Jan P Dumanski

List of Publications by Year
in descending order

99 papers	7,632 citations	69737 41 h-index	57558 83 g-index
117 all docs	117 docs citations	117 times ranked	9215 citing authors

#	Article	IF	Citations
1	Mosaic Loss of Chromosome Y Is Associated With Functional Outcome After Ischemic Stroke. Stroke, 2023, 54, 2434-2437.	5.3	3
2	Loss of Y and clonal hematopoiesis in bloodâ€"two sides of the same coin?. Leukemia, 2022, 36, 889-891.	7.5	27
3	Comprehensive cancer-oriented biobanking resource of human samples for studies of post-zygotic genetic variation involved in cancer predisposition. PLoS ONE, 2022, 17, e0266111.	2.5	6
4	High prevalence of somatic PIK3CA and TP53 pathogenic variants in the normal mammary gland tissue of sporadic breast cancer patients revealed by duplex sequencing. Npj Breast Cancer, 2022, 8, .	5.4	8
5	Loss of Y in leukocytes as a risk factor for critical COVID-19 in men. Genome Medicine, 2022, 14, .	8.5	9
6	Immune cells lacking Y chromosome show dysregulation of autosomal gene expression. Cellular and Molecular Life Sciences, 2021, 78, 4019-4033.	5.5	62
7	Leukocytes with chromosome Y loss have reduced abundance of the cell surface immunoprotein CD99. Scientific Reports, 2021, 11, 15160.	3.4	29
8	Variable degree of mosaicism for tetrasomy 18p in phenotypically discordant monozygotic twinsâ€"Diagnostic implications. Molecular Genetics & Genomic Medicine, 2021, 9, e1526.	1.3	8
9	Longitudinal changes in the frequency of mosaic chromosome Y loss in peripheral blood cells of aging men varies profoundly between individuals. European Journal of Human Genetics, 2020, 28, 349-357.	2.9	53
10	Genetic predisposition to mosaic Y chromosome loss in blood. Nature, 2019, 575, 652-657.	36.2	217
11	PRR14L mutations are associated with chromosome 22 acquired uniparental disomy, age-related clonal hematopoiesis and myeloid neoplasia. Leukemia, 2019, 33, 1184-1194.	7.5	11
12	Mosaic loss of chromosome Y in leukocytes matters. Nature Genetics, 2019, 51, 4-7.	20.4	53
13	Mosaicism in health and disease â€" clones picking up speed. Nature Reviews Genetics, 2017, 18, 128-142.	16.7	214
14	Loss of Chromosome Y in Leukocytes and Major Cardiovascular Events. Circulation: Cardiovascular Genetics, 2017, 10, e001820.	5.1	5
15	Mosaic Loss of Chromosome Y in Blood Is Associated with Alzheimer Disease. American Journal of Human Genetics, 2016, 98, 1208-1219.	6.1	175
16	Concurrent DNA Copy-Number Alterations and Mutations in Genes Related to Maintenance of Genome Stability in Uninvolved Mammary Glandular Tissue from Breast Cancer Patients. Human Mutation, 2015, 36, 1088-1099.	2.8	12
17	Signatures of post-zygotic structural genetic aberrations in the cells of histologically normal breast tissue that can predispose to sporadic breast cancer. Genome Research, 2015, 25, 1521-1535.	5.6	28
18	Mosaic loss of chromosome Y in peripheral blood is associated with shorter survival and higher risk of cancer. Nature Genetics, 2014, 46, 624-628.	20.4	338

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19	Post-Zygotic and Inter-Individual Structural Genetic Variation in a Presumptive Enhancer Element of the Locus between the IL10RÎ² and IFNAR1 Genes. PLoS ONE, 2013, 8, e67752.	2.5	2
20	Procoagulant activity in patients with sickle cell trait. Blood Coagulation and Fibrinolysis, 2012, 23, 268-270.	1.1	9
21	Structural Genetic Variation in the Context of Somatic Mosaicism. Methods in Molecular Biology, 2012, 838, 249-272.	0.0	34
22	Age-Related Somatic Structural Changes in the Nuclear Genome of Human Blood Cells. American Journal of Human Genetics, 2012, 90, 217-228.	6.1	176
23	Common pathogenetic mechanism involving human chromosome 18 in familial and sporadic ileal carcinoid tumors. Genes Chromosomes and Cancer, 2011, 50, 82-94.	3.3	81
24	Focal amplifications are associated with high grade and recurrences in stage Ta bladder carcinoma. International Journal of Cancer, 2010, 126, 1390-1402.	5.4	59
25	Somatic mosaicism for chromosome X and Y aneuploidies in monozygotic twins heterozygous for sickle cell disease mutation. American Journal of Medical Genetics, Part A, 2010, 152A, 2595-2598.	1.5	19
26	Frequent genetic differences between matched primary and metastatic breast cancer provide an approach to identification of biomarkers for disease progression. European Journal of Human Genetics, 2010, 18, 560-568.	2.9	42
27	Integrative epigenomic and genomic analysis of malignant pheochromocytoma. Experimental and Molecular Medicine, 2010, 42, 484.	7.8	32
28	Genome-wide microarray-based comparative genomic hybridization analysis of lymphoplasmacytic lymphomas reveals heterogeneous aberrations. Leukemia and Lymphoma, 2009, 50, 1528-1534.	1.4	8
29	Characterization of novel and complex genomic aberrations in glioblastoma using a 32K BAC array. Neuro-Oncology, 2009, 11, 803-818.	1.2	43
30	Genomeâ€wide highâ€resolution analysis of DNA copy number alterations in NF1â€associated malignant peripheral nerve sheath tumors using 32K BAC array. Genes Chromosomes and Cancer, 2009, 48, 897-907.	3.3	50
31	Tissue-specific variation in DNA methylation levels along human chromosome 1. Epigenetics and Chromatin, 2009, 2, 7.	3.9	55
32	Profiling of copy number variations (CNVs) in healthy individuals from three ethnic groups using a human genome 32â€‰K BAC-clone-based array. Human Mutation, 2008, 29, 398-408.	2.8	47
33	Somatic mosaicism for copy number variation in differentiated human tissues. Human Mutation, 2008, 29, 1118-1124.	2.8	186
34	Distal 22q11.2 microduplication encompassing the BCR gene. American Journal of Medical Genetics, Part A, 2008, 146A, 3075-3081.	1.5	30
35	Phenotypically Concordant and Discordant Monozygotic Twins Display Different DNA Copy-Number-Variation Profiles. American Journal of Human Genetics, 2008, 82, 763-771.	6.1	535
36	Autoantibodies to glutathione S-transferase theta 1 in patients with primary sclerosing cholangitis and other autoimmune diseases. Journal of Autoimmunity, 2008, 30, 273-282.	6.7	17

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37	A segmental maximum a posteriori approach to genome-wide copy number profiling. Bioinformatics, 2008, 24, 751-758.	4.2	30
38	The Mechanism of Cystic Fibrosis Transmembrane Conductance Regulator Transcriptional Repression during the Unfolded Protein Response. Journal of Biological Chemistry, 2008, 283, 12154-12165.	3.5	66
39	High-Resolution DNA Copy Number Profiling of Malignant Peripheral Nerve Sheath Tumors Using Targeted Microarray-Based Comparative Genomic Hybridization. Clinical Cancer Research, 2008, 14, 1015-1024.	7.2	120
40	A previously unrecognized microdeletion syndrome on chromosome 22 band q11.2 encompassing the BCR gene. American Journal of Medical Genetics, Part A, 2007, 143A, 2178-2184.	1.5	42
41	Comprehensive genetic and epigenetic analysis of sporadic meningioma for macro-mutations on 22q and micro-mutations within the NF2 locus. BMC Genomics, 2007, 8, 16.	2.9	68
42	Analysis of copy number variation in the normal human population within a region containing complex segmental duplications on 22q11 using high-resolution array-CGH. Genomics, 2006, 88, 152-162.	2.9	13
43	Microarray-based survey of CpG islands identifies concurrent hyper- and hypomethylation patterns in tissues derived from patients with breast cancer. Genes Chromosomes and Cancer, 2006, 45, 656-667.	3.3	41
44	Detailed assessment of chromosome 22 aberrations in sporadic pheochromocytoma using array-CGH. International Journal of Cancer, 2006, 118, 1159-1164.	5.4	24
45	Identification of limited regions of genetic aberrations in patients affected with Wilms' tumor using a tiling-path chromosome 22 array. International Journal of Cancer, 2006, 119, 571-578.	5.4	10
46	Copy-number polymorphisms: mining the tip of an iceberg. Trends in Genetics, 2005, 21, 315-317.	6.9	46
47	Highâ€resolution gene copy number and expression profiling of human chromosome 22 in ovarian carcinomas. Genes Chromosomes and Cancer, 2005, 42, 228-237.	3.3	21
48	Localization of a putative low-penetrance ependymoma susceptibility locus to 22q11 using a chromosome 22 tiling-path genomic microarray. Genes Chromosomes and Cancer, 2005, 43, 329-338.	3.3	26
49	Chromosome 22 tiling-path array-CGH analysis identifies germ-line- and tumor-specific aberrations in patients with glioblastoma multiforme. Genes Chromosomes and Cancer, 2005, 44, 161-169.	3.3	33
50	Identification of genetic aberrations on chromosome 22 outside theNF2locus in schwannomatosis and neurofibromatosis type 2. Human Mutation, 2005, 26, 540-549.	2.8	29
51	High-resolution array-CGH profiling of germline and tumor-specific copy number alterations on chromosome 22 in patients affected with schwannomas. Human Genetics, 2005, 118, 35-44.	3.8	19
52	Exon Array CGH: Detection of Copy-Number Changes at the Resolution of Individual Exons in the Human Genome. American Journal of Human Genetics, 2005, 76, 750-762.	6.1	132
53	LARGE can functionally bypass Î±-dystroglycan glycosylation defects in distinct congenital muscular dystrophies. Nature Medicine, 2004, 10, 696-703.	30.1	253
54	Genomic microarrays in the spotlight. Trends in Genetics, 2004, 20, 87-94.	6.9	155

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55	Molecular Recognition by LARGE Is Essential for Expression of Functional Dystroglycan. Cell, 2004, 117, 953-964.	27.8	245
56	NF2 Tumor Suppressor Gene: A Comprehensive and Efficient Detection of Somatic Mutations by Denaturing HPLC and Microarray-CGH. NeuroMolecular Medicine, 2003, 3, 41-52.	3.4	12
57	Development of NF2 gene specific, strictly sequence defined diagnostic microarray for deletion detection. Journal of Molecular Medicine, 2003, 81, 443-451.	4.0	31
58	Strong conservation of the human NF2 locus based on sequence comparison in five species. Mammalian Genome, 2003, 14, 526-536.	2.3	7
59	Does chromosome 22 have anything to do with sex determination: Further studies on a 46,XX,22q11.2 del male. American Journal of Medical Genetics Part A, 2003, 123A, 64-67.	2.3	10
60	Coincidence of synteny breakpoints with malignancy-related deletions on human chromosome 3. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 6622-6627.	7.6	14
61	The transcriptional map of the common eliminated region 1 (C3CER1) in 3p21.3. European Journal of Human Genetics, 2002, 10, 52-61.	2.9	37
62	A full-coverage, high-resolution human chromosome 22 genomic microarray for clinical and research applications. Human Molecular Genetics, 2002, 11, 3221-3229.	3.0	131
63	Mouse cytosolic and mitochondrial deoxyribonucleotidases: cDNA cloning of the mitochondrial enzyme, gene structures, chromosomal mapping and comparison with the human orthologs. Gene, 2002, 294, 109-117.	2.3	10
64	Comparative human/murine sequence analysis of the common eliminated region 1 from human 3p21.3. Mammalian Genome, 2002, 13, 646-655.	2.3	16
65	The LZTFL1 Gene Is a Part of a Transcriptional Map Covering 250 kb within the Common Eliminated Region 1 (C3CER1) in 3p21.3. Genomics, 2001, 73, 10-19.	2.9	34
66	Analysis of short stature homeobox-containing gene (SHOX) and auxological phenotype in dyschondrosteosis and isolated Madelung deformity. Human Genetics, 2001, 109, 551-558.	3.8	60
67	High resolution deletion analysis of constitutional DNA from neurofibromatosis type 2 (NF2) patients using microarray-CGH. Human Molecular Genetics, 2001, 10, 271-282.	3.0	148
68	A case of dermatofibrosarcoma protuberans of the vulva with a COL1A1/PDGFB fusion identical to a case of giant cell fibroblastoma. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2000, 437, 95-100.	2.9	43
69	Fine mapping of the constitutional translocation t(11;22)(q23;q11). Human Genetics, 2000, 106, 506-516.	3.8	20
70	Characterization of Five Novel Human Genes in the 11q13-q22 Region. Biochemical and Biophysical Research Communications, 2000, 273, 90-94.	2.2	13
71	Duplications on Human Chromosome 22 Reveal a Novel Ret Finger Protein-Like Gene Family with Sense and Endogenous Antisense Transcripts. Genome Research, 1999, 9, 803-814.	5.6	32
72	Psoriasis Upregulated Phorbolin-1 Shares Structural but not Functional Similarity to the mRNA-Editing Protein Apobec-1. Journal of Investigative Dermatology, 1999, 113, 162-169.	0.7	54

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73	Severe phenotype of neurofibromatosis type 2 in a patient with a 7.4-MB constitutional deletion on chromosome 22: Possible localization of a neurofibromatosis type 2 modifier gene?. Genes Chromosomes and Cancer, 1999, 25, 184-190.	3.3	37
74	TOM1Genes Map to Human Chromosome 22q13.1 and Mouse Chromosome 8C1 and Encode Proteins Similar to the Endosomal Proteins HGS and STAM. Genomics, 1999, 57, 380-388.	2.9	26
75	A 1-Mb PAC Contig Spanning the Common Eliminated Region 1 (CER1) in Microcell Hybrid-Derived SCID Tumors. Genomics, 1999, 62, 147-155.	2.9	28
76	Genomic Structure, 5â€² Flanking Sequences, and Precise Localization in 1P31.1 of the Human Prostaglandin F Receptor Gene. Biochemical and Biophysical Research Communications, 1999, 254, 413-416.	2.2	17
77	The Mouse Ortholog of the HumanSMARCB1Gene Encodes Two Splice Forms. Biochemical and Biophysical Research Communications, 1999, 257, 886-890.	2.2	27
78	Various regions within the alpha-helical domain of theCOL1A1 gene are fused to the second exon of thePDGFB gene in dermatofibrosarcomas and giant-cell fibroblastomas. Genes Chromosomes and Cancer, 1998, 23, 187-193.	3.3	160
79	Characterization of the human NIPSNAP1 gene from 22q12: a member of a novel gene family. Gene, 1998, 212, 13-20.	2.3	48
80	A case of dermatofibrosarcoma protuberans with a ring chromosome 5 and a rearranged chromosome 22 containing amplified COL1A1 and PDGFB sequences. Cancer Letters, 1998, 133, 129-134.	7.3	35
81	Cloning, Expression Pattern, and Chromosomal Assignment to 16q23 of the Human Î³-Adaptin Gene (ADTG). Genomics, 1998, 50, 275-280.	2.9	7
82	Deregulation of the platelet-derived growth factor Î²-chain gene via fusion with collagen gene COL1A1 in dermatof ibrosarcoma protuberans and giant-cell fibroblastoma. Nature Genetics, 1997, 15, 95-98.	20.4	513
83	Characterization of the mouse beta-prime adaptin gene; cDNA sequence, genomic structure, and chromosomal localization. Mammalian Genome, 1997, 8, 651-656.	2.3	7
84	1p and 3p deletions in meningiomas without detectable aberrations of chromosome 22 identified by comparative genomic hybridization. Genes Chromosomes and Cancer, 1997, 20, 419-424.	3.3	30
85	Regional Localization of over 300 Loci on Human Chromosome 22 Using a Somatic Cell Hybrid Mapping Panel. Genomics, 1996, 35, 275-288.	2.9	28
86	Structure of the Promoter and Genomic Organization of the Human Î²â€²-Adaptin Gene (BAM22) from Chromosome 22q12. Genomics, 1996, 36, 112-117.	2.9	27
87	Sequence and Expression of the Mouse Homologue to Human Phospholipase C Î²3 Neighboring Gene. Biochemical and Biophysical Research Communications, 1996, 223, 335-340.	2.2	9
88	Characterization of a second human clathrin heavy chain polypeptide gene (CLH-22) from chromosome 22q11. Human Molecular Genetics, 1996, 5, 625-631.	3.0	54
89	Isolation and mapping of cosmid markers on human chromosome 22, including one within the submicroscopically deleted region of DiGeorge syndrome. Human Genetics, 1994, 93, 248-254.	3.8	30
90	Deletions on chromosome 22 in sporadic meningioma. Genes Chromosomes and Cancer, 1994, 10, 122-130.	3.3	116

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91	Chromosomal deletions in anaplastic meningiomas suggest multiple regions outside chromosome 22 as important in tumor progression. International Journal of Cancer, 1994, 56, 354-357.	5.4	91
92	Evidence for the complete inactivation of the NF2 gene in the majority of sporadic meningiomas. Nature Genetics, 1994, 6, 180-184.	20.4	518
93	Physical Mapping of the NF2/Meningioma Region on Human Chromosome 22q12. Genomics, 1994, 19, 52-59.	2.9	18
94	Regional fine mapping of the Î² crystallin genes on chromosome 22 excludes these genes as physically linked markers for neurofibromatosis type 2. Genes Chromosomes and Cancer, 1993, 8, 112-118.	3.3	10
95	The Genes for Oncostatin M (OSM) and Leukemia Inhibitory Factor (LIF) Are Tightly Linked on Human Chromosome 22. Genomics, 1993, 17, 136-140.	2.9	52
96	Microdeletions within 22q11 associated with sporadic and familial DiGeorge syndrome. Genomics, 1991, 10, 201-206.	2.9	241
97	A map of 22 loci on human chromosome 22. Genomics, 1991, 11, 709-719.	2.9	30
98	The Molecular Genetics of Meningiomas. Brain Pathology, 1990, 1, 19-24.	4.2	55
99	Isolation of anonymous, polymorphic DNA fragments from human chromosome 22q12-qter. Human Genetics, 1990, 84, 219-222.	3.8	26