43	DNA methylation episignature testing improves molecular diagnosis of Mendelian chromatinopathies. Genetics in Medicine, 2022, 24, 51-60.	1.1	24
44	Diagnostic Utility of Genome-Wide DNA Methylation Analysis in Mendelian Neurodevelopmental Disorders. International Journal of Molecular Sciences, 2020, 21, 9303.	1.8	23
45	Functional annotation of genomic variation: DNA methylation episignatures in neurodevelopmental Mendelian disorders. Human Molecular Genetics, 2020, 29, R27-R32.	1.4	23
46	Characterization of functional elements in the neurofibromatosis (NF1) proximal promoter region. Oncogene, 2004, 23, 330-339.	2.6	22
47	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	1.8	22
48	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.	1.1	21
49	Clinical evaluation of a hemochromatosis nextâ€generation sequencing gene panel. European Journal of Haematology, 2017, 98, 228-234.	1.1	20
50	Genotypes of chronic progressive external ophthalmoplegia in a large adult-onset cohort. Mitochondrion, 2019, 49, 227-231.	1.6	20
51	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.	1.5	20
52	Cross-oncopanel study reveals high sensitivity and accuracy with overall analytical performance depending on genomic regions. Genome Biology, 2021, 22, 109.	3.8	20
53	Movement Disorders Associated With Hemochromatosis. Canadian Journal of Neurological Sciences, 2016, 43, 801-808.	0.3	19
54	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16

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55	Constitutional Epi/Genetic Conditions: Genetic, Epigenetic, and Environmental Factors. Journal of Pediatric Genetics, 2017, 06, 030-041.	0.3	15
56	Identification of a DNA Methylation Episignature in the 22q11.2 Deletion Syndrome. International Journal of Molecular Sciences, 2021, 22, 8611.	1.8	15
57	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	1.1	14
58	Genomeâ€wide <scp>DNA</scp> methylation profiling confirms a case of lowâ€level mosaic Kabuki syndrome 1. American Journal of Medical Genetics, Part A, 2022, 188, 2217-2225.	0.7	14
59	Array Comparative Genomic Hybridization in Osteosarcoma. Methods in Molecular Biology, 2013, 973, 227-247.	0.4	12
60	Immunohistochemical Expression and Cluster Analysis of Mesenchymal and Neural Stem Cellâ€“Associated Proteins in Pediatric Soft Tissue Sarcomas. Pediatric and Developmental Pathology, 2011, 14, 259-272.	0.5	11
61	Incidental findings from cancer next generation sequencing panels. Npj Genomic Medicine, 2021, 6, 63.	1.7	11
62	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. Npj Genomic Medicine, 2021, 6, 92.	1.7	11
63	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.	1.1	10
64	Glucose-induced, duration-dependent genome-wide DNA methylation changes in human endothelial cells. American Journal of Physiology - Cell Physiology, 2020, 319, C268-C276.	2.1	10
65	Discovery of a novel <scp>CHD7 CHARGE</scp> syndrome variant by integrated omics analyses. American Journal of Medical Genetics, Part A, 2021, 185, 544-548.	0.7	10
66	Detection of a DNA Methylation Signature for the Intellectual Developmental Disorder, X-Linked, Syndromic, Armfield Type. International Journal of Molecular Sciences, 2021, 22, 1111.	1.8	10
67	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, 2106-2112.	1.3	8
68	Clinical value of nextâ€generation sequencing compared to cytogenetics in patients with suspected myelodysplastic syndrome. British Journal of Haematology, 2021, 192, 729-736.	1.2	8
69	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.	1.1	8
70	CDK13-related disorder: Report of a series of 18 previously unpublished individuals and description of an epigenetic signature. Genetics in Medicine, 2022, 24, 1096-1107.	1.1	8
71	Clinical Utility of a Unique Genome-Wide DNA Methylation Signature for KMT2A-Related Syndrome. International Journal of Molecular Sciences, 2022, 23, 1815.	1.8	8
72	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, 26, 333-343.	1.6	8

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73	DNA Methylation Episignatures in Neurodevelopmental Disorders Associated with Large Structural Copy Number Variants: Clinical Implications. International Journal of Molecular Sciences, 2022, 23, 7862.	1.8	8
74	Validation and clinical performance of a combined nuclearâ€mitochondrial nextâ€generation sequencing and copy number variant analysis panel in a Canadian population. American Journal of Medical Genetics, Part A, 2020, 185, 486-499.	0.7	7
75	Consensus Recommendations for MRD Testing in Adult B-Cell Acute Lymphoblastic Leukemia in Ontario. Current Oncology, 2021, 28, 1376-1387.	0.9	7
76	Clinical findings and a DNA methylation signature in kindreds with alterations in ZNF711. European Journal of Human Genetics, 2022, 30, 420-427.	1.4	7
77	Implementation of Epilepsy Multigene Panel Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2020, 47, 61-68.	0.3	6
78	DNA methylation episignature in Gabriele-de VriesÂsyndrome. Genetics in Medicine, 2022, 24, 905-914.	1.1	6
79	CPEO â€“ Like mitochondrial myopathy associated with m.8340G>A mutation. Mitochondrion, 2019, 46, 69-72.	1.6	5
80	Bone marrowâ€derived mitochondrial DNA has limited capacity for interâ€tissue transfer in vivo. FASEB Journal, 2020, 34, 9297-9306.	0.2	5
81	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. Genes, 2021, 12, 1275.	1.0	5
82	Epigenomic Mechanisms of Human Developmental Disorders. , 2018, , 837-859.		4
83	Genetic Testing in Children with Epilepsy: Report of a Single-Center Experience. Canadian Journal of Neurological Sciences, 2021, 48, 233-244.	0.3	4
84	Contribution of DNA methylation profiling to the reclassification of a variant of uncertain significance in the KDM5C gene. European Journal of Medical Genetics, 2022, 65, 104556.	0.7	4
85	Genomics and epigenomics in pediatric oncology and clinical laboratory genetics. Clinical Biochemistry, 2014, 47, 731-732.	0.8	3
86	EpiSigns. Advances in Molecular Pathology, 2020, 3, 29-39.	0.2	3
87	Reducing cytogenetic testing in the era of next generation sequencing: Are we choosing wisely?. International Journal of Laboratory Hematology, 2022, 44, 333-341.	0.7	3
88	Near complete deletion of <scp>KMT2D</scp> in a college student. American Journal of Medical Genetics, Part A, 2022, 188, 1550-1555.	0.7	3
89	Serum erythropoietin levels in 696 patients investigated for erythrocytosis with <scp>JAK2</scp> mutation analysis. American Journal of Hematology, 2022, 97, .	2.0	3
90	Genome-wide DNA methylation profiling and exome sequencing resolved a long-time misdiagnosed case. Journal of Human Genetics, 2022, 67, 547-551.	1.1	3

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91	Genomic data in prognostic modelsâ€"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.	1.2	2
92	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.	1.1	2
93	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.	1.1	2
94	A Pan-Canadian Validation Study for the Detection of EGFR T790M Mutation Using Circulating Tumor DNA From Peripheral Blood. JTO Clinical and Research Reports, 2021, 2, 100212.	0.6	2
95	Investigating Erythrocytosis: Changing Practice Patterns in the Era of Molecular Diagnostics. Blood, 2021, 138, 4630-4630.	0.6	2
96	The Impact of Artificial Intelligence on Health Equity in Oncology: A Scoping Review. Blood, 2021, 138, 4934-4934.	0.6	2
97	A 42-year-old man with elevated ferritin. Cmaj, 2015, 187, 820-821.	0.9	1
98	Complete elimination of a pathogenic homoplasmic mtDNA mutation in one generation. Mitochondrion, 2019, 45, 18-21.	1.6	1
99	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	1
100	A Prediction Rule to Guide JAK2 Testing in Patients with Suspected Polycythemia Vera. Blood, 2021, 138, 4635-4635.	0.6	1
101	Utility of Genomic Methylation Microarrays to Measure Spreading of X-Inactivation in Association with X;Autosome Translocations. Cancer Genetics, 2016, 209, 299.	0.2	0
102	Novel pathogenic sequence variants in Yin Yang 1 (YY1) transcription factor and abnormal DNA methylation profile. Molecular Genetics and Metabolism, 2021, 132, S254-S255.	0.5	0
103	A case of congenital prothrombin deficiency with two concurrent mutations in the prothrombin gene. Research and Practice in Thrombosis and Haemostasis, 2021, 5, e12510.	1.0	0
104	Retrospective Evaluation of Patients Referred for Hemochromatosis Genetic Testing. Blood, 2014, 124, 4035-4035.	0.6	0
105	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.	0.6	0
106	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.	0.8	0
107	Identifying Myeloid Mutations By NGS in Patients with Unexplained Erythrocytosis. Blood, 2020, 136, 47-47.	0.6	0
108	Reducing Cytogenetic Testing in the Era of Next Generation Sequencing (NGS); Are We Choosing Wisely?. Blood, 2020, 136, 12-13.	0.6	0

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109	eP159: A TOPBP1 variant associated With BAFopathy methylation pattern. Genetics in Medicine, 2022, 24, S97.	1.1	0
110	eP248: Impact of DNA methylation signature exploration for variants of uncertain significance within epigenetic syndromes. Genetics in Medicine, 2022, 24, S157-S158.	1.1	0