Jennifer Kerkhof

List of Publications by Year
in descending order

31 papers	978 citations	623734 14 h-index	501196 28 g-index
33 all docs	33 docs citations	33 times ranked	1517 citing authors

#	Article	IF	Citations
1	DNA methylation episignature testing improves molecular diagnosis of Mendelian chromatinopathies. Genetics in Medicine, 2022, 24, 51-60.	2.4	24
2	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
3	Clinical findings and a DNA methylation signature in kindreds with alterations in ZNF711. European Journal of Human Genetics, 2022, 30, 420-427.	2.8	7
4	Near complete deletion of <scp>KMT2D</scp> in a college student. American Journal of Medical Genetics, Part A, 2022, 188, 1550-1555.	1.2	3
5	CDK13-related disorder: Report of a series of 18 previously unpublished individuals and description of an epigenetic signature.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.	2.4	8
6	Clinical Utility of a Unique Genome-Wide DNA Methylation Signature for KMT2A-Related Syndrome. International Journal of Molecular Sciences, 2022, 23, 1815.	4.1	8
7	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.	2.4	14
8	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.	1.2	14
9	Genome-wide DNA methylation profiling and exome sequencing resolved a long-time misdiagnosed case. Journal of Human Genetics, 2022, 67, 547-551.	2.3	3
10	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.	3.2	20
11	Clinical value of nextâ€generation sequencing compared to cytogenetics in patients with suspected myelodysplastic syndrome. British Journal of Haematology, 2021, 192, 729-736.	2.5	8
12	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.	2.3	2
13	Detection of a DNA Methylation Signature for the Intellectual Developmental Disorder, X-Linked, Syndromic, Armfield Type. International Journal of Molecular Sciences, 2021, 22, 1111.	4.1	10
14	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. Genetics in Medicine, 2021, 23, 1065-1074.	2.4	88
15	Cross-oncopanel study reveals high sensitivity and accuracy with overall analytical performance depending on genomic regions. Genome Biology, 2021, 22, 109.	8.8	20
16	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.	2.3	2
17	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
18	Identification of a DNA Methylation Episignature in the 22q11.2 Deletion Syndrome. International Journal of Molecular Sciences, 2021, 22, 8611.	4.1	15

#	Article	IF	Citations
19	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. Npj Genomic Medicine, 2021, 6, 92.	3.8	11
20	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	4.1	40
21	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.	2.4	31
22	Bone marrowâ€derived mitochondrial DNA has limited capacity for interâ€tissue transfer in vivo. FASEB Journal, 2020, 34, 9297-9306.	0.5	5
23	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.	1.2	7
24	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.	2.3	10
25	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	6.2	171
26	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.	2.5	27
27	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.	2.2	21
28	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64.	4.1	71
29	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.	6.2	125
30	Clinical Validation of Copy Number Variant Detection from Targeted Next-Generation Sequencing Panels. Journal of Molecular Diagnostics, 2017, 19, 905-920.	2.8	104
31	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.	2.8	47