Egbert Bakker

List of Publications by Year
in descending order

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135 papers	10,568 citations	30047 54 h-index	33869 99 g-index
139 all docs	139 docs citations	139 times ranked	10904 citing authors

#	Article	IF	Citations
1	Development of a comprehensive noninvasive prenatal test. Genetics and Molecular Biology, 2018, 41, 545-554.	0.6	8
2	Toward health technology assessment of whole-genome sequencing diagnostic tests: challenges and solutions. Personalized Medicine, 2017, 14, 235-247.	0.8	17
3	Critical points for an accurate human genome analysis. Human Mutation, 2017, 38, 912-921.	1.1	5
4	European registration process for Clinical Laboratory Geneticists in genetic healthcare. European Journal of Human Genetics, 2017, 25, 515-519.	1.4	13
5	A novel keratin 13 variant in a fourâ€generation family with white sponge nevus. Clinical Case Reports (discontinued), 2017, 5, 1503-1509.	0.2	12
6	Guidelines for diagnostic next-generation sequencing. European Journal of Human Genetics, 2016, 24, 2-5.	1.4	389
7	Novel Leptin Receptor Mutations Identified in Two Girls with Severe Obesity Are Associated with Increased Bone Mineral Density. Hormone Research in Paediatrics, 2016, 85, 412-420.	0.8	34
8	Broader Spectrum ofÎ²-Thalassemia Mutations in Oman: Regional Distribution and Comparison with Neighboring Countries. Hemoglobin, 2015, 39, 107-110.	0.4	5
9	Noninvasive prenatal diagnosis of Huntington disease: detection of the paternally inherited expanded CAG repeat in maternal plasma. Prenatal Diagnosis, 2015, 35, 945-949.	1.1	23
10	A Novel Targeted Approach for Noninvasive Detection of Paternally Inherited Mutations in Maternal Plasma. Journal of Molecular Diagnostics, 2015, 17, 590-596.	1.2	7
11	Hemizygosity forSMCHD1in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. Human Mutation, 2015, 36, 679-683.	1.1	32
12	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. Human Molecular Genetics, 2015, 24, 659-669.	1.4	130
13	Genetic Epidemiology and Preventive Healthcare in Multiethnic Societies: The Hemoglobinopathies. International Journal of Environmental Research and Public Health, 2014, 11, 6136-6146.	1.2	27
14	Molecular Spectrum ofÎ±-Globin Gene Defects in the Omani Population. Hemoglobin, 2014, 38, 422-426.	0.4	5
15	Primary Prevention of Hemoglobinopathies by Prenatal Diagnosis and Selective Pregnancy Termination in a Muslim Country: Oman. Thalassemia Reports, 2014, 4, 4171.	0.1	0
16	Population-based incidence and prevalence of facioscapulohumeral dystrophy. Neurology, 2014, 83, 1056-1059.	1.5	278
17	Known and New Î´-Globin Gene Mutations and Other Factors Influencing Hb A₂Measurement in the Omani Population. Hemoglobin, 2014, 38, 299-302.	0.4	5
18	The FSHD2 Gene SMCHD1 Is a Modifier of Disease Severity in Families Affected by FSHD1. American Journal of Human Genetics, 2013, 93, 744-751.	2.6	154

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19	Van Buchem disease: Clinical, biochemical, and densitometric features of patients and disease carriers. Journal of Bone and Mineral Research, 2013, 28, 848-854.	3.1	102
20	Successful Noninvasive Trisomy 18 Detection Using Single Molecule Sequencing. Clinical Chemistry, 2013, 59, 705-709.	1.5	11
21	A variant in FTO shows association with melanoma risk not due to BMI. Nature Genetics, 2013, 45, 428-432.	9.4	111
22	An urgent need for a change in policy revealed by a study on prenatal testing for Duchenne muscular dystrophy. European Journal of Human Genetics, 2013, 21, 21-26.	1.4	35
23	Mrassf1a-Pap, a Novel Methylation-Based Assay for the Detection of Cell-Free Fetal DNA in Maternal Plasma. PLoS ONE, 2013, 8, e84051.	1.1	9
24	Phenotypic characterization of patients with deletions in the 3â€™-flankingÂSHOXregion. PeerJ, 2013, 1, e35.	0.9	14
25	Single Molecule Sequencing of Free DNA from Maternal Plasma for Noninvasive Trisomy 21 Detection. Clinical Chemistry, 2012, 58, 699-706.	1.5	39
26	Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. Nature Genetics, 2012, 44, 1370-1374.	9.4	582
27	Nonâ€•invasive prenatal diagnosis of betaâ€†thalassemia and sickleâ€•cell disease using pyrophosphorolysisâ€•activated polymerization and melting curve analysis. Prenatal Diagnosis, 2012, 32, 578-587.	1.1	38
28	Fine-tiling array CGH to improve diagnostics for Î±- and Î²-thalassemia rearrangements. Human Mutation, 2012, 33, 272-280.	1.1	37
29	Candidate Gene-Based Association Study of Antipsychotic-Induced Movement Disorders in Long-Stay Psychiatric Patients: A Prospective Study. PLoS ONE, 2012, 7, e36561.	1.1	22
30	Genome-wide association study identifies novel loci predisposing to cutaneous melanomaâ€. Human Molecular Genetics, 2011, 20, 5012-5023.	1.4	187
31	Genome-wide association study identifies three new melanoma susceptibility loci. Nature Genetics, 2011, 43, 1108-1113.	9.4	230
32	Three new cases with a mosaicism involving a normal cell line and a cryptic unbalanced autosomal reciprocal translocation. European Journal of Medical Genetics, 2011, 54, e409-e412.	0.7	15
33	Experiences with array-based sequence capture; toward clinical applications. European Journal of Human Genetics, 2011, 19, 50-55.	1.4	13
34	The Jumping SHOX Geneâ€”Crossover in the Pseudoautosomal Region Resulting in Unusual Inheritance of Leri-Weill Dyschondrosteosis. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E356-E359.	1.8	14
35	Keratosis Follicularis Spinulosa Decalvans is caused by mutations in MBTPS2. Human Mutation, 2010, 31, 1125-1133.	1.1	67
36	Preâ€•and postsynaptic neuromuscular junction abnormalities in musk myasthenia. Muscle and Nerve, 2010, 42, 283-288.	1.0	53

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37	Melanocytic Nevi, Nevus Genes, and Melanoma Risk in a Large Case-Control Study in the United Kingdom. Cancer Epidemiology Biomarkers and Prevention, 2010, 19, 2043-2054.	1.1	102
38	Additional cryptic CNVs in mentally retarded patients with apparently balanced karyotypes. European Journal of Medical Genetics, 2010, 53, 227-233.	0.7	20
39	No Haploinsufficiency but Loss of Heterozygosity for EXT in Multiple Osteochondromas. American Journal of Pathology, 2010, 177, 1946-1957.	1.9	67
40	Emerging Technologies, Need for Quality Assessment. , 2010, , 333-340.		0
41	Multiple genomic aberrations in a patient with mental retardation and hypogonadism: 45,X/46,X,psu dic(Y) karyotype, thyroid hormone receptor beta (THRB) mutation and heterozygosity for Wilson disease. American Journal of Medical Genetics, Part A, 2009, 149A, 2231-2235.	0.7	2
42	Intronic variants inBRCA1andBRCA2that affect RNA splicing can be reliably selected by splice-site prediction programs. Human Mutation, 2009, 30, 107-114.	1.1	97
43	Diagnostic guidelines for high-resolution melting curve (HRM) analysis: An interlaboratory validation ofBRCA1mutation scanning using the 96-well LightScannerâ„¢. Human Mutation, 2009, 30, 899-909.	1.1	122
44	Multiple osteochondromas: mutation update and description of the multiple osteochondromas mutation database (MOdb). Human Mutation, 2009, 30, 1620-1627.	1.1	176
45	A new diagnostic workflow for patients with mental retardation and/or multiple congenital abnormalities: test arrays first. European Journal of Human Genetics, 2009, 17, 1394-1402.	1.4	70
46	Genome-wide association study identifies three loci associated with melanoma risk. Nature Genetics, 2009, 41, 920-925.	9.4	422
47	Rapid and cost effective detection of small mutations in the DMD gene by high resolution melting curve analysis. Neuromuscular Disorders, 2009, 19, 383-390.	0.3	33
48	Identification of copy number variants associated with BPES-like phenotypes. Human Genetics, 2008, 124, 489-498.	1.8	15
49	Genomeâ€wide linkage scan in Dutch hereditary nonâ€BRCA1/2 breast cancer families identifies 9q21â€22 as a putative breast cancer susceptibility locus. Genes Chromosomes and Cancer, 2008, 47, 947-956.	1.5	16
50	Search for copy number alterations in the MEFV gene using multiplex ligation probe amplification, experience from three diagnostic centres. European Journal of Human Genetics, 2008, 16, 1404-1406.	1.4	17
51	A 400kb duplication, 2.4Mb triplication and 130kbduplication of 9q34.3 in a patient with severe mental retardation. European Journal of Medical Genetics, 2008, 51, 479-487.	0.7	11
52	A novel (Leu183Pro-)mutation in the HFE-gene co-inherited with the Cys282Tyr mutation in two unrelated Dutch hemochromatosis patients. Blood Cells, Molecules, and Diseases, 2008, 40, 334-338.	0.6	10
53	Y chromosome detection by Real Time PCR and pyrophosphorolysis-activated polymerisation using free fetal DNA isolated from maternal plasma. Prenatal Diagnosis, 2007, 27, 932-937.	1.1	28
54	Ring chromosome formation as a novel escape mechanism in patients with inverted duplication and terminal deletion. European Journal of Human Genetics, 2007, 15, 548-555.	1.4	73

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55	A t(4;6)(q12;p23) translocation disrupts a membrane-associated O-acetyl transferase gene (MBOAT1) in a patient with a novel brachydactylyâ€“syndactyly syndrome. European Journal of Human Genetics, 2007, 15, 743-751.	1.4	25
56	Genotypeâ€“phenotype correlations in 19 Dutch cases with APC gene deletions and a literature review. European Journal of Human Genetics, 2007, 15, 1034-1042.	1.4	36
57	Molecular diagnostics of Meckelâ€“Gruber syndrome highlights phenotypic differences between MKS1 and MKS3. Human Genetics, 2007, 121, 591-599.	1.8	74
58	Haplotypes encoding the factor VIII 1241Glu variation, factor VIII levels and the risk of venous thrombosis. Thrombosis and Haemostasis, 2006, 95, 942-948.	1.8	17
59	Is the DNA Sequence the Gold Standard in Genetic Testing? Quality of Molecular Genetic Tests Assessed. Clinical Chemistry, 2006, 52, 557-558.	1.5	43
60	Mutation screening of EXT1 and EXT2 by direct sequence analysis and MLPA in patients with multiple osteochondromas: splice site mutations and exonic deletions account for more than half of the mutations. European Journal of Human Genetics, 2005, 13, 470-474.	1.4	49
61	Unclassified variants in disease-causing genes: nonuniformity of genetic testing and counselling, a proposal for guidelines. European Journal of Human Genetics, 2005, 13, 525-527.	1.4	25
62	Dystrophin analysis in carriers of Duchenne and Becker muscular dystrophy. Neurology, 2005, 65, 1984-1986.	1.5	36
63	Array-CGH detection of micro rearrangements in mentally retarded individuals: clinical significance of imbalances present both in affected children and normal parents. Journal of Medical Genetics, 2005, 43, 180-186.	1.5	190
64	Duchenne and Becker Muscular Dystrophy. , 2004, 92, 311-342.		3
65	Somatic mosaicism in FSHD often goes undetected. Annals of Neurology, 2004, 55, 845-850.	2.8	69
66	DGGE-based whole-gene mutation scanning of the dystrophin gene in Duchenne and Becker muscular dystrophy patients. Human Mutation, 2004, 23, 57-66.	1.1	50
67	Two-color multiplex ligation-dependent probe amplification: Detecting genomic rearrangements in hereditary multiple exostoses. Human Mutation, 2004, 24, 86-92.	1.1	142
68	Comprehensive Detection of Genomic Duplications and Deletions in the DMD Gene, by Use of Multiplex Amplifiable Probe Hybridization. American Journal of Human Genetics, 2002, 71, 365-374.	2.6	163
69	An Extensive Analysis of Y-Chromosomal Microsatellite Haplotypes in Globally Dispersed Human Populations. American Journal of Human Genetics, 2001, 68, 990-1018.	2.6	186
70	Molecular Analysis of Facioscapulohumeral Muscular Dystrophy (FSHD1). , 2001, , 305-316.		0
71	DNA-Based Techniques for Detection of Carriers of Duchenne and Becker Muscular Dystrophy. , 2001, , 111-135.		2
72	Complete allele information in the diagnosis of facioscapulohumeral muscular dystrophy by triple DNA analysis. Annals of Neurology, 2001, 50, 816-819.	2.8	91

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73	Dystrophin nonsense mutation induces different levels of exon 29 skipping and leads to variable phenotypes within one BMD family. European Journal of Human Genetics, 2000, 8, 793-796.	1.4	85
74	De Novo Facioscapulohumeral Muscular Dystrophy: Frequent Somatic Mosaicism, Sex-Dependent Phenotype, and the Role of Mitotic Transchromosomal Repeat Interaction between Chromosomes 4 and 10. American Journal of Human Genetics, 2000, 66, 26-35.	2.6	136
75	Clinical and Genetic Aspects of Hereditary Cerebral Hemorrhage with Amyloidosis Dutch Type (HCHW) Tj ETQq1 1 0.784314 rgBT /Ov		1
76	A European pilot quality assessment scheme for molecular diagnosis of Huntington's disease. European Journal of Human Genetics, 1999, 7, 217-222.	1.4	27
77	EXT-Mutation Analysis and Loss of Heterozygosity in Sporadic and Hereditary Osteochondromas and Secondary Chondrosarcomas. American Journal of Human Genetics, 1999, 65, 689-698.	2.6	174
78	A protein truncation test for Emeryâ€“Dreifuss muscular dystrophy (EMD): detection of N-terminal truncating mutations. Neuromuscular Disorders, 1999, 9, 247-250.	0.3	5
79	New possibilities for prenatal diagnosis of muscular dystrophies: forced myogenesis with an adenoviral MyoD-vector. Lancet, The, 1999, 353, 727-728.	6.3	17
80	Asn540Thr substitution in the fibroblast growth factor receptor 3 tyrosine kinase domain causing hypochondroplasia. Human Mutation, 1998, 11, S62-S65.	1.1	34
81	Mutations in the EXT1 and EXT2 Genes in Hereditary Multiple Exostoses. American Journal of Human Genetics, 1998, 62, 346-354.	2.6	174
82	Clinical Findings with Implications for Genetic Testing in Families with Clustering of Colorectal Cancer. New England Journal of Medicine, 1998, 339, 511-518.	13.9	386
83	Somatic Triple Mosaicism in a Carrier of X-Linked Chronic Granulomatous Disease. Blood, 1998, 91, 252-257.	0.6	11
84	Somatic Triple Mosaicism in a Carrier of X-Linked Chronic Granulomatous Disease. Blood, 1998, 91, 252-257.	0.6	0
85	Estimating Y Chromosome Specific Microsatellite Mutation Frequencies using Deep Rooting Pedigrees. Human Molecular Genetics, 1997, 6, 799-803.	1.4	234
86	Hereditary Nonpolyposis Colorectal Cancer Families Not Complying with the Amsterdam Criteria Show Extremely Low Frequency of Mismatch-Repair-Gene Mutations. American Journal of Human Genetics, 1997, 61, 329-335.	2.6	216
87	BRCA1 genomic deletions are major founder mutations in Dutch breast cancer patients. Nature Genetics, 1997, 17, 341-345.	9.4	414
88	Molecular genetic analysis of two families with keratosis follicularis spinulosa decalvans: refinement of gene localization and evidence for genetic heterogeneity. Human Genetics, 1997, 100, 520-524.	1.8	26
89	Evolution of cardiac abnormalities in Becker muscular dystrophy over a 13-year period. Journal of Neurology, 1997, 244, 657-663.	1.8	63
90	The Dutch uniform multicenter registration system for genetic disorders and malformation syndromes. , 1997, 70, 444-447.		6

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91	Location on the Human Genetic Linkage Map of 26 Genes Involved in Blood Coagulation. Thrombosis and Haemostasis, 1997, 77, 0873-0878.	1.8	20
92	Analysis of molecular variance (AMOVA) of Y-chromosome-specific microsatellites in two closely related human populations [published erratum appears in Hum Mol Genet 1997 May;6(5):828]. Human Molecular Genetics, 1996, 5, 1029-1033.	1.4	173
93	An Xp22.1-p22.2 YAC Contig Encompassing the Disease Loci for RS, KFSD, CLS, HYP and RP15: Refined Localization of RS. European Journal of Human Genetics, 1996, 4, 101-104.	1.4	19
94	The FSHD-linked locus D4F104S1 (p13E-11) ON 4q35 has a homologue on 10qter. Muscle and Nerve, 1995, 18, S39-S44.	1.0	106
95	Early onset facioscapulohumeral muscular dystrophy. Muscle and Nerve, 1995, 18, S67-S72.	1.0	53
96	Point mutation screening for 16 exons of the dystrophin gene by multiplex single-strand conformation polymorphism analysis. Human Mutation, 1995, 5, 235-242.	1.1	26
97	Rapid detection of BRCA1 mutations by the protein truncation test. Nature Genetics, 1995, 10, 208-212.	9.4	307
98	Confirmation of the 2p Locus for the Mild Autosomal Recessive Limb-Girdle Muscular Dystrophy Gene (LGMD2B) in Three Families Allows Refinement of the Candidate Region. Genomics, 1995, 27, 192-195.	1.3	41
99	Denaturing and non-denaturing gel electrophoresis as methods for the detection of junctional diversity in rearranged T cell receptor sequences. Journal of Immunological Methods, 1995, 181, 101-114.	0.6	12
100	The apolipoprotein E ?4 allele does not influence the clinical expression of the amyloid precursor protein gene codon 693 or 692 mutations. Annals of Neurology, 1994, 36, 434-437.	2.8	58
101	Evidence for the Absence of Intron H of the Histidine-Rich Glycoprotein (HRG) Gene: Genetic Mapping and in Situ Localization of HRG to Chromosome 3q28-q29. Genomics, 1994, 19, 195-197.	1.3	16
102	Severe nonspecific X-linked mental retardation caused by a proximally Xp located gene: Intragenic heterogeneity or a new form of X-linked mental retardation?. American Journal of Medical Genetics Part A, 1993, 46, 172-175.	2.4	24
103	Defining the Proximal Border of the Huntington Disease Candidate Region by Multipoint Recombination Analyses. Genomics, 1993, 16, 599-604.	1.3	3
104	Genetic heterogeneity for Duchenne-like muscular dystrophy (DLMD) based on linkage and 50 DAG analysis. Human Molecular Genetics, 1993, 2, 1945-1947.	1.4	50
105	No evidence of genetic heterogeneity in Brazilian facioscapulohumeral muscular dystrophy familes (FSHD) with 4q markers. Human Molecular Genetics, 1993, 2, 557-562.	1.4	23
106	Mapping around the Xq13.1 breakpoints of two X/A translocations in hypohidrotic ectodermal dysplasia (EDA) female patients. Genomics, 1992, 14, 523-525.	1.3	8
107	Parental origin and germline mosaicism of deletions and duplications of the dystrophin gene: a European study. Human Genetics, 1992, 88, 249-257.	1.8	100
108	Alzheimer's disease and hereditary cerebral hemorrhage with amyloidosis-dutch type share a decrease in cerebrospinal fluid levels of amyloid ?-protein precursor. Annals of Neurology, 1992, 32, 215-218.	2.8	29

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109	Presymptomatic, prenatal, and exclusion testing for Huntington disease using seven closely linked DNA markers. American Journal of Medical Genetics Part A, 1991, 39, 217-222.	2.4	20
110	An infrequent DNA polymorphism associated with severe von Willebrand's disease. British Journal of Haematology, 1990, 75, 78-81.	1.2	9
111	Somatic origin of inherited haemophilia A. Human Genetics, 1990, 85, 288-292.	1.8	50
112	An unusual variant of Becker muscular dystrophy. Annals of Neurology, 1990, 27, 578-581.	2.8	21
113	Two step procedure for early diagnosis of polycystic kidney disease with polymorphic DNA markers on both sides of the gene.. Journal of Medical Genetics, 1990, 27, 614-617.	1.5	20
114	Detection of Truncated Dystrophin in Fetal DMD Myotubes. Advances in Experimental Medicine and Biology, 1990, 280, 17-23.	0.8	2
115	High resoluation deletion breakpoint mapping in the DMD gene by whole cosmid hybridization. Nucleic Acids Research, 1989, 17, 5611-5621.	6.5	60
116	Germinal mosaicism increases the recurrence risk for 'new' Duchenne muscular dystrophy mutations.. Journal of Medical Genetics, 1989, 26, 553-559.	1.5	187
117	IMMUNOLOGICAL STUDY OF DYSTROPHIN IN DUCHENNE FETUS. Lancet, The, 1989, 334, 1212-1213.	6.3	27
118	The DMDgene analysed by field inversion gel electrophoresis. British Medical Bulletin, 1989, 45, 644-658.	2.7	19
119	First trimester prenatal diagnosis of haemophilia A: Two years' experience. Prenatal Diagnosis, 1988, 8, 411-421.	1.1	9
120	A deletion hot spot in the Duchenne muscular dystrophy gene. Genomics, 1988, 2, 101-108.	1.3	115
121	The fragile X syndrome in a large family. III. Investigations on linkage of flanking DNA markers with the fragile site Xq27.. Journal of Medical Genetics, 1987, 24, 413-421.	1.5	39
122	Two additional RFLPs at the D4S10 locus, useful for Huntington's disease (HD)-family studies. Nucleic Acids Research, 1987, 15, 9100-9100.	6.5	13
123	Long-range genomic map of the Duchenne muscular dystrophy (DMD) gene: Isolation and use of J66 (DXS268), a distal intragenic marker. Genomics, 1987, 1, 329-336.	1.3	71
124	Germline mosaicism and Duchenne muscular dystrophy mutations. Nature, 1987, 329, 554-556.	13.7	201
125	Direct detection of more than 50% of the Duchenne muscular dystrophy mutations by field inversion gels. Nature, 1987, 329, 640-642.	13.7	202
126	A physical map of 4 million bp around the Duchenne muscular dystrophy gene on the human X-chromosome. Cell, 1986, 47, 499-504.	13.5	155

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127	Two subsets of human alphoid repetitive DNA show distinct preferential localization in the pericentric regions of chromosomes 13, 18, and 21. Cytogenetic and Genome Research, 1986, 41, 193-201.	0.6	150
128	DNA probe analysis for carrier detection and prenatal diagnosis of Duchenne muscular dystrophy: a standard diagnostic procedure.. Journal of Medical Genetics, 1986, 23, 573-580.	1.5	56
129	Fine mapping of the Huntington disease linked D4S10 locus by non-radioactive in situ hybridization. Human Genetics, 1986, 73, 354-357.	1.8	63
130	Development of additional RFLP probes near the locus for Duchenne muscular dystrophy by cosmid cloning of the DXS84 (754) locus. Human Genetics, 1986, 74, 270-274.	1.8	37
131	Isolation of a random cosmid clone, cX5, which defines a new polymorphic locus DXS148 near the locus for Duchenne muscular dystrophy. Human Genetics, 1986, 74, 275-279.	1.8	19
132	Isolation of probes detecting restriction fragment length polymorphisms from X chromosome-specific libraries: potential use for diagnosis of Duchenne muscular dystrophy. Human Genetics, 1985, 70, 148-156.	1.8	222
133	An anonymous single copy chromosome 22 clone, D22S10 (22c1-18) identifies an RFLP with PstI. Nucleic Acids Research, 1985, 13, 7167-7167.	6.5	15
134	Zygosity determination in newborn twins using DNA variants.. Journal of Medical Genetics, 1985, 22, 279-282.	1.5	77
135	Localization of the polymorphic human calcitonin gene on chromosome 11. Human Genetics, 1984, 66, 309-312.	1.8	77