Egbert Bakker

List of Publications by Year in Descending Order

134 papers	9,051 citations	53 h-index	93 g-index
139 ext. papers	9,746 ext. citations	8.7 avg, IF	5 L-index

#	Paper	IF	Citations
134	Development of a comprehensive noninvasive prenatal test. *Genetics and Molecular Biology*, **2018**, 41, 545-554	2	7
133	Toward health technology assessment of whole-genome sequencing diagnostic tests: challenges and solutions. *Personalized Medicine*, **2017**, 14, 235-247	2.2	12
132	Critical points for an accurate human genome analysis. *Human Mutation*, **2017**, 38, 912-921	4.7	5
131	European registration process for Clinical Laboratory Geneticists in genetic healthcare. *European Journal of Human Genetics*, **2017**, 25, 515-519	5.3	9
130	A novel keratin 13 variant in a four-generation family with white sponge nevus. *Clinical Case Reports (discontinued)*, **2017**, 5, 1503-1509	0.7	7
129	Guidelines for diagnostic next-generation sequencing. *European Journal of Human Genetics*, **2016**, 24, 2-5	5.3	268
128	Novel Leptin Receptor Mutations Identified in Two Girls with Severe Obesity Are Associated with Increased Bone Mineral Density. *Hormone Research in Paediatrics*, **2016**, 85, 412-20	3.3	31
127	Broader spectrum of β-thalassemia mutations in Oman: regional distribution and comparison with neighboring countries. *Hemoglobin*, **2015**, 39, 107-10	0.6	2
126	Noninvasive prenatal diagnosis of Huntington disease: detection of the paternally inherited expanded CAG repeat in maternal plasma. *Prenatal Diagnosis*, **2015**, 35, 945-9	3.2	16
125	A Novel Targeted Approach for Noninvasive Detection of Paternally Inherited Mutations in Maternal Plasma. *Journal of Molecular Diagnostics*, **2015**, 17, 590-6	5.1	6
124	Hemizygosity for SMCHD1 in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. *Human Mutation*, **2015**, 36, 679-83	4.7	24
123	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. *Human Molecular Genetics*, **2015**, 24, 659-69	5.6	106
122	Population-based incidence and prevalence of facioscapulohumeral dystrophy. *Neurology*, **2014**, 83, 1056-9	6.9	203
121	Known and new β-globin gene mutations and other factors influencing Hb A2 measurement in the Omani population. *Hemoglobin*, **2014**, 38, 299-302	0.6	4
120	Genetic epidemiology and preventive healthcare in multiethnic societies: the hemoglobinopathies. *International Journal of Environmental Research and Public Health*, **2014**, 11, 6136-46	4.6	17
119	Molecular spectrum of β-globin gene defects in the Omani population. *Hemoglobin*, **2014**, 38, 422-6	0.6	5
118	The FSHD2 gene SMCHD1 is a modifier of disease severity in families affected by FSHD1. *American Journal of Human Genetics*, **2013**, 93, 744-51	11	126

117	Van Buchem disease: clinical, biochemical, and densitometric features of patients and disease carriers. *Journal of Bone and Mineral Research*, **2013**, 28, 848-54	6.3	89
116	Successful noninvasive trisomy 18 detection using single molecule sequencing. *Clinical Chemistry*, **2013**, 59, 705-9	5.5	9
115	A variant in FTO shows association with melanoma risk not due to BMI. *Nature Genetics*, **2013**, 45, 428-32, 432e1	36.3	95
114	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, 21-6	5.3	30
113	Mrassf1a-pap, a novel methylation-based assay for the detection of cell-free fetal DNA in maternal plasma. *PLoS ONE*, **2013**, 8, e84051	3.7	9
112	Phenotypic characterization of patients with deletions in the 3′ flanking SHOX region. *PeerJ*, **2013**, 1, e35	3.1	12
111	Fine-tiling array CGH to improve diagnostics for and thalassemia rearrangements. *Human Mutation*, **2012**, 33, 272-80	4.7	31
110	Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. *Nature Genetics*, **2012**, 44, 1370-4	36.3	405
109	Non-invasive prenatal diagnosis of beta-thalassemia and sickle-cell disease using pyrophosphorolysis-activated polymerization and melting curve analysis. *Prenatal Diagnosis*, **2012**, 32, 578-87	3.2	28
108	Single molecule sequencing of free DNA from maternal plasma for noninvasive trisomy 21 detection. *Clinical Chemistry*, **2012**, 58, 699-706	5.5	33
107	Candidate gene-based association study of antipsychotic-induced movement disorders in long-stay psychiatric patients: a prospective study. *PLoS ONE*, **2012**, 7, e36561	3.7	15
106	Genome-wide association study identifies three new melanoma susceptibility loci. *Nature Genetics*, **2011**, 43, 1108-13	36.3	203
105	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, e409-12	2.6	15
104	Experiences with array-based sequence capture; toward clinical applications. *European Journal of Human Genetics*, **2011**, 19, 50-5	5.3	12
103	Genome-wide association study identifies novel loci predisposing to cutaneous melanoma. *Human Molecular Genetics*, **2011**, 20, 5012-23	5.6	164
102	The jumping SHOX gene--crossover in the pseudoautosomal region resulting in unusual inheritance of Leri-Weill dyschondrosteosis. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, E356-9	5.6	14
101	Melanocytic nevi, nevus genes, and melanoma risk in a large case-control study in the United Kingdom. *Cancer Epidemiology Biomarkers and Prevention*, **2010**, 19, 2043-54	4	83
100	Additional cryptic CNVs in mentally retarded patients with apparently balanced karyotypes. *European Journal of Medical Genetics*, **2010**, 53, 227-33	2.6	19

99	No haploinsufficiency but loss of heterozygosity for EXT in multiple osteochondromas. *American Journal of Pathology*, **2010**, 177, 1946-57	5.8	59
98	Keratosis Follicularis Spinulosa Decalvans is caused by mutations in MBTPS2. *Human Mutation*, **2010**, 31, 1125-33	4.7	52
97	Pre- and postsynaptic neuromuscular junction abnormalities in musk myasthenia. *Muscle and Nerve*, **2010**, 42, 283-8	3.4	45
96	Emerging Technologies, Need for Quality Assessment **2010**, 333-340		
95	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, 2231-5	2.5	2
94	Intronic variants in BRCA1 and BRCA2 that affect RNA splicing can be reliably selected by splice-site prediction programs. *Human Mutation*, **2009**, 30, 107-14	4.7	88
93	Diagnostic guidelines for high-resolution melting curve (HRM) analysis: an interlaboratory validation of BRCA1 mutation scanning using the 96-well LightScanner. *Human Mutation*, **2009**, 30, 899-909	4.7	116
92	Multiple osteochondromas: mutation update and description of the multiple osteochondromas mutation database (MOdb). *Human Mutation*, **2009**, 30, 1620-7	4.7	154
91	A new diagnostic workflow for patients with mental retardation and/or multiple congenital abnormalities: test arrays first. *European Journal of Human Genetics*, **2009**, 17, 1394-402	5.3	63
90	Genome-wide association study identifies three loci associated with melanoma risk. *Nature Genetics*, **2009**, 41, 920-5	36.3	360
89	Rapid and cost effective detection of small mutations in the DMD gene by high resolution melting curve analysis. *Neuromuscular Disorders*, **2009**, 19, 383-90	2.9	28
88	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, 1404-6	5.3	16
87	A 400kb duplication, 2.4Mb triplication and 130kb duplication of 9q34.3 in a patient with severe mental retardation. *European Journal of Medical Genetics*, **2008**, 51, 479-87	2.6	11
86	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, 334-8	2.1	10
85	Identification of copy number variants associated with BPES-like phenotypes. *Human Genetics*, **2008**, 124, 489-98	6.3	14
84	Genome-wide linkage scan in Dutch hereditary non-BRCA1/2 breast cancer families identifies 9q21-22 as a putative breast cancer susceptibility locus. *Genes Chromosomes and Cancer*, **2008**, 47, 947-56	5	14
83	Y chromosome detection by Real Time PCR and pyrophosphorolysis-activated polymerisation using free fetal DNA isolated from maternal plasma. *Prenatal Diagnosis*, **2007**, 27, 932-7	3.2	24
82	Ring chromosome formation as a novel escape mechanism in patients with inverted duplication and terminal deletion. *European Journal of Human Genetics*, **2007**, 15, 548-55	5.3	63

81	A t(4;6)(q12;p23) translocation disrupts a membrane-associated O-acetyl transferase gene (MBOAT1) in a patient with a novel brachydactyly-syndactyly syndrome. *European Journal of Human Genetics*, **2007**, 15, 743-51	5.3	18
80	Genotype-phenotype correlations in 19 Dutch cases with APC gene deletions and a literature review. *European Journal of Human Genetics*, **2007**, 15, 1034-42	5.3	32
79	Molecular diagnostics of Meckel-Gruber syndrome highlights phenotypic differences between MKS1 and MKS3. *Human Genetics*, **2007**, 121, 591-9	6.3	62
78	Is the DNA sequence the gold standard in genetic testing? Quality of molecular genetic tests assessed. *Clinical Chemistry*, **2006**, 52, 557-8	5.5	38
77	Haplotypes encoding the factor VIII 1241 Glu variation, factor VIII levels and the risk of venous thrombosis. *Thrombosis and Haemostasis*, **2006**, 95, 942-8	7	15
76	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*, **2006**, 43, 180-6	5.8	161
75	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, 470-4	5.3	44
74	Unclassified variants in disease-causing genes: nonuniformity of genetic testing and counselling, a proposal for guidelines. *European Journal of Human Genetics*, **2005**, 13, 525-7	5.3	25
73	Dystrophin analysis in carriers of Duchenne and Becker muscular dystrophy. *Neurology*, **2005**, 65, 1984-6	6.5	31
72	Duchenne and Becker muscular dystrophy. *Methods in Molecular Medicine*, **2004**, 92, 311-41		
71	Somatic mosaicism in FSHD often goes undetected. *Annals of Neurology*, **2004**, 55, 845-50	9.4	53
70	DGGE-based whole-gene mutation scanning of the dystrophin gene in Duchenne and Becker muscular dystrophy patients. *Human Mutation*, **2004**, 23, 57-66	4.7	45
69	Two-color multiplex ligation-dependent probe amplification: detecting genomic rearrangements in hereditary multiple exostoses. *Human Mutation*, **2004**, 24, 86-92	4.7	136
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, 365-74	11	155
67	Molecular Analysis of Facioscapulohumeral Muscular Dystrophy (FSHD1) **2001**, 305-316		
66	DNA-Based Techniques for Detection of Carriers of Duchenne and Becker Muscular Dystrophy **2001** , 111-135		1
65	Complete allele information in the diagnosis of facioscapulohumeral muscular dystrophy by triple DNA analysis. *Annals of Neurology*, **2001**, 50, 816-9	9.4	80
64	An extensive analysis of Y-chromosomal microsatellite haplotypes in globally dispersed human populations. *American Journal of Human Genetics*, **2001**, 68, 990-1018	11	167

63	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, 793-6	5.3	75
62	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	11	118
61	Clinical and Genetic Aspects of Hereditary Cerebral Hemorrhage with Amyloidosis Dutch Type (HCHW A-D) **2000**, 103-119		1
60	A European pilot quality assessment scheme for molecular diagnosis of Huntington disease. *European Journal of Human Genetics*, **1999**, 7, 217-22	5.3	26
59	EXT-mutation analysis and loss of heterozygosity in sporadic and hereditary osteochondromas and secondary chondrosarcomas. *American Journal of Human Genetics*, **1999**, 65, 689-98	11	151
58	A protein truncation test for Emery-Dreifuss muscular dystrophy (EMD): detection of N-terminal truncating mutations. *Neuromuscular Disorders*, **1999**, 9, 247-50	2.9	4
57	New possibilities for prenatal diagnosis of muscular dystrophies: forced myogenesis with an adenoviral MyoD-vector. *Lancet, The*, **1999**, 353, 727-8	40	15
56	Asn540Thr substitution in the fibroblast growth factor receptor 3 tyrosine kinase domain causing hypochondroplasia. *Human Mutation*, **1998**, Suppl 1, S62-5	4.7	26
55	Mutations in the EXT1 and EXT2 genes in hereditary multiple exostoses. *American Journal of Human Genetics*, **1998**, 62, 346-54	11	149
54	Clinical findings with implications for genetic testing in families with clustering of colorectal cancer. *New England Journal of Medicine*, **1998**, 339, 511-8	59.2	346
53	Somatic Triple Mosaicism in a Carrier of X-Linked Chronic Granulomatous Disease. *Blood*, **1998**, 91, 252-257	9.2	10
52	Somatic Triple Mosaicism in a Carrier of X-Linked Chronic Granulomatous Disease. *Blood*, **1998**, 91, 252-257	9.2	
51	Estimating Y chromosome specific microsatellite mutation frequencies using deep rooting pedigrees. *Human Molecular Genetics*, **1997**, 6, 799-803	5.6	218
50	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, 329-35	11	204
49	BRCA1 genomic deletions are major founder mutations in Dutch breast cancer patients. *Nature Genetics*, **1997**, 17, 341-5	36.3	379
48	Molecular genetic analysis of two families with keratosis follicularis spinulosa decalvans: refinement of gene localization and evidence for genetic heterogeneity. *Human Genetics*, **1997**, 100, 520-4	6.3	23
47	Evolution of cardiac abnormalities in Becker muscular dystrophy over a 13-year period. *Journal of Neurology*, **1997**, 244, 657-63	5.5	55
46	The Dutch Uniform Multicenter Registration system for genetic disorders and malformation syndromes. *American Journal of Medical Genetics Part A*, **1997**, 70, 444-7		6

45	Location on the Human Genetic Linkage Map of 26 Genes Involved in Blood Coagulation. *Thrombosis and Haemostasis*, **1997**, 77, 0873-0878	7	13
44	Analysis of molecular variance (AMOVA) of Y-chromosome-specific microsatellites in two closely related human populations. *Human Molecular Genetics*, **1996**, 5, 1029-33	5.6	126
43	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, 101-4	5.3	17
42	Rapid detection of BRCA1 mutations by the protein truncation test. *Nature Genetics*, **1995**, 10, 208-12	36.3	283
41	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, 192-5	4.3	39
40	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, 101-14	2.5	12
39	The FSHD-linked locus D4F104S1 (p13E-11) ON 4q35 has a homologue on 10qter. *Muscle and Nerve*, **1995**, 18, S39-S44	3.4	79
38	Early onset facioscapulohumeral muscular dystrophy. *Muscle and Nerve*, **1995**, 18, S67-S72	3.4	44
37	Point mutation screening for 16 exons of the dystrophin gene by multiplex single-strand conformation polymorphism analysis. *Human Mutation*, **1995**, 5, 235-42	4.7	24
36	The apolipoprotein E epsilon 4 allele does not influence the clinical expression of the amyloid precursor protein gene codon 693 or 692 mutations. *Annals of Neurology*, **1994**, 36, 434-7	9.4	52
35	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, 195-7	4.3	14
34	Defining the proximal border of the Huntington disease candidate region by multipoint recombination analyses. *Genomics*, **1993**, 16, 599-604	4.3	2
33	Genetic heterogeneity for Duchenne-like muscular dystrophy (DLMD) based on linkage and 50 DAG analysis. *Human Molecular Genetics*, **1993**, 2, 1945-7	5.6	44
32	No evidence of genetic heterogeneity in Brazilian facioscapulohumeral muscular dystrophy families (FSHD) with 4q markers. *Human Molecular Genetics*, **1993**, 2, 557-62	5.6	20
31	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, 172-5		22
30	Mapping around the Xq13.1 breakpoints of two X/A translocations in hypohidrotic ectodermal dysplasia (EDA) female patients. *Genomics*, **1992**, 14, 523-5	4.3	8
29	Parental origin and germline mosaicism of deletions and duplications of the dystrophin gene: a European study. *Human Genetics*, **1992**, 88, 249-57	6.3	92
28	Alzheimer's disease and hereditary cerebral hemorrhage with amyloidosis-Dutch type share a decrease in cerebrospinal fluid levels of amyloid beta-protein precursor. *Annals of Neurology*, **1992**, 32, 215-8	9.4	27

27	Presymptomatic, prenatal, and exclusion testing for Huntington disease using seven closely linked DNA markers. *American Journal of Medical Genetics Part A*, **1991**, 39, 217-22		19
26	An infrequent DNA polymorphism associated with severe von Willebrand disease. *British Journal of Haematology*, **1990**, 75, 78-81	4.5	8
25	Somatic origin of inherited haemophilia A. *Human Genetics*, **1990**, 85, 288-92	6.3	49
24	An unusual variant of Becker muscular dystrophy. *Annals of Neurology*, **1990**, 27, 578-81	9.4	19
23	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, 614-7	5.8	18
22	Detection of truncated dystrophin in fetal DMD myotubes. *Advances in Experimental Medicine and Biology*, **1990**, 280, 17-23	3.6	2
21	High resolution deletion breakpoint mapping in the DMD gene by whole cosmid hybridization. *Nucleic Acids Research*, **1989**, 17, 5611-21	20.1	56
20	Germinal mosaicism increases the recurrence risk for "new" Duchenne muscular dystrophy mutations. *Journal of Medical Genetics*, **1989**, 26, 553-9	5.8	163
19	Immunological study of dystrophin in Duchenne fetus. *Lancet, The*, **1989**, 2, 1212-3	40	15
18	The DMD gene analysed by field inversion gel electrophoresis. *British Medical Bulletin*, **1989**, 45, 644-58	5.4	14
17	First trimester prenatal diagnosis of haemophilia A: two years' experience. *Prenatal Diagnosis*, **1988**, 8, 411-21	3.2	9
16	A deletion hot spot in the Duchenne muscular dystrophy gene. *Genomics*, **1988**, 2, 101-8	4.3	106
15	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, 413-21	5.8	38
14	Two additional RFLPs at the D4S10 locus, useful for Huntington's disease (HD)-family studies. *Nucleic Acids Research*, **1987**, 15, 9100	20.1	11
13	Long-range genomic map of the Duchenne muscular dystrophy (DMD) gene: isolation and use of J66 (DXS268), a distal intragenic marker. *Genomics*, **1987**, 1, 329-36	4.3	69
12	Germline mosaicism and Duchenne muscular dystrophy mutations. *Nature*, **1987**, 329, 554-6	50.4	187
11	Direct detection of more than 50% of the Duchenne muscular dystrophy mutations by field inversion gels. *Nature*, **1987**, 329, 640-2	50.4	191
10	DNA probe analysis for carrier detection and prenatal diagnosis of Duchenne muscular dystrophy: a standard diagnostic procedure. *Journal of Medical Genetics*, **1986**, 23, 573-80	5.8	53

9	Fine mapping of the Huntington disease linked D4S10 locus by non-radioactive in situ hybridization. *Human Genetics*, **1986**, 73, 354-7	6.3	57
8	Development of additional RFLP probes near the locus for Duchenne muscular dystrophy by cosmid cloning of the DXS84 (754) locus. *Human Genetics*, **1986**, 74, 270-4	6.3	36
7	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, 275-9	6.3	18
6	A physical map of 4 million bp around the Duchenne muscular dystrophy gene on the human X-chromosome. *Cell*, **1986**, 47, 499-504	56.2	142
5	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	1.9	142
4	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, 148-56	6.3	215
3	An anonymous single copy chromosome 22 clone, D22S10 (22c1-18) identifies an RFLP with PstI. *Nucleic Acids Research*, **1985**, 13, 7167	20.1	14
2	Zygosity determination in newborn twins using DNA variants. *Journal of Medical Genetics*, **1985**, 22, 279-82	3.8	63
1	Localization of the polymorphic human calcitonin gene on chromosome 11. *Human Genetics*, **1984**, 66, 309-12	6.3	76