Uta Francke

List of Publications by Citations

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	Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. *Nature Genetics*, **1999**, 23, 185-8	36.3	3780
175	Isolation of a novel gene mutated in Wiskott-Aldrich syndrome. *Cell*, **1994**, 78, 635-44	56.2	806
174	Wiskott-Aldrich syndrome protein, a novel effector for the GTPase CDC42Hs, is implicated in actin polymerization. *Cell*, **1996**, 84, 723-34	56.2	772
173	The CD40 ligand, gp39, is defective in activated T cells from patients with X-linked hyper-IgM syndrome. *Cell*, **1993**, 72, 291-300	56.2	706
172	NFAT dysregulation by increased dosage of DSCR1 and DYRK1A on chromosome 21. *Nature*, **2006**, 441, 595-600	50.4	534
171	An intronless gene encoding a potential member of the family of receptors coupled to guanine nucleotide regulatory proteins. *Nature*, **1987**, 329, 75-9	50.4	489
170	The gene for the peripheral myelin protein PMP-22 is a candidate for Charcot-Marie-Tooth disease type 1A. *Nature Genetics*, **1992**, 1, 159-65	36.3	469
169	Identification of a gene (GPR30) with homology to the G-protein-coupled receptor superfamily associated with estrogen receptor expression in breast cancer. *Genomics*, **1997**, 45, 607-17	4.3	419
168	Rett syndrome and beyond: recurrent spontaneous and familial MECP2 mutations at CpG hotspots. *American Journal of Human Genetics*, **1999**, 65, 1520-9	11	417
167	Trembler mouse carries a point mutation in a myelin gene. *Nature*, **1992**, 356, 241-4	50.4	400
166	Web-based genome-wide association study identifies two novel loci and a substantial genetic component for Parkinson's disease. *PLoS Genetics*, **2011**, 7, e1002141	6	387
165	A system of nomenclature for band patterns of mouse chromosomes. *Chromosoma*, **1973**, 41, 145-58	2.8	360
164	Molecular analysis and chromosomal mapping of amplified genes isolated from a transformed mouse 3T3 cell line. *Somatic Cell and Molecular Genetics*, **1987**, 13, 235-44		258
163	A mouse model for Prader-Willi syndrome imprinting-centre mutations. *Nature Genetics*, **1998**, 19, 25-31	36.3	255
162	Small nuclear ribonucleoprotein polypeptide N (SNRPN), an expressed gene in the Prader-Willi syndrome critical region. *Nature Genetics*, **1992**, 2, 265-9	36.3	242
161	Maternal imprinting of the mouse Snrpn gene and conserved linkage homology with the human Prader-Willi syndrome region. *Nature Genetics*, **1992**, 2, 259-64	36.3	238
160	Quantitative analysis of high-resolution trypsin-giemsa bands on human prometaphase chromosomes. *Human Genetics*, **1978**, 45, 137-65	6.3	235

159	Human chromosomal mapping of genes for insulin-like growth factors I and II and epidermal growth factor. *Nature*, **1984**, 310, 781-4	50.4	234
158	Identification of a novel paternally expressed gene in the Prader-Willi syndrome region. *Human Molecular Genetics*, **1994**, 3, 1877-82	5.6	229
157	SnoRNA Snord116 (Pwcr1/MBII-85) deletion causes growth deficiency and hyperphagia in mice. *PLoS ONE*, **2008**, 3, e1709	3.7	218
156	Human dystroglycan: skeletal muscle cDNA, genomic structure, origin of tissue specific isoforms and chromosomal localization. *Human Molecular Genetics*, **1993**, 2, 1651-7	5.6	213
155	Widespread changes in dendritic and axonal morphology in Mecp2-mutant mouse models of Rett syndrome: evidence for disruption of neuronal networks. *Journal of Comparative Neurology*, **2009**, 514, 240-58	3.4	194
154	Genome-wide analysis points to roles for extracellular matrix remodeling, the visual cycle, and neuronal development in myopia. *PLoS Genetics*, **2013**, 9, e1003299	6	190
153	Hereditary spherocytosis associated with deletion of human erythrocyte ankyrin gene on chromosome 8. *Nature*, **1990**, 345, 736-9	50.4	179
152	Transforming growth factor beta gene maps to human chromosome 19 long arm and to mouse chromosome 7. *Somatic Cell and Molecular Genetics*, **1986**, 12, 281-8		170
151	Cloning of MITF, the human homolog of the mouse microphthalmia gene and assignment to chromosome 3p14.1-p12.3. *Human Molecular Genetics*, **1994**, 3, 553-7	5.6	164
150	Human tyrosinase gene, mapped to chromosome 11 (q14----q21), defines second region of homology with mouse chromosome 7. *Genomics*, **1988**, 3, 17-24	4.3	150
149	Roberts syndrome: a review of 100 cases and a new rating system for severity. *American Journal of Medical Genetics Part A*, **1993**, 47, 1104-23		149
148	Silent mutation induces exon skipping of fibrillin-1 gene in Marfan syndrome. *Nature Genetics*, **1997** , 16, 328-9	36.3	147
147	Digitized and differentially shaded human chromosome ideograms for genomic applications. *Cytogenetic and Genome Research*, **1994**, 65, 206-18	1.9	144
146	p48 Activates a UV-damaged-DNA binding factor and is defective in xeroderma pigmentosum group E cells that lack binding activity. *Molecular and Cellular Biology*, **1998**, 18, 4391-9	4.8	142
145	NFATc3, a lymphoid-specific NFATc family member that is calcium-regulated and exhibits distinct DNA binding specificity. *Journal of Biological Chemistry*, **1995**, 270, 19898-907	5.4	142
144	A physical map, including a BAC/PAC clone contig, of the Williams-Beuren syndrome--deletion region at 7q11.23. *American Journal of Human Genetics*, **2000**, 66, 47-68	11	141
143	Molecular analysis of the cDNA for human SPARC/osteonectin/BM-40: sequence, expression, and localization of the gene to chromosome 5q31-q33. *Genomics*, **1988**, 2, 37-47	4.3	138
142	A novel human homologue of the Drosophila frizzled wnt receptor gene binds wingless protein and is in the Williams syndrome deletion at 7q11.23. *Human Molecular Genetics*, **1997**, 6, 465-72	5.6	135

141	Diffuse polyclonal B-cell lymphoma during primary infection with Epstein-Barr virus. *New England Journal of Medicine*, **1980**, 302, 1293-7	59.2	135
140	The genes for growth hormone and chorionic somatomammotropin are on the long arm of human chromosome 17 in region q21 to qter. *Human Genetics*, **1981**, 57, 138-41	6.3	130
139	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. *American Journal of Human Genetics*, **2000**, 67, 1067-82	11	125
138	A partial deletion of the muscular dystrophy gene transmitted twice by an unaffected male. *Nature*, **1987**, 329, 556-8	50.4	125
137	Conservation of autosomal gene synteny groups in mouse and man. *Nature*, **1978**, 274, 160-3	50.4	123
136	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. *Human Molecular Genetics*, **1998**, 7, 325-34	5.6	122
135	Guidelines for reporting clinical features in cases with MECP2 mutations. *Brain and Development*, **2001**, 23, 208-11	2.2	121
134	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. *Pediatrics*, **2009**, 123, 391-8	7.4	120
133	The aniridia-Wilms tumor association: The critical role of chromosome band 11p13. *Cancer Genetics and Cytogenetics*, **1980**, 2, 131-137		120
132	Premature termination mutations in FBN1: distinct effects on differential allelic expression and on protein and clinical phenotypes. *American Journal of Human Genetics*, **2002**, 71, 223-37	11	113
131	The TSG101 tumor susceptibility gene is located in chromosome 11 band p15 and is mutated in human breast cancer. *Cell*, **1997**, 88, 143-54	56.2	109
130	Inactivating mutations in ESCO2 cause SC phocomelia and Roberts syndrome: no phenotype-genotype correlation. *American Journal of Human Genetics*, **2005**, 77, 1117-28	11	108
129	Mutation creating a new splice site in the growth hormone receptor genes of 37 Ecuadorean patients with Laron syndrome. *Human Mutation*, **1992**, 1, 24-32	4.7	107
128	Novel associations for hypothyroidism include known autoimmune risk loci. *PLoS ONE*, **2012**, 7, e34442	3.7	105
127	Evidence for the role of PWCR1/HBII-85 C/D box small nucleolar RNAs in Prader-Willi syndrome. *American Journal of Human Genetics*, **2002**, 71, 669-78	11	104
126	Efficient replication of over 180 genetic associations with self-reported medical data. *PLoS ONE*, **2011**, 6, e23473	3.7	101
125	A new Rett syndrome family consistent with X-linked inheritance expands the X chromosome exclusion map. *American Journal of Human Genetics*, **1997**, 61, 634-41	11	100
124	Cardiovascular manifestations in men and women carrying a FBN1 mutation. *European Heart Journal*, **2010**, 31, 2223-9	9.5	98

123	Comparative study of brain morphology in Mecp2 mutant mouse models of Rett syndrome. *Journal of Comparative Neurology*, **2008**, 508, 184-95	3.4	98
122	Molecular evolution of the human interleukin-8 receptor gene cluster. *Nature Genetics*, **1992**, 2, 31-6	36.3	98
121	Cerebellar gene expression profiles of mouse models for Rett syndrome reveal novel MeCP2 targets. *BMC Medical Genetics*, **2007**, 8, 36	2.1	97
120	A severely affected male born into a Rett syndrome kindred supports X-linked inheritance and allows extension of the exclusion map. *American Journal of Human Genetics*, **1998**, 63, 267-9	11	95
119	RBM3, a novel human gene in Xp11.23 with a putative RNA-binding domain. *Human Molecular Genetics*, **1995**, 4, 2307-11	5.6	92
118	Mutation screening of complete fibrillin-1 coding sequence: report of five new mutations, including two in 8-cysteine domains. *Human Molecular Genetics*, **1993**, 2, 1813-21	5.6	91
117	Sporadic bilateral retinoblastoma and 13q- chromosomal deletion. *Medical and Pediatric Oncology*, **1976**, 2, 379-85		89
116	Inverted tandem ("mirror") duplications in human chromosomes: -nv dup 8p, 4q, 22q. *American Journal of Medical Genetics Part A*, **1977**, 1, 3-19		88
115	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. *Somatic Cell Genetics*, **1976**, 2, 27-40		87
114	Frizzled 9 knock-out mice have abnormal B-cell development. *Blood*, **2005**, 105, 2487-94	2.2	86
113	Control of bone formation by the serpentine receptor Frizzled-9. *Journal of Cell Biology*, **2011**, 192, 1057-72	7.3	84
112	Denaturing HPLC-identified novel FBN1 mutations, polymorphisms, and sequence variants in Marfan syndrome and related connective tissue disorders. *Genetic Testing and Molecular Biomarkers* , **1997**, 1, 237-42		82
111	An atypical deletion of the Williams-Beuren syndrome interval implicates genes associated with defective visuospatial processing and autism. *Journal of Medical Genetics*, **2007**, 44, 136-43	5.8	82
110	Human genes for U2 small nuclear RNA map to a major adenovirus 12 modification site on chromosome 17. *Nature*, **1985**, 314, 115-6	50.4	80
109	Gene expression patterns vary in clonal cell cultures from Rett syndrome females with eight different MECP2 mutations. *BMC Medical Genetics*, **2002**, 3, 12	2.1	79
108	Comparison of family history and SNPs for predicting risk of complex disease. *PLoS Genetics*, **2012**, 8, e1002973	6	78
107	A novel GC-rich human macrosatellite VNTR in Xq24 is differentially methylated on active and inactive X chromosomes. *Nature Genetics*, **1992**, 1, 137-43	36.3	78
106	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. *American Journal of Medical Genetics Part A*, **1987**, 27, 275-83		78

105	Chromosome assignment of a murine glucocorticoid receptor gene (Grl-1) using intraspecies somatic cell hybrids. *Cell*, **1980**, 22, 657-64	56.2	77
104	Characterization and expression pattern of the frizzled gene Fzd9, the mouse homolog of FZD9 which is deleted in Williams-Beuren syndrome. *Genomics*, **1999**, 57, 235-48	4.3	75
103	Induced chromosome deletions cause hypersociability and other features of Williams-Beuren syndrome in mice. *EMBO Molecular Medicine*, **2009**, 1, 50-65	12	74
102	Fibrillin abnormalities and prognosis in Marfan syndrome and related disorders. *American Journal of Medical Genetics Part A*, **1995**, 58, 169-76		73
101	cDNA cloning of human oxysterol-binding protein and localization of the gene to human chromosome 11 and mouse chromosome 19. *Genomics*, **1990**, 7, 65-74	4.3	72
100	Direct method for prenatal diagnosis and carrier detection in Duchenne/Becker muscular dystrophy using the entire dystrophin cDNA. *American Journal of Medical Genetics Part A*, **1988**, 29, 713-26		72
99	Mutant fibrillin-1 monomers lacking EGF-like domains disrupt microfibril assembly and cause severe marfan syndrome. *Human Molecular Genetics*, **1996**, 5, 1581-7	5.6	70
98	Lack of Pwcr1/MBII-85 snoRNA is critical for neonatal lethality in Prader-Willi syndrome mouse models. *Mammalian Genome*, **2005**, 16, 424-31	3.2	69
97	Chromosomal mapping of the gene for the type II insulin-like growth factor receptor/cation-independent mannose 6-phosphate receptor in man and mouse. *Genomics*, **1988**, 3, 224-9	4.3	69
96	Assignment of the gene for cystathionine beta-synthase to human chromosome 21 in somatic cell hybrids. *Human Genetics*, **1984**, 65, 291-4	6.3	68
95	The c-Ha-ras1, insulin and beta-globin loci map outside the deletion associated with aniridia-WilmsR tumour. *Nature*, **1983**, 305, 641-3	50.4	67
94	The myelin-associated glycoprotein gene: mapping to human chromosome 19 and mouse chromosome 7 and expression in quivering mice. *Genomics*, **1987**, 1, 107-12	4.3	65
93	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. *Somatic Cell Genetics*, **1982**, 8, 95-104		65
92	Serotonin receptor 1c gene assigned to X chromosome in human (band q24) and mouse (bands D-F4). *Human Molecular Genetics*, **1992**, 1, 681-4	5.6	63
91	Partial triplication and deletion of 13q: study of a family presenting with bilateral retinoblastomas. *Clinical Genetics*, **1979**, 15, 332-45	4	62
90	Skeletogenic phenotype of human Marfan embryonic stem cells faithfully phenocopied by patient-specific induced-pluripotent stem cells. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 215-20	11.5	60
89	Manifestations and linkage analysis in X-linked autoimmunity-immunodeficiency syndrome. *American Journal of Medical Genetics Part A*, **2000**, 90, 390-7		59
88	Novel mutations in the Wiskott-Aldrich syndrome protein gene and their effects on transcriptional, translational, and clinical phenotypes. *Human Mutation*, **1999**, 14, 54-66	4.7	58

87	Human luteinizing hormone-releasing hormone gene (LHRH) is located on short arm of chromosome 8 (region 8p11.2----p21). *Somatic Cell and Molecular Genetics*, **1986**, 12, 95-100		58
86	Aniridia caused by a heritable chromosome 11 deletion. *Ophthalmology*, **1979**, 86, 1173-83	7.3	58
85	MECP2 truncating mutations cause histone H4 hyperacetylation in Rett syndrome. *Human Molecular Genetics*, **2001**, 10, 1085-92	5.6	57
84	Dealing with the unexpected: consumer responses to direct-access BRCA mutation testing. *PeerJ*, **2013**, 1, e8	3.1	55
83	Identification of cis-regulatory elements for MECP2 expression. *Human Molecular Genetics*, **2006**, 15, 1769-82	5.6	55
82	In vivo nuclease hypersensitivity studies reveal multiple sites of parental origin-dependent differential chromatin conformation in the 150 kb SNRPN transcription unit. *Human Molecular Genetics*, **1999**, 8, 555-66	5.6	54
81	Mouse chromosome 5 codes for ecotropic murine leukaemia virus cell-surface receptor. *Nature*, **1978**, 274, 60-2	50.4	53
80	Gene for lymphoid enhancer-binding factor 1 (LEF1) mapped to human chromosome 4 (q23-q25) and mouse chromosome 3 near Egf. *Genomics*, **1991**, 11, 1040-8	4.3	51
79	Localisation of the G gamma-, A gamma-, delta- and beta-globin genes on the short arm of human chromosome 11. *Nature*, **1979**, 281, 606-8	50.4	51
78	Duplication 12q mosaicism in two unrelated patients with a similar syndrome. *American Journal of Medical Genetics Part A*, **1980**, 7, 123-9		51
77	Genetic variants associated with breast size also influence breast cancer risk. *BMC Medical Genetics*, **2012**, 13, 53	2.1	50
76	Isolation, chromosomal mapping, and expression of the mouse tyrosinase gene. *Journal of Investigative Dermatology*, **1989**, 93, 589-94	4.3	50
75	Structural, functional analysis and localization of the human CAP18 gene. *FEBS Letters*, **1996**, 398, 74-80	3.8	49
74	Prognosis factors in probands with an FBN1 mutation diagnosed before the age of 1 year. *Pediatric Research*, **2011**, 69, 265-70	3.2	48
73	Molecular breakpoint cloning and gene expression studies of a novel translocation t(4;15)(q27;q11.2) associated with Prader-Willi syndrome. *BMC Medical Genetics*, **2005**, 6, 18	2.1	48
72	Williams (Williams Beuren) syndrome: a distinct neurobehavioral disorder. *Journal of Child Neurology*, **2001**, 16, 177-90	2.5	47
71	Growth hormone receptor deficiency in Ecuador. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 4436-43	5.6	47
70	Mechanisms of disease: neurogenetics of MeCP2 deficiency. *Nature Clinical Practice Neurology*, **2006**, 2, 212-21		45

69	DLX5 and DLX6 expression is biallelic and not modulated by MeCP2 deficiency. *American Journal of Human Genetics*, **2007**, 81, 492-506	11	44
68	Four contiguous amino acid substitutions, identified in patients with Laron syndrome, differently affect the binding affinity and intracellular trafficking of the growth hormone receptor. *Journal of Clinical Endocrinology and Metabolism*, **1998**, 83, 4481-9	5.6	43
67	Utilization of purines by an HPRT variant in an intelligent, nonmutilative patient with features of the Lesch-Nyhan syndrome. *Pediatric Research*, **1979**, 13, 1365-70	3.2	43
66	Multi-exon deletions of the FBN1 gene in Marfan syndrome. *BMC Medical Genetics*, **2001**, 2, 11	2.1	42
65	Genes for the CPE receptor (CPETR1) and the human homolog of RVP1 (CPETR2) are localized within the Williams-Beuren syndrome deletion. *Genomics*, **1998**, 54, 453-9	4.3	41
64	The question of heterogeneity in Marfan syndrome. *Nature Genetics*, **1995**, 9, 228-31	36.3	41
63	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. *Human Genetics*, **1986**, 72, 221-4	6.3	41
62	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. *Human Genetics*, **1983**, 63, 374-9	6.3	41
61	The beta-globin gene is on the short arm of human chromosome 11. *Nature*, **1980**, 283, 683-6	50.4	41
60	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. *Genomics*, **1995**, 29, 471-7	4.3	40
59	A mouse single-copy gene, Gtf2i, the homolog of human GTF2I, that is duplicated in the Williams-Beuren syndrome deletion region. *Genomics*, **1998**, 48, 163-70	4.3	39
58	Diagnostic test for the Prader-Willi syndrome by SNRPN expression in blood. *Lancet, The*, **1996**, 348, 1068-9	40	36
57	Chromosomal mapping of brain-derived neurotrophic factor and neurotrophin-3 genes in man and mouse. *Genomics*, **1991**, 10, 569-75	4.3	35
56	The N-ras oncogene assigned to the short arm of human chromosome 1. *Nucleic Acids Research*, **1983**, 11, 5267-75	20.1	34
55	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. *Somatic Cell and Molecular Genetics*, **1984**, 10, 589-99		34
54	Spectrum of MECP2 mutations in Rett syndrome. *Brain and Development*, **2001**, 23 Suppl 1, S138-43	2.2	33
53	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. *Somatic Cell Genetics*, **1981**, 7, 171-91		33
52	Human SSAV-related endogenous retroviral element: LTR-like sequence and chromosomal localization to 18q21. *Genomics*, **1989**, 4, 68-75	4.3	32

51	A single mutation that results in an Asp to His substitution and partial exon skipping in a family with congenital contractural arachnodactyly. *Human Genetics*, **1998**, 103, 22-8	6.3	31
50	Identification of the mouse chromosomes by quinacrine mustard staining. *Cytogenetic and Genome Research*, **1971**, 10, 356-66	1.9	30
49	The involvement of 6p in melanoma. *Cancer Genetics and Cytogenetics*, **1986**, 20, 255-61		29
48	Partial trisomy for the distal long arm of chromosome 5 (region q34-qter). A new clinically recognizable syndrome. *Clinical Genetics*, **2008**, 15, 454-461	4	28
47	Fibrillin genes map to regions of conserved mouse/human synteny on mouse chromosomes 2 and 18. *Genomics*, **1993**, 18, 667-72	4.3	28
46	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). *Experimental Eye Research*, **1991**, 52, 549-61	3.7	28
45	frizzled 9 is expressed in neural precursor cells in the developing neural tube. *Development Genes and Evolution*, **2001**, 211, 453-7	1.8	27
44	Direct assignment of orosomucoid to human chromosome 9 and alpha 2HS-glycoprotein to chromosome 3 using human fetal liver x rat hepatoma hybrids. *Human Genetics*, **1985**, 70, 109-15	6.3	27
43	The gene encoding the Ia-associated invariant chain is located on chromosome 18 in the mouse. *Immunogenetics*, **1985**, 21, 83-90	3.2	27
42	Duplication 11p11.3 leads to 14.1 to meiotic crossing--over. *American Journal of Medical Genetics Part A*, **1980**, 7, 15-20		27
41	Reduction of NADPH-oxidase activity ameliorates the cardiovascular phenotype in a mouse model of Williams-Beuren Syndrome. *PLoS Genetics*, **2012**, 8, e1002458	6	26
40	Ube3a expression is not altered in Mecp2 mutant mice. *Human Molecular Genetics*, **2006**, 15, 2210-5	5.6	26
39	Conserved chromosomal location and genomic structure of human and mouse fatty-acid amide hydrolase genes and evaluation of clasper as a candidate neurological mutation. *Genomics*, **1998**, 54, 408-14	4.3	26
38	Guidelines for human linkage maps. An International System for Human Linkage Maps (ISLM, 1990). *Annals of Human Genetics*, **1991**, 55, 1-6	2.2	26
37	Partial trisomy 20p derived from a t(18;20) translocation. *Human Genetics*, **1976**, 34, 155-62	6.3	26
36	Diverse deletions in the growth hormone receptor gene cause growth hormone insensitivity syndrome. *Human Mutation*, **2000**, 16, 323-33	4.7	25
35	The mouse pink-eyed dilution gene: association with hypopigmentation in Prader-Willi and Angelman syndromes and with human OCA2. *Pigment Cell & Melanoma Research*, **1994**, 7, 398-402		24
34	Regional mapping of human genes for hexosaminidase B and diphtheria toxin sensitivity on chromosome 5 using mouse X human hybrid cells. *Somatic Cell Genetics*, **1977**, 3, 629-38		24

33	Mutation analysis and prenatal diagnosis in a Lesch-Nyhan family showing non-random X-inactivation interfering with carrier detection tests. *Human Genetics*, **1992**, 89, 395-400	6.3	23
32	Stature in Ecuadorians heterozygous for growth hormone receptor gene E180 splice mutation does not differ from that of homozygous normal relatives. *Journal of Clinical Endocrinology and Metabolism*, **1998**, 83, 2373-5	5.6	22
31	Sensitivity of Roberts syndrome cells to gamma radiation, mitomycin C, and protein synthesis inhibitors. *Somatic Cell and Molecular Genetics*, **1993**, 19, 377-92		22
30	Neonatal maternal deprivation response and developmental changes in gene expression revealed by hypothalamic gene expression profiling in mice. *PLoS ONE*, **2010**, 5, e9402	3.7	22
29	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. *Human Genetics*, **1999**, 104, 326-32	6.3	21
28	Identity of human Lyb-2 and CD72 and localization of the gene to chromosome 9. *European Journal of Immunology*, **1991**, 21, 1425-31	6.1	20
27	Brief clinical report: aqueductal stenosis leading to hydrocephalus--an unusual manifestation of neurofibromatosis. *American Journal of Medical Genetics Part A*, **1983**, 14, 577-81		20
26	Minute chromosomes replacing the Y chromosome carry Y-specific sequences by restriction fragment analysis and in situ hybridization. *American Journal of Medical Genetics Part A*, **1985**, 22, 361-74		20
25	Induced chromosome deletion in a Williams-Beuren syndrome mouse model causes cardiovascular abnormalities. *Journal of Vascular Research*, **2011**, 48, 119-29	1.9	18
24	Evaluation of two X chromosomal candidate genes for Rett syndrome: Glutamate dehydrogenase-2 (GLUD2) and rab GDP-dissociation inhibitor (GDI1). *American Journal of Medical Genetics Part A*, **1998**, 78, 169-172		18
23	Normal histone modifications on the inactive X chromosome in ICF and Rett syndrome cells: implications for methyl-CpG binding proteins. *BMC Biology*, **2004**, 2, 21	7.3	18
22	Cytological identification of the chromosomes involved in Searle's translocation and the location of the centromere in the X chromosome of the mouse. *Genetics*, **1972**, 71, 643-8	4	18
21	A Marfan syndrome gene expression phenotype in cultured skin fibroblasts. *BMC Genomics*, **2007**, 8, 319	4.5	17
20	Cloning and characterization of a novel zinc finger gene in Xp11.2. *Genomics*, **1995**, 30, 361-5	4.3	17
19	Restriction of human adenovirus replication in Chinese hamster cell lines and their hybrids with human cells. *Virus Research*, **1987**, 8, 277-99	6.4	15
18	Analysis of the T(3;?)6 Ca and T(14;17)264Ca translocations in the mouse by quinacrine mustard staining. *Genetics*, **1971**, 69, 517-22	4	14
17	Intrachromosomal gene mapping in man: the gene for tryptophyl-tRNA synthetase maps in region q21 leads to qter of chromosome 14. *Somatic Cell Genetics*, **1977**, 3, 381-9		12
16	Exclusion of the gastrin-releasing peptide receptor (GRPR) locus as a candidate gene for Rett syndrome. *American Journal of Medical Genetics Part A*, **1998**, 78, 173-5		11

15	Assignment of Amog (adhesion molecule on glia) gene to mouse chromosome 11 near Zfp-3 and Asgr-1,2 and to human chromosome 17. *Somatic Cell and Molecular Genetics*, **1990**, 16, 401-5		10
14	Properties of cystinotic fibroblast-D98 cell hybrids studied by somatic cell hybridization. *Nature*, **1973**, 244, 289-92	50.4	10
13	H-Y antigen expression in patients with X-autosomal translocations and gonadal dysgenesis. *American Journal of Medical Genetics Part A*, **1982**, 13, 115-23		9
12	Localization of the human HIP1 gene close to the elastin (ELN) locus on 7q11.23. *Genomics*, **1997**, 46, 313-5	4.3	8
11	Induced reciprocal translocation in transgenic mice near sites of transgene integration. *Mammalian Genome*, **1992**, 3, 209-16	3.2	8
10	High resolution cytogenetic evaluation of couples with recurring fetal wastage. *Human Genetics*, **1985**, 69, 246-9	6.3	8
9	Cytogenetic and molecular genetic studies of a patient with atypical lymphoid hyperplasia. *Cancer Genetics and Cytogenetics*, **1987**, 27, 251-9		4
8	Imprinted genes in the Prader-Willi deletion. *Novartis Foundation Symposium*, **1998**, 214, 264-75; discussion 275-9		3
7	2012 William Allan Award: Adventures in cytogenetics. *American Journal of Human Genetics*, **2013**, 92, 325-37	11	2
6	Novel mutations in the Wiskott-Aldrich syndrome protein gene and their effects on transcriptional, translational, and clinical phenotypes **1999**, 14, 54		2
5	Report of the Phenotype of a Patient with Roberts Syndrome and a Rare Variant. *Journal of Pediatric Genetics*, **2020**, 9, 58-62	0.7	1
4	Diverse deletions in the growth hormone receptor gene cause growth hormone insensitivity syndrome **2000**, 16, 323		1
3	Marfan Syndrome **2010**, 495-515		
2	Mammalian Cell. *BioScience*, **1987**, 37, 741-742	5.7	
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