

# Uta Francke

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

176 papers	21,250 citations	74 h-index	144 g-index
183 ext. papers	22,703 ext. citations	13.7 avg, IF	6.12 L-index

#	Paper	IF	Citations
176	MARFAN SYNDROME <b>2021</b> , 577-596		
175	Report of the Phenotype of a Patient with Roberts Syndrome and a Rare Variant. <i>Journal of Pediatric Genetics</i> , <b>2020</b> , 9, 58-62	0.7	1
174	2012 William Allan Award: Adventures in cytogenetics. <i>American Journal of Human Genetics</i> , <b>2013</b> , 92, 325-37	11	2
173	Dealing with the unexpected: consumer responses to direct-access BRCA mutation testing. <i>PeerJ</i> , <b>2013</b> , 1, e8	3.1	55
172	Genome-wide analysis points to roles for extracellular matrix remodeling, the visual cycle, and neuronal development in myopia. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003299	6	190
171	Genetic variants associated with breast size also influence breast cancer risk. <i>BMC Medical Genetics</i> , <b>2012</b> , 13, 53	2.1	50
170	Novel associations for hypothyroidism include known autoimmune risk loci. <i>PLoS ONE</i> , <b>2012</b> , 7, e34442	3.7	105
169	Comparison of family history and SNPs for predicting risk of complex disease. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002973	6	78
168	Reduction of NADPH-oxidase activity ameliorates the cardiovascular phenotype in a mouse model of Williams-Beuren Syndrome. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002458	6	26
167	Skeletogenic phenotype of human Marfan embryonic stem cells faithfully phenocopied by patient-specific induced-pluripotent stem cells. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2012</b> , 109, 215-20	11.5	60
166	Efficient replication of over 180 genetic associations with self-reported medical data. <i>PLoS ONE</i> , <b>2011</b> , 6, e23473	3.7	101
165	Prognosis factors in probands with an FBN1 mutation diagnosed before the age of 1 year. <i>Pediatric Research</i> , <b>2011</b> , 69, 265-70	3.2	48
164	Control of bone formation by the serpentine receptor Frizzled-9. <i>Journal of Cell Biology</i> , <b>2011</b> , 192, 1057-72	7.2	84
163	Induced chromosome deletion in a Williams-Beuren syndrome mouse model causes cardiovascular abnormalities. <i>Journal of Vascular Research</i> , <b>2011</b> , 48, 119-29	1.9	18
162	Web-based genome-wide association study identifies two novel loci and a substantial genetic component for Parkinson's disease. <i>PLoS Genetics</i> , <b>2011</b> , 7, e1002141	6	387
161	Cardiovascular manifestations in men and women carrying a FBN1 mutation. <i>European Heart Journal</i> , <b>2010</b> , 31, 2223-9	9.5	98
160	Marfan Syndrome <b>2010</b> , 495-515		

159	Neonatal maternal deprivation response and developmental changes in gene expression revealed by hypothalamic gene expression profiling in mice. <i>PLoS ONE</i> , <b>2010</b> , 5, e9402	3.7	22
158	Clinical and molecular study of 320 children with Marfan syndrome and related type I fibrillinopathies in a series of 1009 probands with pathogenic FBN1 mutations. <i>Pediatrics</i> , <b>2009</b> , 123, 391-8	7.4	120
157	Widespread changes in dendritic and axonal morphology in Mecp2-mutant mouse models of Rett syndrome: evidence for disruption of neuronal networks. <i>Journal of Comparative Neurology</i> , <b>2009</b> , 514, 240-58	3.4	194
156	Induced chromosome deletions cause hypersociability and other features of Williams-Beuren syndrome in mice. <i>EMBO Molecular Medicine</i> , <b>2009</b> , 1, 50-65	12	74
155	Partial trisomy for the distal long arm of chromosome 5 (region q34-qter). A new clinically recognizable syndrome. <i>Clinical Genetics</i> , <b>2008</b> , 15, 454-461	4	28
154	Comparative study of brain morphology in Mecp2 mutant mouse models of Rett syndrome. <i>Journal of Comparative Neurology</i> , <b>2008</b> , 508, 184-95	3.4	98
153	SnoRNA Snord116 (Pwcr1/MBII-85) deletion causes growth deficiency and hyperphagia in mice. <i>PLoS ONE</i> , <b>2008</b> , 3, e1709	3.7	218
152	Cerebellar gene expression profiles of mouse models for Rett syndrome reveal novel MeCP2 targets. <i>BMC Medical Genetics</i> , <b>2007</b> , 8, 36	2.1	97
151	A Marfan syndrome gene expression phenotype in cultured skin fibroblasts. <i>BMC Genomics</i> , <b>2007</b> , 8, 3194.5	17	
150	An atypical deletion of the Williams-Beuren syndrome interval implicates genes associated with defective visuospatial processing and autism. <i>Journal of Medical Genetics</i> , <b>2007</b> , 44, 136-43	5.8	82
149	DLX5 and DLX6 expression is biallelic and not modulated by MeCP2 deficiency. <i>American Journal of Human Genetics</i> , <b>2007</b> , 81, 492-506	11	44
148	Mechanisms of disease: neurogenetics of MeCP2 deficiency. <i>Nature Clinical Practice Neurology</i> , <b>2006</b> , 2, 212-21		45
147	Identification of cis-regulatory elements for MECP2 expression. <i>Human Molecular Genetics</i> , <b>2006</b> , 15, 1769-82	5.6	55
146	Ube3a expression is not altered in Mecp2 mutant mice. <i>Human Molecular Genetics</i> , <b>2006</b> , 15, 2210-5	5.6	26
145	NFAT dysregulation by increased dosage of DSCR1 and DYRK1A on chromosome 21. <i>Nature</i> , <b>2006</b> , 441, 595-600	50.4	534
144	Inactivating mutations in ESCO2 cause SC phocomelia and Roberts syndrome: no phenotype-genotype correlation. <i>American Journal of Human Genetics</i> , <b>2005</b> , 77, 1117-28	11	108
143	Frizzled 9 knock-out mice have abnormal B-cell development. <i>Blood</i> , <b>2005</b> , 105, 2487-94	2.2	86
142	Lack of Pwcr1/MBII-85 snoRNA is critical for neonatal lethality in Prader-Willi syndrome mouse models. <i>Mammalian Genome</i> , <b>2005</b> , 16, 424-31	3.2	69

141	Molecular breakpoint cloning and gene expression studies of a novel translocation t(4;15)(q27;q11.2) associated with Prader-Willi syndrome. <i>BMC Medical Genetics</i> , <b>2005</b> , 6, 18	2.1	48
140	Normal histone modifications on the inactive X chromosome in ICF and Rett syndrome cells: implications for methyl-CpG binding proteins. <i>BMC Biology</i> , <b>2004</b> , 2, 21	7.3	18
139	Gene expression patterns vary in clonal cell cultures from Rett syndrome females with eight different MECP2 mutations. <i>BMC Medical Genetics</i> , <b>2002</b> , 3, 12	2.1	79
138	Premature termination mutations in FBN1: distinct effects on differential allelic expression and on protein and clinical phenotypes. <i>American Journal of Human Genetics</i> , <b>2002</b> , 71, 223-37	11	113
137	Evidence for the role of PWCR1/HBII-85 C/D box small nucleolar RNAs in Prader-Willi syndrome. <i>American Journal of Human Genetics</i> , <b>2002</b> , 71, 669-78	11	104
136	frizzled 9 is expressed in neural precursor cells in the developing neural tube. <i>Development Genes and Evolution</i> , <b>2001</b> , 211, 453-7	1.8	27
135	Multi-exon deletions of the FBN1 gene in Marfan syndrome. <i>BMC Medical Genetics</i> , <b>2001</b> , 2, 11	2.1	42
134	MECP2 truncating mutations cause histone H4 hyperacetylation in Rett syndrome. <i>Human Molecular Genetics</i> , <b>2001</b> , 10, 1085-92	5.6	57
133	Guidelines for reporting clinical features in cases with MECP2 mutations. <i>Brain and Development</i> , <b>2001</b> , 23, 208-11	2.2	121
132	Spectrum of MECP2 mutations in Rett syndrome. <i>Brain and Development</i> , <b>2001</b> , 23 Suppl 1, S138-43	2.2	33
131	Williams (Williams Beuren) syndrome: a distinct neurobehavioral disorder. <i>Journal of Child Neurology</i> , <b>2001</b> , 16, 177-90	2.5	47
130	Manifestations and linkage analysis in X-linked autoimmunity-immunodeficiency syndrome. <i>American Journal of Medical Genetics Part A</i> , <b>2000</b> , 90, 390-7		59
129	Diverse deletions in the growth hormone receptor gene cause growth hormone insensitivity syndrome. <i>Human Mutation</i> , <b>2000</b> , 16, 323-33	4.7	25
128	A physical map, including a BAC/PAC clone contig, of the Williams-Beuren syndrome--deletion region at 7q11.23. <i>American Journal of Human Genetics</i> , <b>2000</b> , 66, 47-68	11	141
127	Small evolutionarily conserved RNA, resembling C/D box small nucleolar RNA, is transcribed from PWCR1, a novel imprinted gene in the Prader-Willi deletion region, which is highly expressed in brain. <i>American Journal of Human Genetics</i> , <b>2000</b> , 67, 1067-82	11	125
126	Diverse deletions in the growth hormone receptor gene cause growth hormone insensitivity syndrome <b>2000</b> , 16, 323		1
125	Growth hormone receptor deficiency in Ecuador. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1999</b> , 84, 4436-43	5.6	47
124	In vivo nuclease hypersensitivity studies reveal multiple sites of parental origin-dependent differential chromatin conformation in the 150 kb SNRPN transcription unit. <i>Human Molecular Genetics</i> , <b>1999</b> , 8, 555-66	5.6	54

123	Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. <i>Nature Genetics</i> , <b>1999</b> , 23, 185-8	36.3	3780
122	The human neuregulin-2 (NRG2) gene: cloning, mapping and evaluation as a candidate for the autosomal recessive form of Charcot-Marie-Tooth disease linked to 5q. <i>Human Genetics</i> , <b>1999</b> , 104, 326-32	6.3	21
121	Novel mutations in the Wiskott-Aldrich syndrome protein gene and their effects on transcriptional, translational, and clinical phenotypes. <i>Human Mutation</i> , <b>1999</b> , 14, 54-66	4.7	58
120	Rett syndrome and beyond: recurrent spontaneous and familial MECP2 mutations at CpG hotspots. <i>American Journal of Human Genetics</i> , <b>1999</b> , 65, 1520-9	11	417
119	Characterization and expression pattern of the frizzled gene Fzd9, the mouse homolog of FZD9 which is deleted in Williams-Beuren syndrome. <i>Genomics</i> , <b>1999</b> , 57, 235-48	4.3	75
118	Novel mutations in the Wiskott-Aldrich syndrome protein gene and their effects on transcriptional, translational, and clinical phenotypes <b>1999</b> , 14, 54		2
117	A mouse model for Prader-Willi syndrome imprinting-centre mutations. <i>Nature Genetics</i> , <b>1998</b> , 19, 25-31	36.3	255
116	Evaluation of two X chromosomal candidate genes for Rett syndrome: Glutamate dehydrogenase-2 (GLUD2) and rab GDP-dissociation inhibitor (GDI1). <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 78, 169-172		18
115	Exclusion of the gastrin-releasing peptide receptor (GRPR) locus as a candidate gene for Rett syndrome. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 78, 173-5		11
114	A severely affected male born into a Rett syndrome kindred supports X-linked inheritance and allows extension of the exclusion map. <i>American Journal of Human Genetics</i> , <b>1998</b> , 63, 267-9	11	95
113	A single mutation that results in an Asp to His substitution and partial exon skipping in a family with congenital contractural arachnodactyly. <i>Human Genetics</i> , <b>1998</b> , 103, 22-8	6.3	31
112	A mouse single-copy gene, Gtf2i, the homolog of human GTF2I, that is duplicated in the Williams-Beuren syndrome deletion region. <i>Genomics</i> , <b>1998</b> , 48, 163-70	4.3	39
111	Conserved chromosomal location and genomic structure of human and mouse fatty-acid amide hydrolase genes and evaluation of clasper as a candidate neurological mutation. <i>Genomics</i> , <b>1998</b> , 54, 408-14	4.3	26
110	Genes for the CPE receptor (CPETR1) and the human homolog of RVP1 (CPETR2) are localized within the Williams-Beuren syndrome deletion. <i>Genomics</i> , <b>1998</b> , 54, 453-9	4.3	41
109	Four contiguous amino acid substitutions, identified in patients with Laron syndrome, differently affect the binding affinity and intracellular trafficking of the growth hormone receptor. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1998</b> , 83, 4481-9	5.6	43
108	Stature in Ecuadorians heterozygous for growth hormone receptor gene E180 splice mutation does not differ from that of homozygous normal relatives. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1998</b> , 83, 2373-5	5.6	22
107	A duplicated gene in the breakpoint regions of the 7q11.23 Williams-Beuren syndrome deletion encodes the initiator binding protein TFII-I and BAP-135, a phosphorylation target of BTK. <i>Human Molecular Genetics</i> , <b>1998</b> , 7, 325-34	5.6	122
106	p48 Activates a UV-damaged-DNA binding factor and is defective in xeroderma pigmentosum group E cells that lack binding activity. <i>Molecular and Cellular Biology</i> , <b>1998</b> , 18, 4391-9	4.8	142

105	Imprinted genes in the Prader-Willi deletion. <i>Novartis Foundation Symposium</i> , <b>1998</b> , 214, 264-75; discussion 275-9		3
104	A novel human homologue of the Drosophila frizzled wnt receptor gene binds wingless protein and is in the Williams syndrome deletion at 7q11.23. <i>Human Molecular Genetics</i> , <b>1997</b> , 6, 465-72	5.6	135
103	Denaturing HPLC-identified novel FBN1 mutations, polymorphisms, and sequence variants in Marfan syndrome and related connective tissue disorders. <i>Genetic Testing and Molecular Biomarkers</i> , <b>1997</b> , 1, 237-42		82
102	Identification of a gene (GPR30) with homology to the G-protein-coupled receptor superfamily associated with estrogen receptor expression in breast cancer. <i>Genomics</i> , <b>1997</b> , 45, 607-17	4.3	419
101	Localization of the human HIP1 gene close to the elastin (ELN) locus on 7q11.23. <i>Genomics</i> , <b>1997</b> , 46, 313-5	4.3	8
100	The TSG101 tumor susceptibility gene is located in chromosome 11 band p15 and is mutated in human breast cancer. <i>Cell</i> , <b>1997</b> , 88, 143-54	56.2	109
99	A new Rett syndrome family consistent with X-linked inheritance expands the X chromosome exclusion map. <i>American Journal of Human Genetics</i> , <b>1997</b> , 61, 634-41	11	100
98	Silent mutation induces exon skipping of fibrillin-1 gene in Marfan syndrome. <i>Nature Genetics</i> , <b>1997</b> , 16, 328-9	36.3	147
97	Mutant fibrillin-1 monomers lacking EGF-like domains disrupt microfibril assembly and cause severe marfan syndrome. <i>Human Molecular Genetics</i> , <b>1996</b> , 5, 1581-7	5.6	70
96	Diagnostic test for the Prader-Willi syndrome by SNRPN expression in blood. <i>Lancet, The</i> , <b>1996</b> , 348, 1068-9	40	36
95	Structural, functional analysis and localization of the human CAP18 gene. <i>FEBS Letters</i> , <b>1996</b> , 398, 74-80	3.8	49
94	Wiskott-Aldrich syndrome protein, a novel effector for the GTPase CDC42Hs, is implicated in actin polymerization. <i>Cell</i> , <b>1996</b> , 84, 723-34	56.2	772
93	The question of heterogeneity in Marfan syndrome. <i>Nature Genetics</i> , <b>1995</b> , 9, 228-31	36.3	41
92	NFATc3, a lymphoid-specific NFATc family member that is calcium-regulated and exhibits distinct DNA binding specificity. <i>Journal of Biological Chemistry</i> , <b>1995</b> , 270, 19898-907	5.4	142
91	Cloning and characterization of a novel zinc finger gene in Xp11.2. <i>Genomics</i> , <b>1995</b> , 30, 361-5	4.3	17
90	The mouse homolog of the Wiskott-Aldrich syndrome protein (WASP) gene is highly conserved and maps near the scurfy (sf) mutation on the X chromosome. <i>Genomics</i> , <b>1995</b> , 29, 471-7	4.3	40
89	RBM3, a novel human gene in Xp11.23 with a putative RNA-binding domain. <i>Human Molecular Genetics</i> , <b>1995</b> , 4, 2307-11	5.6	92
88	Fibrillin abnormalities and prognosis in Marfan syndrome and related disorders. <i>American Journal of Medical Genetics Part A</i> , <b>1995</b> , 58, 169-76		73

87	Cloning of MITF, the human homolog of the mouse microphthalmia gene and assignment to chromosome 3p14.1-p12.3. <i>Human Molecular Genetics</i> , <b>1994</b> , 3, 553-7	5.6	164
86	The mouse pink-eyed dilution gene: association with hypopigmentation in Prader-Willi and Angelman syndromes and with human OCA2. <i>Pigment Cell &amp; Melanoma Research</i> , <b>1994</b> , 7, 398-402		24
85	Isolation of a novel gene mutated in Wiskott-Aldrich syndrome. <i>Cell</i> , <b>1994</b> , 78, 635-44	56.2	806
84	Identification of a novel paternally expressed gene in the Prader-Willi syndrome region. <i>Human Molecular Genetics</i> , <b>1994</b> , 3, 1877-82	5.6	229
83	Digitized and differentially shaded human chromosome ideograms for genomic applications. <i>Cytogenetic and Genome Research</i> , <b>1994</b> , 65, 206-18	1.9	144
82	Fibrillin genes map to regions of conserved mouse/human synteny on mouse chromosomes 2 and 18. <i>Genomics</i> , <b>1993</b> , 18, 667-72	4.3	28
81	The CD40 ligand, gp39, is defective in activated T cells from patients with X-linked hyper-IgM syndrome. <i>Cell</i> , <b>1993</b> , 72, 291-300	56.2	706
80	Mutation screening of complete fibrillin-1 coding sequence: report of five new mutations, including two in 8-cysteine domains. <i>Human Molecular Genetics</i> , <b>1993</b> , 2, 1813-21	5.6	91
79	Human dystroglycan: skeletal muscle cDNA, genomic structure, origin of tissue specific isoforms and chromosomal localization. <i>Human Molecular Genetics</i> , <b>1993</b> , 2, 1651-7	5.6	213
78	Roberts syndrome: a review of 100 cases and a new rating system for severity. <i>American Journal of Medical Genetics Part A</i> , <b>1993</b> , 47, 1104-23		149
77	Sensitivity of Roberts syndrome cells to gamma radiation, mitomycin C, and protein synthesis inhibitors. <i>Somatic Cell and Molecular Genetics</i> , <b>1993</b> , 19, 377-92		22
76	Serotonin receptor 1c gene assigned to X chromosome in human (band q24) and mouse (bands D-F4). <i>Human Molecular Genetics</i> , <b>1992</b> , 1, 681-4	5.6	63
75	Induced reciprocal translocation in transgenic mice near sites of transgene integration. <i>Mammalian Genome</i> , <b>1992</b> , 3, 209-16	3.2	8
74	Mutation analysis and prenatal diagnosis in a Lesch-Nyhan family showing non-random X-inactivation interfering with carrier detection tests. <i>Human Genetics</i> , <b>1992</b> , 89, 395-400	6.3	23
73	Trembler mouse carries a point mutation in a myelin gene. <i>Nature</i> , <b>1992</b> , 356, 241-4	50.4	400
72	A novel GC-rich human macrosatellite VNTR in Xq24 is differentially methylated on active and inactive X chromosomes. <i>Nature Genetics</i> , <b>1992</b> , 1, 137-43	36.3	78
71	The gene for the peripheral myelin protein PMP-22 is a candidate for Charcot-Marie-Tooth disease type 1A. <i>Nature Genetics</i> , <b>1992</b> , 1, 159-65	36.3	469
70	Molecular evolution of the human interleukin-8 receptor gene cluster. <i>Nature Genetics</i> , <b>1992</b> , 2, 31-6	36.3	98



69	Maternal imprinting of the mouse Snrpn gene and conserved linkage homology with the human Prader-Willi syndrome region. <i>Nature Genetics</i> , <b>1992</b> , 2, 259-64	36.3	238
68	Small nuclear ribonucleoprotein polypeptide N (SNRPN), an expressed gene in the Prader-Willi syndrome critical region. <i>Nature Genetics</i> , <b>1992</b> , 2, 265-9	36.3	242
67	Mutation creating a new splice site in the growth hormone receptor genes of 37 Ecuadorean patients with Laron syndrome. <i>Human Mutation</i> , <b>1992</b> , 1, 24-32	4.7	107
66	Identity of human Lyb-2 and CD72 and localization of the gene to chromosome 9. <i>European Journal of Immunology</i> , <b>1991</b> , 21, 1425-31	6.1	20
65	Gene for lymphoid enhancer-binding factor 1 (LEF1) mapped to human chromosome 4 (q23-q25) and mouse chromosome 3 near Egf. <i>Genomics</i> , <b>1991</b> , 11, 1040-8	4.3	51
64	Chromosomal mapping of brain-derived neurotrophic factor and neurotrophin-3 genes in man and mouse. <i>Genomics</i> , <b>1991</b> , 10, 569-75	4.3	35
63	Guidelines for human linkage maps. An International System for Human Linkage Maps (ISLM, 1990). <i>Annals of Human Genetics</i> , <b>1991</b> , 55, 1-6	2.2	26
62	Sequence analysis, expression and chromosomal localization of a gene, isolated from a subtracted human retina cDNA library, that encodes an insulin-like growth factor binding protein (IGFBP2). <i>Experimental Eye Research</i> , <b>1991</b> , 52, 549-61	3.7	28
61	Hereditary spherocytosis associated with deletion of human erythrocyte ankyrin gene on chromosome 8. <i>Nature</i> , <b>1990</b> , 345, 736-9	50.4	179
60	Assignment of Amog (adhesion molecule on glia) gene to mouse chromosome 11 near Zfp-3 and Asgr-1,2 and to human chromosome 17. <i>Somatic Cell and Molecular Genetics</i> , <b>1990</b> , 16, 401-5		10
59	cDNA cloning of human oxysterol-binding protein and localization of the gene to human chromosome 11 and mouse chromosome 19. <i>Genomics</i> , <b>1990</b> , 7, 65-74	4.3	72
58	Isolation, chromosomal mapping, and expression of the mouse tyrosinase gene. <i>Journal of Investigative Dermatology</i> , <b>1989</b> , 93, 589-94	4.3	50
57	Human SSAV-related endogenous retroviral element: LTR-like sequence and chromosomal localization to 18q21. <i>Genomics</i> , <b>1989</b> , 4, 68-75	4.3	32
56	Direct method for prenatal diagnosis and carrier detection in Duchenne/Becker muscular dystrophy using the entire dystrophin cDNA. <i>American Journal of Medical Genetics Part A</i> , <b>1988</b> , 29, 713-26		72
55	Molecular analysis of the cDNA for human SPARC/osteonectin/BM-40: sequence, expression, and localization of the gene to chromosome 5q31-q33. <i>Genomics</i> , <b>1988</b> , 2, 37-47	4.3	138
54	Human tyrosinase gene, mapped to chromosome 11 (q14----q21), defines second region of homology with mouse chromosome 7. <i>Genomics</i> , <b>1988</b> , 3, 17-24	4.3	150
53	Chromosomal mapping of the gene for the type II insulin-like growth factor receptor/cation-independent mannose 6-phosphate receptor in man and mouse. <i>Genomics</i> , <b>1988</b> , 3, 224-9	4.3	69
52	Cytogenetic and molecular genetic studies of a patient with atypical lymphoid hyperplasia. <i>Cancer Genetics and Cytogenetics</i> , <b>1987</b> , 27, 251-9		4



51	Restriction of human adenovirus replication in Chinese hamster cell lines and their hybrids with human cells. <i>Virus Research</i> , <b>1987</b> , 8, 277-99	6.4	15
50	Mammalian Cell. <i>BioScience</i> , <b>1987</b> , 37, 741-742	5.7	
49	The myelin-associated glycoprotein gene: mapping to human chromosome 19 and mouse chromosome 7 and expression in quivering mice. <i>Genomics</i> , <b>1987</b> , 1, 107-12	4.3	65
48	Mosaic tetrasomy 12p: four new cases, and confirmation of the chromosomal origin of the supernumerary chromosome in one of the original Pallister-Mosaic syndrome cases. <i>American Journal of Medical Genetics Part A</i> , <b>1987</b> , 27, 275-83		78
47	Molecular analysis and chromosomal mapping of amplified genes isolated from a transformed mouse 3T3 cell line. <i>Somatic Cell and Molecular Genetics</i> , <b>1987</b> , 13, 235-44		258
46	An intronless gene encoding a potential member of the family of receptors coupled to guanine nucleotide regulatory proteins. <i>Nature</i> , <b>1987</b> , 329, 75-9	50.4	489
45	A partial deletion of the muscular dystrophy gene transmitted twice by an unaffected male. <i>Nature</i> , <b>1987</b> , 329, 556-8	50.4	125
44	The human tyrosine aminotransferase gene mapped to the long arm of chromosome 16 (region 16q22----q24) by somatic cell hybrid analysis and in situ hybridization. <i>Human Genetics</i> , <b>1986</b> , 72, 221-4	6.3	41
43	Human luteinizing hormone-releasing hormone gene (LHRH) is located on short arm of chromosome 8 (region 8p11.2----p21). <i>Somatic Cell and Molecular Genetics</i> , <b>1986</b> , 12, 95-100		58
42	Transforming growth factor beta gene maps to human chromosome 19 long arm and to mouse chromosome 7. <i>Somatic Cell and Molecular Genetics</i> , <b>1986</b> , 12, 281-8		170
41	The involvement of 6p in melanoma. <i>Cancer Genetics and Cytogenetics</i> , <b>1986</b> , 20, 255-61		29
40	Direct assignment of orosomuroid to human chromosome 9 and alpha 2HS-glycoprotein to chromosome 3 using human fetal liver x rat hepatoma hybrids. <i>Human Genetics</i> , <b>1985</b> , 70, 109-15	6.3	27
39	High resolution cytogenetic evaluation of couples with recurring fetal wastage. <i>Human Genetics</i> , <b>1985</b> , 69, 246-9	6.3	8
38	The gene encoding the Ia-associated invariant chain is located on chromosome 18 in the mouse. <i>Immunogenetics</i> , <b>1985</b> , 21, 83-90	3.2	27
37	Human genes for U2 small nuclear RNA map to a major adenovirus 12 modification site on chromosome 17. <i>Nature</i> , <b>1985</b> , 314, 115-6	50.4	80
36	Minute chromosomes replacing the Y chromosome carry Y-specific sequences by restriction fragment analysis and in situ hybridization. <i>American Journal of Medical Genetics Part A</i> , <b>1985</b> , 22, 361-74		20
35	Comparative analysis of mouse-human hybrids with rearranged chromosomes 1 by in situ hybridization and Southern blotting: high-resolution mapping of NRAS, NGFB, and AMY on human chromosome 1. <i>Somatic Cell and Molecular Genetics</i> , <b>1984</b> , 10, 589-99		34
34	Human chromosomal mapping of genes for insulin-like growth factors I and II and epidermal growth factor. <i>Nature</i> , <b>1984</b> , 310, 781-4	50.4	234

33	Assignment of the gene for cystathionine beta-synthase to human chromosome 21 in somatic cell hybrids. <i>Human Genetics</i> , <b>1984</b> , 65, 291-4	6.3	68
32	Regional assignment of the human gene for platelet-type phosphofructokinase (PFKP) to chromosome 10p: novel use of polyspecific rodent antisera to localize human enzyme genes. <i>Human Genetics</i> , <b>1983</b> , 63, 374-9	6.3	41
31	Brief clinical report: aqueductal stenosis leading to hydrocephalus--an unusual manifestation of neurofibromatosis. <i>American Journal of Medical Genetics Part A</i> , <b>1983</b> , 14, 577-81		20
30	The N-ras oncogene assigned to the short arm of human chromosome 1. <i>Nucleic Acids Research</i> , <b>1983</b> , 11, 5267-75	20.1	34
29	The c-Ha-ras1, insulin and beta-globin loci map outside the deletion associated with aniridia-WilmsR tumour. <i>Nature</i> , <b>1983</b> , 305, 641-3	50.4	67
28	Assignment of the human gene for muscle-type phosphofructokinase (PFKM) to chromosome 1 (region cen leads to q32) using somatic cell hybrids and monoclonal anti-M antibody. <i>Somatic Cell Genetics</i> , <b>1982</b> , 8, 95-104		65
27	H-Y antigen expression in patients with X-autosomal translocations and gonadal dysgenesis. <i>American Journal of Medical Genetics Part A</i> , <b>1982</b> , 13, 115-23		9
26	The genes for growth hormone and chorionic somatomammotropin are on the long arm of human chromosome 17 in region q21 to qter. <i>Human Genetics</i> , <b>1981</b> , 57, 138-41	6.3	130
25	Requirement of the human chromosome 11 long arm for replication of herpes simplex virus type 1 in nonpermissive Chinese hamster x human diploid fibroblast hybrids. <i>Somatic Cell Genetics</i> , <b>1981</b> , 7, 171-91		33
24	Duplication 11p11.3 leads to 14.1 to meiotic crossing-over. <i>American Journal of Medical Genetics Part A</i> , <b>1980</b> , 7, 15-20		27
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22	The beta-globin gene is on the short arm of human chromosome 11. <i>Nature</i> , <b>1980</b> , 283, 683-6	50.4	41
21	Diffuse polyclonal B-cell lymphoma during primary infection with Epstein-Barr virus. <i>New England Journal of Medicine</i> , <b>1980</b> , 302, 1293-7	59.2	135
20	The aniridia-Wilms tumor association: The critical role of chromosome band 11p13. <i>Cancer Genetics and Cytogenetics</i> , <b>1980</b> , 2, 131-137		120
19	Chromosome assignment of a murine glucocorticoid receptor gene (Grl-1) using intraspecies somatic cell hybrids. <i>Cell</i> , <b>1980</b> , 22, 657-64	56.2	77
18	Partial triplication and deletion of 13q: study of a family presenting with bilateral retinoblastomas. <i>Clinical Genetics</i> , <b>1979</b> , 15, 332-45	4	62
17	Localisation of the G gamma-, A gamma-, delta- and beta-globin genes on the short arm of human chromosome 11. <i>Nature</i> , <b>1979</b> , 281, 606-8	50.4	51
16	Aniridia caused by a heritable chromosome 11 deletion. <i>Ophthalmology</i> , <b>1979</b> , 86, 1173-83	7.3	58

15	Utilization of purines by an HPRT variant in an intelligent, nonmutilative patient with features of the Lesch-Nyhan syndrome. <i>Pediatric Research</i> , <b>1979</b> , 13, 1365-70	3.2	43
14	Quantitative analysis of high-resolution trypsin-giemsa bands on human prometaphase chromosomes. <i>Human Genetics</i> , <b>1978</b> , 45, 137-65	6.3	235
13	Mouse chromosome 5 codes for ecotropic murine leukaemia virus cell-surface receptor. <i>Nature</i> , <b>1978</b> , 274, 60-2	50.4	53
12	Conservation of autosomal gene syntenic groups in mouse and man. <i>Nature</i> , <b>1978</b> , 274, 160-3	50.4	123
11	Regional mapping of human genes for hexosaminidase B and diphtheria toxin sensitivity on chromosome 5 using mouse X human hybrid cells. <i>Somatic Cell Genetics</i> , <b>1977</b> , 3, 629-38		24
10	Intrachromosomal gene mapping in man: the gene for tryptophyl-tRNA synthetase maps in region q21 leads to qter of chromosome 14. <i>Somatic Cell Genetics</i> , <b>1977</b> , 3, 381-9		12
9	Inverted tandem ("mirror") duplications in human chromosomes: -nv dup 8p, 4q, 22q. <i>American Journal of Medical Genetics Part A</i> , <b>1977</b> , 1, 3-19		88
8	Intrachromosomal gene mapping in man: assignment of nucleoside phosphorylase to region 14cen leads to 14q21 by interspecific hybridization of cells with a t(X;14) (p22;q21) translocation. <i>Somatic Cell Genetics</i> , <b>1976</b> , 2, 27-40		87
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3	Cytological identification of the chromosomes involved in Searle's translocation and the location of the centromere in the X chromosome of the mouse. <i>Genetics</i> , <b>1972</b> , 71, 643-8	4	18
2	Identification of the mouse chromosomes by quinacrine mustard staining. <i>Cytogenetic and Genome Research</i> , <b>1971</b> , 10, 356-66	1.9	30
1	Analysis of the T(3;?)6 Ca and T(14;17)264Ca translocations in the mouse by quinacrine mustard staining. <i>Genetics</i> , <b>1971</b> , 69, 517-22	4	14