Andreas Rump

List of Publications by Year
in descending order

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Version: 2024-02-01

69 papers	27,125 citations	186265 28 h-index	91884 69 g-index
71 all docs	71 docs citations	71 times ranked	32260 citing authors

#	Article	IF	Citations
1	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	6.3	19
2	Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	1.6	90
3	Prevalence of Cancer Predisposition Germline Variants in Male Breast Cancer Patients: Results of the German Consortium for Hereditary Breast and Ovarian Cancer. Cancers, 2022, 14, 3292.	3.7	11
4	Case Report: ANXA2 Associated Life-Threatening Coagulopathy With Hyperfibrinolysis in a Patient With Non-APL Acute Myeloid Leukemia. Frontiers in Oncology, 2021, 11, 666014.	2.8	2
5	Comprehensive Genomic and Transcriptomic Analysis for Guiding Therapeutic Decisions in Patients with Rare Cancers. Cancer Discovery, 2021, 11, 2780-2795.	9.4	125
6	Performance of Breast Cancer Polygenic Risk Scores in 760 FemaleCHEK2Germline Mutation Carriers. Journal of the National Cancer Institute, 2021, 113, 893-899.	6.3	21
7	Genetic and genomic studies of pathogenic EXOSC2 mutations in the newly described disease SHRF implicate the autophagy pathway in disease pathogenesis. Human Molecular Genetics, 2020, 29, 541-553.	2.9	21
8	Novel dominant-negative NR2F1 frameshift mutation and a phenotypic expansion of the Bosch-Boonstra-Schaaf optic atrophy syndrome. European Journal of Medical Genetics, 2020, 63, 104019.	1.3	9
9	Criteria of the German Consortium for Hereditary Breast and Ovarian Cancer for the Classification of Germline Sequence Variants in Risk Genes for Hereditary Breast and Ovarian Cancer. Geburtshilfe Und Frauenheilkunde, 2020, 80, 410-429.	1.8	18
10	Novel truncating PPM1D mutation in a patient with intellectual disability. European Journal of Medical Genetics, 2019, 62, 70-72.	1.3	10
11	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	2.5	102
12	Optimizing Genetic Workup in Pheochromocytoma and Paraganglioma by Integrating Diagnostic and Research Approaches. Cancers, 2019, 11, 809.	3.7	23
13	Targeted capture-based NGS is superior to multiplex PCR-based NGS for hereditary BRCA1 and BRCA2 gene analysis in FFPE tumor samples. BMC Cancer, 2019, 19, 396.	2.6	30
14	Gene panel testing of 5589 <scp>BRCA</scp>1/2â€negative index patients with breast cancer in a routine diagnostic setting: results of the German Consortium for Hereditary Breast and Ovarian Cancer. Cancer Medicine, 2018, 7, 1349-1358.	2.8	126
15	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	2.4	92
16	A frameshift mutation in BRCA1 leads to hereditary breast and ovarian cancer in one part of a family and to familial pancreatic cancer in another. Breast Cancer Research and Treatment, 2018, 167, 305-307.	2.5	1
17	Next-generation panel sequencing identifies NF1 germline mutations in three patients with pheochromocytoma but no clinical diagnosis of neurofibromatosis type 1. European Journal of Endocrinology, 2018, 178, K1-K9.	3.7	19
18	Variants in exons 5 and 6 of ACTB cause syndromic thrombocytopenia. Nature Communications, 2018, 9, 4250.	12.8	38

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19	Diagnostic value of partial exome sequencing in developmental disorders. PLoS ONE, 2018, 13, e0201041.	2.5	36
20	Functional monosomy of 6q27â€qter and functional disomy of Xpterâ€p22.11 due to X;6 translocation with an atypical Xâ€inactivation pattern. American Journal of Medical Genetics, Part A, 2017, 173, 1334-1341.	1.2	5
21	New gain-of-function mutation shows CACNA1D as recurrently mutated gene in autism spectrum disorders and epilepsy. Human Molecular Genetics, 2017, 26, 2923-2932.	2.9	85
22	The contribution of homology arms to nuclease-assisted genome engineering. Nucleic Acids Research, 2017, 45, 8105-8115.	14.5	23
23	Pierpont syndrome: report of a new patient. Clinical Dysmorphology, 2017, 26, 205-208.	0.3	12
24	Spectrum of genetic variants of BRCA1 and BRCA2 in a German single center study. Archives of Gynecology and Obstetrics, 2017, 295, 1227-1238.	1.7	18
25	BRCA1/2 missense mutations and the value of in-silico analyses. European Journal of Medical Genetics, 2017, 60, 572-577.	1.3	7
26	Comprehensive molecular characterization of multifocal glioblastoma proves its monoclonal origin and reveals novel insights into clonal evolution and heterogeneity of glioblastomas. Neuro-Oncology, 2017, 19, 546-557.	1.2	86
27	Identification and Functional Testing of ERCC2 Mutations in a Multi-national Cohort of Patients with Familial Breast- and Ovarian Cancer. PLoS Genetics, 2016, 12, e1006248.	3.5	22
28	Novel ADAMTSL2-mutations in a patient with geleophysic dysplasia type I. Clinical Dysmorphology, 2016, 25, 106-109.	0.3	5
29	Update on the ACTG1â€associated Baraitserâ€“Winter cerebrofrontofacial syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2644-2651.	1.2	28
30	Identification and Characterization of a Novel ConstitutionalPIK3CAMutation in a Child Lacking the Typical Segmental Overgrowth of â€œPIK3CA-Related Overgrowth Spectrumâ€. Human Mutation, 2016, 37, 242-245.	2.5	11
31	Ready to clone: CNV detection and breakpoint fine-mapping in breast and ovarian cancer susceptibility genes by high-resolution array CGH. Breast Cancer Research and Treatment, 2016, 159, 585-590.	2.5	15
32	Tentative clinical diagnosis of Lujanâ€Fryns syndromeâ€”A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	1.2	11
33	An unusual case of Cowden syndrome associated with ganglioneuromatous polyposis. Hereditary Cancer in Clinical Practice, 2016, 14, 11.	1.5	6
34	Mutations inEXOSC2are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. Journal of Medical Genetics, 2016, 53, 419-425.	3.2	69
35	A child with Liâ€“Fraumeni syndrome: Modes to inactivate the second allele of TP53 in three different malignancies. Pediatric Blood and Cancer, 2015, 62, 1481-1484.	1.5	22
36	HBOC multi-gene panel testing: comparison of two sequencing centers. Breast Cancer Research and Treatment, 2015, 152, 129-136.	2.5	38

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37	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
38	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	2.8	115
39	Severe forms of Baraitserâ€“Winter syndrome are caused by ACTB mutations rather than ACTG1 mutations. European Journal of Human Genetics, 2014, 22, 179-183.	2.8	67
40	Severe intellectual disability, West syndrome, Dandyâ€“Walker malformation, and syndactyly in a patient with partial tetrasomy 17q25.3. American Journal of Medical Genetics, Part A, 2013, 161, 3144-3149.	1.2	13
41	Partial deletion of GLRB and GRIA2 in a patient with intellectual disability. European Journal of Human Genetics, 2013, 21, 112-114.	2.8	22
42	A mosaic maternal splice donor mutation in the EHMT1 gene leads to aberrant transcripts and to Kleefstra syndrome in the offspring. European Journal of Human Genetics, 2013, 21, 887-890.	2.8	14
43	Novel CIC Point Mutations and an Exon-Spanning, Homozygous Deletion Identified in Oligodendroglial Tumors by a Comprehensive Genomic Approach Including Transcriptome Sequencing. PLoS ONE, 2013, 8, e76623.	2.5	16
44	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	13.7	940
45	A misplaced lncRNA causes brachydactyly in humans. Journal of Clinical Investigation, 2012, 122, 3990-4002.	8.2	108
46	A Homozygous Microdeletion withinADAMTSL4in Patients with Isolated Ectopia Lentis: Evidence of a Founder Mutation. , 2011, 52, 695.		21
47	A cis-regulatory site downregulates PTHLH in translocation t(8;12)(q13;p11.2) and leads to Brachydactyly Type E. Human Molecular Genetics, 2010, 19, 848-860.	2.9	67
48	Characterization of a new Xâ€linked mental retardation syndrome with microcephaly, cortical malformation, and thin habitus. American Journal of Medical Genetics, Part A, 2009, 149A, 2469-2478.	1.2	27
49	Identification and characterization of CaApe2 Ã¢Â€Â“ a neutral arginine/alanine/leucine-specific metallo-aminopeptidase fromCandida albicans. FEMS Yeast Research, 2008, 8, 858-869.	2.3	14
50	A splice-supporting intronic mutation in the last bp position of a cryptic exon within intron 6 of the CYBB gene induces its incorporation into the mRNA causing chronic granulomatous disease (CGD). Gene, 2006, 371, 174-181.	2.2	22
51	In acute leukemia, the polymorphism â^’211C>T in the promoter region of the multidrug resistance-associated protein 3 (MRP3) does not determine the expression level of the gene. Pharmacogenetics and Genomics, 2006, 16, 149-150.	1.5	20
52	Identification of a Set of Seven Genes for the Monitoring of Minimal Residual Disease in Pediatric Acute Myeloid Leukemia. Clinical Cancer Research, 2006, 12, 2434-2441.	7.0	111
53	Expression levels of the putative zinc transporter LIV-1 are associated with a better outcome of breast cancer patients. International Journal of Cancer, 2005, 117, 961-973.	5.1	75
54	Small Reciprocal Insertion detected by Spectral Karyotyping (SKY) and delimited by Array-CGH Analysis. European Journal of Medical Genetics, 2005, 48, 328-338.	1.3	9

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55	High-resolution analysis of chromosomal imbalances using the Affymetrix 10K SNP genotyping chip. Genomics, 2005, 85, 392-400.	2.9	26
56	Expression of mouse Tbx22 supports its role in palatogenesis and glossogenesis. Developmental Dynamics, 2003, 226, 579-586.	1.8	31
57	Defects in whirlin, a PDZ domain molecule involved in stereocilia elongation, cause deafness in the whirler mouse and families with DFNB31. Nature Genetics, 2003, 34, 421-428.	21.4	293
58	Different structural organization of the encephalopsin gene in man and mouse. Gene, 2002, 295, 27-32.	2.2	20
59	Gene Structure and Regulation of the Murine Epithelial Calcium Channels ECaC1 and 2. Biochemical and Biophysical Research Communications, 2001, 289, 1287-1294.	2.1	118
60	Complex Arrangement of Genes within a 220-kb Region of Double-Duplicated DNA on Human 2q37.1. Genomics, 2001, 73, 50-55.	2.9	6
61	A High-Resolution Genetic, Physical, and Comparative Gene Map of the Doublefoot (Dbf) Region of Mouse Chromosome 1 and the Region of Conserved Synteny on Human Chromosome 2q35. Genomics, 2001, 78, 197-205.	2.9	8
62	Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921.	27.8	21,074
63	The DNA sequence of human chromosome 21. Nature, 2000, 405, 311-319.	27.8	1,144
64	RUMMAGE â€“ a high-throughput sequence annotation system. Trends in Genetics, 2000, 16, 519-521.	6.7	25
65	Comparative Genome Sequence Analysis of the Bpa/Str Region in Mouse and Man. Genome Research, 2000, 10, 758-775.	5.5	48
66	Pseudoautosomal deletions encompassing a novel homeobox gene cause growth failure in idiopathic short stature and Turner syndrome. Nature Genetics, 1997, 16, 54-63.	21.4	867
67	Editing of GluR2 RNA in the Gerbil Hippocampus after Global Cerebral Ischemia. Journal of Cerebral Blood Flow and Metabolism, 1996, 16, 1362-1365.	4.3	22
68	Tandem arrangement of tRNAAsp-encoding genes in Phytophthora spp. Gene, 1991, 102, 51-56.	2.2	4
69	Nucleotide sequence of a 24,206-base-pair DNA fragment carrying the entire nitrogen fixation gene cluster of Klebsiella pneumoniae. Journal of Molecular Biology, 1988, 203, 715-738.	4.2	290