

Mads Bak

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8503584/publications.pdf>

Version: 2024-02-01

65
papers

5,921
citations

201385

27
h-index

123241

61
g-index

66
all docs

66
docs citations

66
times ranked

11720
citing authors

#	ARTICLE	IF	CITATIONS
1	9p21.3 Microdeletion involving <i>CDKN2A/2B</i> in a young patient with multiple primary cancers and review of the literature. <i>Journal of Physical Education and Sports Management</i> , 2022, 8, a006164.	0.5	1
2	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. <i>PLoS Genetics</i> , 2021, 17, e1009679.	1.5	17
3	Case report: "AARS2 leukodystrophy". <i>Molecular Genetics and Metabolism Reports</i> , 2021, 28, 100782.	0.4	7
4	Paroxysmal Cranial Dyskinesia and Nail-Patella Syndrome Caused by a Novel Variant in the <i>LMX1B</i> Gene. <i>Movement Disorders</i> , 2020, 35, 2343-2347.	2.2	2
5	Mitochondrial dysfunction induced by variation in the non-coding genome " A proposed workflow to improve diagnostics. <i>Mitochondrion</i> , 2020, 53, 255-259.	1.6	1
6	Diagnostic pitfalls in vitamin B6-dependent epilepsy caused by mutations in the <i>PLPBP</i> gene. <i>JIMD Reports</i> , 2019, 50, 1-8.	0.7	16
7	Multigenic truncation of the semaphorin-plexin pathway by a germline chromothriptic rearrangement associated with Moebius syndrome. <i>Human Mutation</i> , 2019, 40, 1057-1062.	1.1	4
8	Haploinsufficiency of <i>ARHGAP42</i> is associated with hypertension. <i>European Journal of Human Genetics</i> , 2019, 27, 1296-1303.	1.4	12
9	A splice-site variant in the lncRNA gene cosegregates in the large Volkmann cataract family. <i>Molecular Vision</i> , 2019, 25, 1-11.	1.1	1
10	Very short DNA segments can be detected and handled by the repair machinery during germline chromothriptic chromosome reassembly. <i>Human Mutation</i> , 2018, 39, 709-716.	1.1	19
11	Regulatory variants of <i>FOXP1</i> in the context of its topological domain organisation. <i>European Journal of Human Genetics</i> , 2018, 26, 186-196.	1.4	20
12	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. <i>PLoS Genetics</i> , 2018, 14, e1007780.	1.5	28
13	Aggressiveness of non-EMT breast cancer cells relies on <i>FBXO11</i> activity. <i>Molecular Cancer</i> , 2018, 17, 171.	7.9	20
14	Position effect, cryptic complexity, and direct gene disruption as disease mechanisms in de novo apparently balanced translocation cases. <i>PLoS ONE</i> , 2018, 13, e0205298.	1.1	14
15	Risks and Recommendations in Prenatally Detected De Novo Balanced Chromosomal Rearrangements from Assessment of Long-Term Outcomes. <i>American Journal of Human Genetics</i> , 2018, 102, 1090-1103.	2.6	29
16	Cryptic breakpoint identified by whole-genome mate-pair sequencing in a rare paternally inherited complex chromosomal rearrangement. <i>Molecular Cytogenetics</i> , 2018, 11, 34.	0.4	11
17	Enrichment of megabase-sized DNA molecules for single-molecule optical mapping and next-generation sequencing. <i>Scientific Reports</i> , 2017, 7, 17893.	1.6	5
18	Elevation of brain-enriched miRNAs in cerebrospinal fluid of patients with acute ischemic stroke. <i>Biomarker Research</i> , 2017, 5, 24.	2.8	59

#	ARTICLE	IF	CITATIONS
19	Accurate Breakpoint Mapping in Apparently Balanced Translocation Families with Discordant Phenotypes Using Whole Genome Mate-Pair Sequencing. PLoS ONE, 2017, 12, e0169935.	1.1	31
20	Germline Chromothripsis Driven by L1-Mediated Retrotransposition and Alu/Alu Homologous Recombination. Human Mutation, 2016, 37, 385-395.	1.1	50
21	Genome-wide DNA methylation analysis of transient neonatal diabetes type 1 patients with mutations in ZFP57. BMC Medical Genetics, 2016, 17, 29.	2.1	27
22	Re-Examination of Danish Carriers of Balanced Chromosomal Inversions. Cancer Genetics, 2016, 209, 231.	0.2	0
23	Systematic Mapping of Chromosomal Breakpoints in the Context of Phenotypes and Nuclear Genome Organization. Cancer Genetics, 2016, 209, 301.	0.2	0
24	Phenotypic subregions within the split-hand/foot malformation 1 locus. Human Genetics, 2016, 135, 345-357.	1.8	15
25	Identification of the BRD1 interaction network and its impact on mental disorder risk. Genome Medicine, 2016, 8, 53.	3.6	29
26	The segregation of different submicroscopic imbalances underlying the clinical variability associated with a familial karyotypically balanced translocation. Molecular Cytogenetics, 2015, 8, 106.	0.4	6
27	Dysregulation of FOXC1 by ring chromosome 14. Molecular Cytogenetics, 2015, 8, 24.	0.4	8
28	The small RNA content of human sperm reveals pseudogene-derived piRNAs complementary to protein-coding genes. Rna, 2015, 21, 1085-1095.	1.6	83
29	Partial USH2A deletions contribute to Usher syndrome in Denmark. European Journal of Human Genetics, 2015, 23, 1646-1651.	1.4	8
30	International Breakpoint Mapping Consortium (IBMC). Systematic Mapping of Chromosomal Breakpoints in the Context of Phenotypes and Nuclear Genome Organization. Cancer Genetics, 2015, 208, 359-360.	0.2	0
31	Immature truncated O-glycophenotype of cancer directly induces oncogenic features. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E4066-75.	3.3	251
32	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	1.4	19
33	The strength of combined cytogenetic and mate-pair sequencing techniques illustrated by a germline chromothripsis rearrangement involving FOXP2. European Journal of Human Genetics, 2014, 22, 338-343.	1.4	46
34	Aberrant expression of miR-218 and miR-204 in human mesial temporal lobe epilepsy and hippocampal sclerosis—Convergence on axonal guidance. Epilepsia, 2014, 55, 2017-2027.	2.6	71
35	Neurodevelopmental disorders associated with dosage imbalance of <i>ZBTB20</i> correlate with the morbidity spectrum of ZBTB20 candidate target genes. Journal of Medical Genetics, 2014, 51, 605-613.	1.5	26
36	Hypomorphic Mutations in PGAP2, Encoding a GPI-Anchor-Remodeling Protein, Cause Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2013, 92, 575-583.	2.6	87

#	ARTICLE	IF	CITATIONS
37	Next-generation sequencing: proof of concept for antenatal prediction of the fetal Kell blood group phenotype from cell-free fetal DNA in maternal plasma. <i>Transfusion</i> , 2013, 53, 2892-2898.	0.8	51
38	REST-Mediated Recruitment of Polycomb Repressor Complexes in Mammalian Cells. <i>PLoS Genetics</i> , 2012, 8, e1002494.	1.5	140
39	Haploinsufficiency of CELF4 at 18q12.2 is associated with developmental and behavioral disorders, seizures, eye manifestations, and obesity. <i>European Journal of Human Genetics</i> , 2012, 20, 1315-1319.	1.4	34
40	Sequence and expression analysis of gaps in human chromosome 20. <i>Nucleic Acids Research</i> , 2012, 40, 6660-6672.	6.5	5
41	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of <i>ARID1B</i> . <i>Clinical Genetics</i> , 2012, 82, 248-255.	1.0	126
42	Aberrant brain microRNA target and miRISC gene expression in the anx/anx anorexia mouse model. <i>Gene</i> , 2012, 497, 181-190.	1.0	12
43	Profiling microRNAs in lung tissue from pigs infected with <i>Actinobacillus pleuropneumoniae</i> . <i>BMC Genomics</i> , 2012, 13, 459.	1.2	54
44	Validation of Genome-Wide Intervertebral Disk Calcification Associations in Dachshund and Further Investigation of the Chromosome 12 Susceptibility Locus. <i>Frontiers in Genetics</i> , 2012, 3, 225.	1.1	18
45	YKL-40 Is Differentially Expressed in Human Embryonic Stem Cells and in Cell Progeny of the Three Germ Layers. <i>Journal of Histochemistry and Cytochemistry</i> , 2012, 60, 188-204.	1.3	36
46	Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. <i>European Journal of Medical Genetics</i> , 2011, 54, e383-e388.	0.7	7
47	Cost-effective multiplexing before capture allows screening of 25,000 clinically relevant SNPs in childhood acute lymphoblastic leukemia. <i>Leukemia</i> , 2011, 25, 1001-1006.	3.3	20
48	MicroRNA-138 regulates osteogenic differentiation of human stromal (mesenchymal) stem cells in vivo. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, 6139-6144.	3.3	443
49	Genome-wide detection of chromosomal rearrangements, indels, and mutations in circular chromosomes by short read sequencing. <i>Genome Research</i> , 2011, 21, 1388-1393.	2.4	79
50	Jarid1b targets genes regulating development and is involved in neural differentiation. <i>EMBO Journal</i> , 2011, 30, 4586-4600.	3.5	183
51	JARID2 regulates binding of the Polycomb repressive complex 2 to target genes in ES cells. <i>Nature</i> , 2010, 464, 306-310.	13.7	499
52	Ancient human genome sequence of an extinct Palaeo-Eskimo. <i>Nature</i> , 2010, 463, 757-762.	13.7	750
53	Genome-wide Analysis of CDX2 Binding in Intestinal Epithelial Cells (Caco-2). <i>Journal of Biological Chemistry</i> , 2010, 285, 25115-25125.	1.6	68
54	A Functional Link between the Histone Demethylase PHF8 and the Transcription Factor ZNF711 in X-Linked Mental Retardation. <i>Molecular Cell</i> , 2010, 38, 165-178.	4.5	186

#	ARTICLE	IF	CITATIONS
55	MicroRNA-31 functions as an oncogenic microRNA in mouse and human lung cancer cells by repressing specific tumor suppressors. <i>Journal of Clinical Investigation</i> , 2010, 120, 1298-1309.	3.9	353
56	Uncovering Growth-Suppressive MicroRNAs in Lung Cancer. <i>Clinical Cancer Research</i> , 2009, 15, 1177-1183.	3.2	167
57	GLI1 Is Involved in Cell Cycle Regulation and Proliferation of NT2 Embryonal Carcinoma Stem Cells. <i>DNA and Cell Biology</i> , 2008, 27, 251-256.	0.9	11
58	MicroRNA expression in the adult mouse central nervous system. <i>Rna</i> , 2008, 14, 432-444.	1.6	427
59	Antagonism of microRNA-122 in mice by systemically administered LNA-antimiR leads to up-regulation of a large set of predicted target mRNAs in the liver. <i>Nucleic Acids Research</i> , 2008, 36, 1153-1162.	6.5	630
60	Altered MicroRNA Expression Confined to Specific Epithelial Cell Subpopulations in Breast Cancer. <i>Cancer Research</i> , 2007, 67, 11612-11620.	0.4	515
61	Non-disjunction of chromosome 13. <i>Human Molecular Genetics</i> , 2007, 16, 2004-2010.	1.4	38
62	Evaluation of two methods for generating cRNA for microarray experiments from nanogram amounts of total RNA. <i>Analytical Biochemistry</i> , 2006, 358, 111-119.	1.1	5
63	Trisomy 13 due to <i>rea(13q;13q)</i> is caused by <i>i(13)</i> and not <i>rob(13;13)(q10;q10)</i> in the majority of cases. , 2005, 132A, 310-313.		9
64	Mutation analysis of the Sonic hedgehog promoter and putative enhancer elements in Parkinson's disease patients. <i>Molecular Brain Research</i> , 2004, 126, 207-211.	2.5	4
65	The Hedgehog signaling pathway â€™ implications for drug targets in cancer and neurodegenerative disorders. <i>Pharmacogenomics</i> , 2003, 4, 411-429.	0.6	26