

Jan P Dumanski

List of Publications by Year in descending order

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docs citations

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times ranked

9215
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#	ARTICLE	IF	CITATIONS
1	Phenotypically Concordant and Discordant Monozygotic Twins Display Different DNA Copy-Number-Variation Profiles. <i>American Journal of Human Genetics</i> , 2008, 82, 763-771.	6.1	535
2	Evidence for the complete inactivation of the NF2 gene in the majority of sporadic meningiomas. <i>Nature Genetics</i> , 1994, 6, 180-184.	20.4	518
3	Deregulation of the platelet-derived growth factor β -chain gene via fusion with collagen gene COL1A1 in dermatofibrosarcoma protuberans and giant-cell fibroblastoma. <i>Nature Genetics</i> , 1997, 15, 95-98.	20.4	513
4	Mosaic loss of chromosome Y in peripheral blood is associated with shorter survival and higher risk of cancer. <i>Nature Genetics</i> , 2014, 46, 624-628.	20.4	338
5	LARGE can functionally bypass β -dystroglycan glycosylation defects in distinct congenital muscular dystrophies. <i>Nature Medicine</i> , 2004, 10, 696-703.	30.1	253
6	Molecular Recognition by LARGE Is Essential for Expression of Functional Dystroglycan. <i>Cell</i> , 2004, 117, 953-964.	27.8	245
7	Microdeletions within 22q11 associated with sporadic and familial DiGeorge syndrome. <i>Genomics</i> , 1991, 10, 201-206.	2.9	241
8	Genetic predisposition to mosaic Y chromosome loss in blood. <i>Nature</i> , 2019, 575, 652-657.	36.2	217
9	Mosaicism in health and disease “ clones picking up speed. <i>Nature Reviews Genetics</i> , 2017, 18, 128-142.	16.7	214
10	Somatic mosaicism for copy number variation in differentiated human tissues. <i>Human Mutation</i> , 2008, 29, 1118-1124.	2.8	186
11	Age-Related Somatic Structural Changes in the Nuclear Genome of Human Blood Cells. <i>American Journal of Human Genetics</i> , 2012, 90, 217-228.	6.1	176
12	Mosaic Loss of Chromosome Y in Blood Is Associated with Alzheimer Disease. <i>American Journal of Human Genetics</i> , 2016, 98, 1208-1219.	6.1	175
13	Various regions within the alpha-helical domain of the COL1A1 gene are fused to the second exon of the PDGFB gene in dermatofibrosarcomas and giant-cell fibroblastomas. <i>Genes Chromosomes and Cancer</i> , 1998, 23, 187-193.	3.3	160
14	Genomic microarrays in the spotlight. <i>Trends in Genetics</i> , 2004, 20, 87-94.	6.9	155
15	High resolution deletion analysis of constitutional DNA from neurofibromatosis type 2 (NF2) patients using microarray-CGH. <i>Human Molecular Genetics</i> , 2001, 10, 271-282.	3.0	148
16	Exon Array CGH: Detection of Copy-Number Changes at the Resolution of Individual Exons in the Human Genome. <i>American Journal of Human Genetics</i> , 2005, 76, 750-762.	6.1	132
17	A full-coverage, high-resolution human chromosome 22 genomic microarray for clinical and research applications. <i>Human Molecular Genetics</i> , 2002, 11, 3221-3229.	3.0	131
18	High-Resolution DNA Copy Number Profiling of Malignant Peripheral Nerve Sheath Tumors Using Targeted Microarray-Based Comparative Genomic Hybridization. <i>Clinical Cancer Research</i> , 2008, 14, 1015-1024.	7.2	120

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19	Deletions on chromosome 22 in sporadic meningioma. <i>Genes Chromosomes and Cancer</i> , 1994, 10, 122-130.	3.3	116
20	Chromosomal deletions in anaplastic meningiomas suggest multiple regions outside chromosome 22 as important in tumor progression. <i>International Journal of Cancer</i> , 1994, 56, 354-357.	5.4	91
21	Common pathogenetic mechanism involving human chromosome 18 in familial and sporadic ileal carcinoid tumors. <i>Genes Chromosomes and Cancer</i> , 2011, 50, 82-94.	3.3	81
22	Comprehensive genetic and epigenetic analysis of sporadic meningioma for macro-mutations on 22q and micro-mutations within the NF2 locus. <i>BMC Genomics</i> , 2007, 8, 16.	2.9	68
23	The Mechanism of Cystic Fibrosis Transmembrane Conductance Regulator Transcriptional Repression during the Unfolded Protein Response. <i>Journal of Biological Chemistry</i> , 2008, 283, 12154-12165.	3.5	66
24	Immune cells lacking Y chromosome show dysregulation of autosomal gene expression. <i>Cellular and Molecular Life Sciences</i> , 2021, 78, 4019-4033.	5.5	62
25	Analysis of short stature homeobox-containing gene (SHOX) and auxological phenotype in dyschondrosteosis and isolated Madelung deformity. <i>Human Genetics</i> , 2001, 109, 551-558.	3.8	60
26	Focal amplifications are associated with high grade and recurrences in stage Ta bladder carcinoma. <i>International Journal of Cancer</i> , 2010, 126, 1390-1402.	5.4	59
27	The Molecular Genetics of Meningiomas. <i>Brain Pathology</i> , 1990, 1, 19-24.	4.2	55
28	Tissue-specific variation in DNA methylation levels along human chromosome 1. <i>Epigenetics and Chromatin</i> , 2009, 2, 7.	3.9	55
29	Characterization of a second human clathrin heavy chain polypeptide gene (CLH-22) from chromosome 22q11. <i>Human Molecular Genetics</i> , 1996, 5, 625-631.	3.0	54
30	Psoriasis Upregulated Phorbol-1 Shares Structural but not Functional Similarity to the mRNA-Editing Protein Apobec-1. <i>Journal of Investigative Dermatology</i> , 1999, 113, 162-169.	0.7	54
31	Mosaic loss of chromosome Y in leukocytes matters. <i>Nature Genetics</i> , 2019, 51, 4-7.	20.4	53
32	Longitudinal changes in the frequency of mosaic chromosome Y loss in peripheral blood cells of aging men varies profoundly between individuals. <i>European Journal of Human Genetics</i> , 2020, 28, 349-357.	2.9	53
33	The Genes for Oncostatin M (OSM) and Leukemia Inhibitory Factor (LIF) Are Tightly Linked on Human Chromosome 22. <i>Genomics</i> , 1993, 17, 136-140.	2.9	52
34	Genome-wide high-resolution analysis of DNA copy number alterations in NF1-associated malignant peripheral nerve sheath tumors using 32K BAC array. <i>Genes Chromosomes and Cancer</i> , 2009, 48, 897-907.	3.3	50
35	Characterization of the human NIPSNAP1 gene from 22q12: a member of a novel gene family. <i>Gene</i> , 1998, 212, 13-20.	2.3	48
36	Profiling of copy number variations (CNVs) in healthy individuals from three ethnic groups using a human genome 32K BAC-clone-based array. <i>Human Mutation</i> , 2008, 29, 398-408.	2.8	47

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37	Copy-number polymorphisms: mining the tip of an iceberg. <i>Trends in Genetics</i> , 2005, 21, 315-317.	6.9	46
38	A case of dermatofibrosarcoma protuberans of the vulva with a COL1A1/PDGFB fusion identical to a case of giant cell fibroblastoma. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2000, 437, 95-100.	2.9	43
39	Characterization of novel and complex genomic aberrations in glioblastoma using a 32K BAC array. <i>Neuro-Oncology</i> , 2009, 11, 803-818.	1.2	43
40	A previously unrecognized microdeletion syndrome on chromosome 22 band q11.2 encompassing the <i>BCR</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2178-2184.	1.5	42
41	Frequent genetic differences between matched primary and metastatic breast cancer provide an approach to identification of biomarkers for disease progression. <i>European Journal of Human Genetics</i> , 2010, 18, 560-568.	2.9	42
42	Microarray-based survey of CpG islands identifies concurrent hyper- and hypomethylation patterns in tissues derived from patients with breast cancer. <i>Genes Chromosomes and Cancer</i> , 2006, 45, 656-667.	3.3	41
43	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?. <i>Genes Chromosomes and Cancer</i> , 1999, 25, 184-190.	3.3	37
44	The transcriptional map of the common eliminated region 1 (C3CER1) in 3p21.3. <i>European Journal of Human Genetics</i> , 2002, 10, 52-61.	2.9	37
45	A case of dermatofibrosarcoma protuberans with a ring chromosome 5 and a rearranged chromosome 22 containing amplified COL1A1 and PDGFB sequences. <i>Cancer Letters</i> , 1998, 133, 129-134.	7.3	35
46	The LZTFL1 Gene Is a Part of a Transcriptional Map Covering 250 kb within the Common Eliminated Region 1 (C3CER1) in 3p21.3. <i>Genomics</i> , 2001, 73, 10-19.	2.9	34
47	Structural Genetic Variation in the Context of Somatic Mosaicism. <i>Methods in Molecular Biology</i> , 2012, 838, 249-272.	0.0	34
48	Chromosome 22 tiling-path array-CGH analysis identifies germ-line- and tumor-specific aberrations in patients with glioblastoma multiforme. <i>Genes Chromosomes and Cancer</i> , 2005, 44, 161-169.	3.3	33
49	Duplications on Human Chromosome 22 Reveal a Novel Ret Finger Protein-Like Gene Family with Sense and Endogenous Antisense Transcripts. <i>Genome Research</i> , 1999, 9, 803-814.	5.6	32
50	Integrative epigenomic and genomic analysis of malignant pheochromocytoma. <i>Experimental and Molecular Medicine</i> , 2010, 42, 484.	7.8	32
51	Development of NF2 gene specific, strictly sequence defined diagnostic microarray for deletion detection. <i>Journal of Molecular Medicine</i> , 2003, 81, 443-451.	4.0	31
52	A map of 22 loci on human chromosome 22. <i>Genomics</i> , 1991, 11, 709-719.	2.9	30
53	Isolation and mapping of cosmid markers on human chromosome 22, including one within the submicroscopically deleted region of DiGeorge syndrome. <i>Human Genetics</i> , 1994, 93, 248-254.	3.8	30
54	1p and 3p deletions in meningiomas without detectable aberrations of chromosome 22 identified by comparative genomic hybridization. <i>Genes Chromosomes and Cancer</i> , 1997, 20, 419-424.	3.3	30

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55	Distal 22q11.2 microduplication encompassing the <i>BCR</i> gene. American Journal of Medical Genetics, Part A, 2008, 146A, 3075-3081.	1.5	30
56	A segmental maximum a posteriori approach to genome-wide copy number profiling. Bioinformatics, 2008, 24, 751-758.	4.2	30
57	Identification of genetic aberrations on chromosome 22 outside the NF2 locus in schwannomatosis and neurofibromatosis type 2. Human Mutation, 2005, 26, 540-549.	2.8	29
58	Leukocytes with chromosome Y loss have reduced abundance of the cell surface immunoprotein CD99. Scientific Reports, 2021, 11, 15160.	3.4	29
59	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
60	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
61	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
62	Structure of the Promoter and Genomic Organization of the Human <i>BAM22</i> Gene (BAM22) from Chromosome 22q12. Genomics, 1996, 36, 112-117.	2.9	27
63	The Mouse Ortholog of the Human <i>SMARCB1</i> Gene Encodes Two Splice Forms. Biochemical and Biophysical Research Communications, 1999, 257, 886-890.	2.2	27
64	Loss of Y and clonal hematopoiesis in blood – two sides of the same coin?. Leukemia, 2022, 36, 889-891.	7.5	27
65	Isolation of anonymous, polymorphic DNA fragments from human chromosome 22q12-qter. Human Genetics, 1990, 84, 219-222.	3.8	26
66	TOM1 Genes 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
67	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
68	Detailed assessment of chromosome 22 aberrations in sporadic pheochromocytoma using array-CGH. International Journal of Cancer, 2006, 118, 1159-1164.	5.4	24
69	High-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
70	Fine mapping of the constitutional translocation t(11;22)(q23;q11). Human Genetics, 2000, 106, 506-516.	3.8	20
71	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
72	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

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73	Physical Mapping of the NF2/Meningioma Region on Human Chromosome 22q12. <i>Genomics</i> , 1994, 19, 52-59.	2.9	18
74	Genomic Structure, 5' Flanking Sequences, and Precise Localization in 1P31.1 of the Human Prostaglandin F Receptor Gene. <i>Biochemical and Biophysical Research Communications</i> , 1999, 254, 413-416.	2.2	17
75	Autoantibodies to glutathione S-transferase theta 1 in patients with primary sclerosing cholangitis and other autoimmune diseases. <i>Journal of Autoimmunity</i> , 2008, 30, 273-282.	6.7	17
76	Comparative human/murine sequence analysis of the common eliminated region 1 from human 3p21.3. <i>Mammalian Genome</i> , 2002, 13, 646-655.	2.3	16
77	Coincidence of synteny breakpoints with malignancy-related deletions on human chromosome 3. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2003, 100, 6622-6627.	7.6	14
78	Characterization of Five Novel Human Genes in the 11q13-q22 Region. <i>Biochemical and Biophysical Research Communications</i> , 2000, 273, 90-94.	2.2	13
79	Analysis of copy number variation in the normal human population within a region containing complex segmental duplications on 22q11 using high-resolution array-CGH. <i>Genomics</i> , 2006, 88, 152-162.	2.9	13
80	NF2 Tumor Suppressor Gene: A Comprehensive and Efficient Detection of Somatic Mutations by Denaturing HPLC and Microarray-CGH. <i>NeuroMolecular Medicine</i> , 2003, 3, 41-52.	3.4	12
81	Concurrent DNA Copy-Number Alterations and Mutations in Genes Related to Maintenance of Genome Stability in Uninvolved Mammary Glandular Tissue from Breast Cancer Patients. <i>Human Mutation</i> , 2015, 36, 1088-1099.	2.8	12
82	PRR14L mutations are associated with chromosome 22 acquired uniparental disomy, age-related clonal hematopoiesis and myeloid neoplasia. <i>Leukemia</i> , 2019, 33, 1184-1194.	7.5	11
83	Regional fine mapping of the β crystallin genes on chromosome 22 excludes these genes as physically linked markers for neurofibromatosis type 2. <i>Genes Chromosomes and Cancer</i> , 1993, 8, 112-118.	3.3	10
84	Mouse cytosolic and mitochondrial deoxyribonucleotidases: cDNA cloning of the mitochondrial enzyme, gene structures, chromosomal mapping and comparison with the human orthologs. <i>Gene</i> , 2002, 294, 109-117.	2.3	10
85	Does chromosome 22 have anything to do with sex determination: Further studies on a 46,XX,22q11.2 del male. <i>American Journal of Medical Genetics Part A</i> , 2003, 123A, 64-67.	2.3	10
86	Identification of limited regions of genetic aberrations in patients affected with Wilms' tumor using a tiling-path chromosome 22 array. <i>International Journal of Cancer</i> , 2006, 119, 571-578.	5.4	10
87	Sequence and Expression of the Mouse Homologue to Human Phospholipase C β 3 Neighboring Gene. <i>Biochemical and Biophysical Research Communications</i> , 1996, 223, 335-340.	2.2	9
88	Procoagulant activity in patients with sickle cell trait. <i>Blood Coagulation and Fibrinolysis</i> , 2012, 23, 268-270.	1.1	9
89	Loss of Y in leukocytes as a risk factor for critical COVID-19 in men. <i>Genome Medicine</i> , 2022, 14, .	8.5	9
90	Genome-wide microarray-based comparative genomic hybridization analysis of lymphoplasmacytic lymphomas reveals heterogeneous aberrations. <i>Leukemia and Lymphoma</i> , 2009, 50, 1528-1534.	1.4	8

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91	Variable degree of mosaicism for tetrasomy 18p in phenotypically discordant monozygotic twinsâ€™ Diagnostic implications. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1526.	1.3	8
92	High prevalence of somatic PIK3CA and TP53 pathogenic variants in the normal mammary gland tissue of sporadic breast cancer patients revealed by duplex sequencing. <i>Npj Breast Cancer</i> , 2022, 8, .	5.4	8
93	Characterization of the mouse beta-prime adaptin gene; cDNA sequence, genomic structure, and chromosomal localization. <i>Mammalian Genome</i> , 1997, 8, 651-656.	2.3	7
94	Cloning, Expression Pattern, and Chromosomal Assignment to 16q23 of the Human Î³-Adaptin Gene (ADTG). <i>Genomics</i> , 1998, 50, 275-280.	2.9	7
95	Strong conservation of the human NF2 locus based on sequence comparison in five species. <i>Mammalian Genome</i> , 2003, 14, 526-536.	2.3	7
96	Comprehensive cancer-oriented biobanking resource of human samples for studies of post-zygotic genetic variation involved in cancer predisposition. <i>PLoS ONE</i> , 2022, 17, e0266111.	2.5	6
97	Loss of Chromosome Y in Leukocytes and Major Cardiovascular Events. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, e001820.	5.1	5
98	Mosaic Loss of Chromosome Y Is Associated With Functional Outcome After Ischemic Stroke. <i>Stroke</i> , 2023, 54, 2434-2437.	5.3	3
99	Post-Zygotic and Inter-Individual Structural Genetic Variation in a Presumptive Enhancer Element of the Locus between the IL10RÎ² and IFNAR1 Genes. <i>PLoS ONE</i> , 2013, 8, e67752.	2.5	2