

# Andreas Rump

## List of Publications by Year in descending order

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Version: 2024-02-01

69  
papers

27,125  
citations

185998

28  
h-index

91712

69  
g-index

71  
all docs

71  
docs citations

71  
times ranked

32260  
citing authors

#	ARTICLE	IF	CITATIONS
1	Initial sequencing and analysis of the human genome. <i>Nature</i> , 2001, 409, 860-921.	13.7	21,074
2	The DNA sequence of human chromosome 21. <i>Nature</i> , 2000, 405, 311-319.	13.7	1,144
3	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. <i>Lancet</i> , The, 2012, 380, 1674-1682.	6.3	940
4	Pseudoautosomal deletions encompassing a novel homeobox gene cause growth failure in idiopathic short stature and Turner syndrome. <i>Nature Genetics</i> , 1997, 16, 54-63.	9.4	867
5	Defects in whirlin, a PDZ domain molecule involved in stereocilia elongation, cause deafness in the whirler mouse and families with DFNB31. <i>Nature Genetics</i> , 2003, 34, 421-428.	9.4	293
6	Nucleotide sequence of a 24,206-base-pair DNA fragment carrying the entire nitrogen fixation gene cluster of <i>Klebsiella pneumoniae</i> . <i>Journal of Molecular Biology</i> , 1988, 203, 715-738.	2.0	290
7	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	2.6	230
8	Gene panel testing of 5589 <i>BRCA1/2</i> negative index patients with breast cancer in a routine diagnostic setting: results of the German Consortium for Hereditary Breast and Ovarian Cancer. <i>Cancer Medicine</i> , 2018, 7, 1349-1358.	1.3	126
9	Comprehensive Genomic and Transcriptomic Analysis for Guiding Therapeutic Decisions in Patients with Rare Cancers. <i>Cancer Discovery</i> , 2021, 11, 2780-2795.	7.7	125
10	Gene Structure and Regulation of the Murine Epithelial Calcium Channels ECaC1 and 2. <i>Biochemical and Biophysical Research Communications</i> , 2001, 289, 1287-1294.	1.0	118
11	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. <i>European Journal of Human Genetics</i> , 2015, 23, 292-301.	1.4	115
12	Identification of a Set of Seven Genes for the Monitoring of Minimal Residual Disease in Pediatric Acute Myeloid Leukemia. <i>Clinical Cancer Research</i> , 2006, 12, 2434-2441.	3.2	111
13	A misplaced lncRNA causes brachydactyly in humans. <i>Journal of Clinical Investigation</i> , 2012, 122, 3990-4002.	3.9	108
14	Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019, 40, 1557-1578.	1.1	102
15	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. <i>Genetics in Medicine</i> , 2018, 20, 1354-1364.	1.1	92
16	Cancer Risks Associated With <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>Journal of Clinical Oncology</i> , 2022, 40, 1529-1541.	0.8	90
17	Comprehensive molecular characterization of multifocal glioblastoma proves its monoclonal origin and reveals novel insights into clonal evolution and heterogeneity of glioblastomas. <i>Neuro-Oncology</i> , 2017, 19, 546-557.	0.6	86
18	New gain-of-function mutation shows CACNA1D as recurrently mutated gene in autism spectrum disorders and epilepsy. <i>Human Molecular Genetics</i> , 2017, 26, 2923-2932.	1.4	85

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19	Expression levels of the putative zinc transporter LIV-1 are associated with a better outcome of breast cancer patients. <i>International Journal of Cancer</i> , 2005, 117, 961-973.	2.3	75
20	Mutations in <i>EXOSC2</i> are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. <i>Journal of Medical Genetics</i> , 2016, 53, 419-425.	1.5	69
21	A cis-regulatory site downregulates PTHLH in translocation t(8;12)(q13;p11.2) and leads to Brachydactyly Type E. <i>Human Molecular Genetics</i> , 2010, 19, 848-860.	1.4	67
22	Severe forms of Baraitser's Winter syndrome are caused by ACTB mutations rather than ACTG1 mutations. <i>European Journal of Human Genetics</i> , 2014, 22, 179-183.	1.4	67
23	Comparative Genome Sequence Analysis of the Bpa/Str Region in Mouse and Man. <i>Genome Research</i> , 2000, 10, 758-775.	2.4	48
24	HBOC multi-gene panel testing: comparison of two sequencing centers. <i>Breast Cancer Research and Treatment</i> , 2015, 152, 129-136.	1.1	38
25	Variants in exons 5 and 6 of ACTB cause syndromic thrombocytopenia. <i>Nature Communications</i> , 2018, 9, 4250.	5.8	38
26	Diagnostic value of partial exome sequencing in developmental disorders. <i>PLoS ONE</i> , 2018, 13, e0201041.	1.1	36
27	Expression of mouse Tbx22 supports its role in palatogenesis and glossogenesis. <i>Developmental Dynamics</i> , 2003, 226, 579-586.	0.8	31
28	Targeted capture-based NGS is superior to multiplex PCR-based NGS for hereditary BRCA1 and BRCA2 gene analysis in FFPE tumor samples. <i>BMC Cancer</i> , 2019, 19, 396.	1.1	30
29	Update on the <i>ACTG1</i> -associated Baraitser's Winter cerebrofrontofacial syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2644-2651.	0.7	28
30	Characterization of a new X-linked mental retardation syndrome with microcephaly, cortical malformation, and thin habitus. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2469-2478.	0.7	27
31	High-resolution analysis of chromosomal imbalances using the Affymetrix 10K SNP genotyping chip. <i>Genomics</i> , 2005, 85, 392-400.	1.3	26
32	RUMMAGE - a high-throughput sequence annotation system. <i>Trends in Genetics</i> , 2000, 16, 519-521.	2.9	25
33	The contribution of homology arms to nuclease-assisted genome engineering. <i>Nucleic Acids Research</i> , 2017, 45, 8105-8115.	6.5	23
34	Optimizing Genetic Workup in Pheochromocytoma and Paraganglioma by Integrating Diagnostic and Research Approaches. <i>Cancers</i> , 2019, 11, 809.	1.7	23
35	Editing of GluR2 RNA in the Gerbil Hippocampus after Global Cerebral Ischemia. <i>Journal of Cerebral Blood Flow and Metabolism</i> , 1996, 16, 1362-1365.	2.4	22
36	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). <i>Gene</i> , 2006, 371, 174-181.	1.0	22

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37	Partial deletion of GLRB and GRIA2 in a patient with intellectual disability. <i>European Journal of Human Genetics</i> , 2013, 21, 112-114.	1.4	22
38	A child with Li-Fraumeni syndrome: Modes to inactivate the second allele of <i>TP53</i> in three different malignancies. <i>Pediatric Blood and Cancer</i> , 2015, 62, 1481-1484.	0.8	22
39	Identification and Functional Testing of ERCC2 Mutations in a Multi-national Cohort of Patients with Familial Breast- and Ovarian Cancer. <i>PLoS Genetics</i> , 2016, 12, e1006248.	1.5	22
40	A Homozygous Microdeletion within <i>ADAMTSL4</i> in Patients with Isolated Ectopia Lentis: Evidence of a Founder Mutation. , 2011, 52, 695.		21
41	Genetic and genomic studies of pathogenic EXOSC2 mutations in the newly described disease SHRF implicate the autophagy pathway in disease pathogenesis. <i>Human Molecular Genetics</i> , 2020, 29, 541-553.	1.4	21
42	Performance of Breast Cancer Polygenic Risk Scores in 760 Female <i>CHEK2</i> Germline Mutation Carriers. <i>Journal of the National Cancer Institute</i> , 2021, 113, 893-899.	3.0	21
43	Different structural organization of the encephalopsin gene in man and mouse. <i>Gene</i> , 2002, 295, 27-32.	1.0	20
44	In acute leukemia, the polymorphism $\sim 211C>T$ in the promoter region of the multidrug resistance-associated protein 3 (MRP3) does not determine the expression level of the gene. <i>Pharmacogenetics and Genomics</i> , 2006, 16, 149-150.	0.7	20
45	Next-generation panel sequencing identifies NF1 germline mutations in three patients with pheochromocytoma but no clinical diagnosis of neurofibromatosis type 1. <i>European Journal of Endocrinology</i> , 2018, 178, K1-K9.	1.9	19
46	Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2022, 114, 109-122.	3.0	19
47	Spectrum of genetic variants of BRCA1 and BRCA2 in a German single center study. <i>Archives of Gynecology and Obstetrics</i> , 2017, 295, 1227-1238.	0.8	18
48	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. <i>Geburtshilfe Und Frauenheilkunde</i> , 2020, 80, 410-429.	0.8	18
49	Novel CIC Point Mutations and an Exon-Spanning, Homozygous Deletion Identified in Oligodendroglial Tumors by a Comprehensive Genomic Approach Including Transcriptome Sequencing. <i>PLoS ONE</i> , 2013, 8, e76623.	1.1	16
50	Ready to clone: CNV detection and breakpoint fine-mapping in breast and ovarian cancer susceptibility genes by high-resolution array CGH. <i>Breast Cancer Research and Treatment</i> , 2016, 159, 585-590.	1.1	15
51	Identification and characterization of CaApe2 " a neutral arginine/alanine/leucine-specific metallo-aminopeptidase from <i>Candida albicans</i> . <i>FEMS Yeast Research</i> , 2008, 8, 858-869.	1.1	14
52	A mosaic maternal splice donor mutation in the EHMT1 gene leads to aberrant transcripts and to Kleefstra syndrome in the offspring. <i>European Journal of Human Genetics</i> , 2013, 21, 887-890.	1.4	14
53	Severe intellectual disability, West syndrome, Dandy-Walker malformation, and syndactyly in a patient with partial tetrasomy 17q25.3. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 3144-3149.	0.7	13
54	Pierpont syndrome: report of a new patient. <i>Clinical Dysmorphology</i> , 2017, 26, 205-208.	0.1	12

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55	Identification and Characterization of a Novel Constitutional PIK3CA Mutation in a Child Lacking the Typical Segmental Overgrowth of PIK3CA-Related Overgrowth Spectrum Human Mutation, 2016, 37, 242-245.	1.1	11
56	Tentative clinical diagnosis of Lujan-Fryns syndrome "A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	0.7	11
57	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.	1.7	11
58	Novel truncating PPM1D mutation in a patient with intellectual disability. European Journal of Medical Genetics, 2019, 62, 70-72.	0.7	10
59	Small Reciprocal Insertion detected by Spectral Karyotyping (SKY) and delimited by Array-CGH Analysis. European Journal of Medical Genetics, 2005, 48, 328-338.	0.7	9
60	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.	0.7	9
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.	1.3	8
62	BRCA1/2 missense mutations and the value of in-silico analyses. European Journal of Medical Genetics, 2017, 60, 572-577.	0.7	7
63	Complex Arrangement of Genes within a 220-kb Region of Double-Duplicated DNA on Human 2q37.1. Genomics, 2001, 73, 50-55.	1.3	6
64	An unusual case of Cowden syndrome associated with ganglioneuromatous polyposis. Hereditary Cancer in Clinical Practice, 2016, 14, 11.	0.6	6
65	Novel ADAMTSL2-mutations in a patient with geleophysic dysplasia type I. Clinical Dysmorphology, 2016, 25, 106-109.	0.1	5
66	Functional monosomy of 6q27qter and functional disomy of Xpterp22.11 due to X;6 translocation with an atypical X inactivation pattern. American Journal of Medical Genetics, Part A, 2017, 173, 1334-1341.	0.7	5
67	Tandem arrangement of tRNA <sup>Asp</sup> -encoding genes in <i>Phytophthora</i> spp. Gene, 1991, 102, 51-56.	1.0	4
68	Case Report: ANXA2 Associated Life-Threatening Coagulopathy With Hyperfibrinolysis in a Patient With Non-APL Acute Myeloid Leukemia. Frontiers in Oncology, 2021, 11, 666014.	1.3	2
69	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.	1.1	1