Hans Scheffer

List of Publications by Year
in descending order

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199 papers	16,273 citations	20797 60 h-index	18115 120 g-index
203 all docs	203 docs citations	203 times ranked	23649 citing authors

#	Article	IF	Citations
1	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. Nature Genetics, 2014, 46, 989-993.	9.4	1,685
2	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. New England Journal of Medicine, 2012, 367, 1921-1929.	13.9	1,367
3	Imputation of sequence variants for identification of genetic risks for Parkinson's disease: a meta-analysis of genome-wide association studies. Lancet, The, 2011, 377, 641-649.	6.3	845
4	Guidelines for diagnostic next-generation sequencing. European Journal of Human Genetics, 2016, 24, 2-5.	1.4	389
5	POMT2 mutations cause Â-dystroglycan hypoglycosylation and Walker-Warburg syndrome. Journal of Medical Genetics, 2005, 42, 907-912.	1.5	374
6	Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. Brain, 2010, 133, 655-670.	3.7	356
7	Unbiased screen for interactors of leucine-rich repeat kinase 2 supports a common pathway for sporadic and familial Parkinson disease. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 2626-2631.	3.3	342
8	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. American Journal of Human Genetics, 2016, 98, 500-513.	2.6	333
9	Whole-genome sequencing in health care. European Journal of Human Genetics, 2013, 21, 580-584.	1.4	330
10	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	3.7	323
11	PCR-based DNA test to confirm clinical diagnosis of autosomal recessive spinal muscular atrophy. Lancet, The, 1995, 345, 985-986.	6.3	308
12	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. Human Mutation, 2013, 34, 1721-1726.	1.1	303
13	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	1.1	258
14	Geographic distribution and regional origin of 272 cystic fibrosis mutations in European populations. Human Mutation, 1997, 10, 135-154.	1.1	251
15	A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease. PLoS Genetics, 2011, 7, e1002142.	1.5	247
16	Revertant Mosaicism in Epidermolysis Bullosa Caused by Mitotic Gene Conversion. Cell, 1997, 88, 543-551.	13.5	245
17	Best practice guidelines for molecular genetic diagnosis of cystic fibrosis and CFTR-related disorders â€“ updated European recommendations. European Journal of Human Genetics, 2009, 17, 51-65.	1.4	207
18	Next-Generation Sequencing of a 40 Mb Linkage Interval Reveals TSPAN12 Mutations in Patients with Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 240-247.	2.6	202

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19	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	9.4	194
20	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	1.4	176
21	Identification of Patients With Variants in TPMT and Dose Reduction Reduces Hematologic Events During Thiopurine Treatment of Inflammatory Bowel Disease. Gastroenterology, 2015, 149, 907-917.e7.	0.6	169
22	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. Annals of Neurology, 2011, 70, 964-973.	2.8	168
23	Genome-wide association study confirms extant PD risk loci among the Dutch. European Journal of Human Genetics, 2011, 19, 655-661.	1.4	164
24	Association between angiotensin-converting-enzyme gene polymorphism and failure of renoprotective therapy. Lancet, The, 1996, 347, 94-95.	6.3	163
25	A Cystic Fibrosis Mutation Associated with Mild Lung Disease. New England Journal of Medicine, 1995, 333, 95-99.	13.9	162
26	A natural history study of late onset spinal muscular atrophy types 3b and 4. Journal of Neurology, 2008, 255, 1400-1404.	1.8	158
27	Mutations in BICD2, which Encodes a Golgin and Important Motor Adaptor, Cause Congenital Autosomal-Dominant Spinal Muscular Atrophy. American Journal of Human Genetics, 2013, 92, 946-954.	2.6	150
28	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	1.1	143
29	Seizure Control and Acceptance of the Ketogenic Diet in GLUT1 Deficiency Syndrome: A 2- to 5-Year Follow-Up of 15 Children Enrolled Prospectively. Neuropediatrics, 2005, 36, 302-308.	0.3	140
30	ARSACS in the Dutch population: a frequent cause of early-onset cerebellar ataxia. Neurogenetics, 2008, 9, 207-214.	0.7	132
31	Best practice guidelines and recommendations on the molecular diagnosis of myotonic dystrophy types 1 and 2. European Journal of Human Genetics, 2012, 20, 1203-1208.	1.4	129
32	Targeted Next-Generation Sequencing of a 12.5 Mb Homozygous Region Reveals ANO10 Mutations in Patients with Autosomal-Recessive Cerebellar Ataxia. American Journal of Human Genetics, 2010, 87, 813-819.	2.6	125
33	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2013, 22, 1039-1049.	1.4	122
34	Randomized sequential trial of valproic acid in amyotrophic lateral sclerosis. Annals of Neurology, 2009, 66, 227-234.	2.8	111
35	<scp>RYR</scp>1â€related myopathies: a wide spectrum of phenotypes throughout life. European Journal of Neurology, 2015, 22, 1094-1112.	1.7	111
36	Genotypeâ€“phenotype correlations in spastic paraplegia type 7: a study in a large Dutch cohort. Brain, 2012, 135, 2994-3004.	3.7	107

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37	The expanding phenotype ofPOMT1mutations: from Walker-Warburg syndrome to congenital muscular dystrophy, microcephaly, and mental retardation. Human Mutation, 2006, 27, 453-459.	1.1	106
38	Cerebrospinal Fluid Analysis in the Workup of GLUT1 Deficiency Syndrome. JAMA Neurology, 2013, 70, 1440.	4.5	106
39	Diagnostic exome sequencing in 266 Dutch patients with visual impairment. European Journal of Human Genetics, 2017, 25, 591-599.	1.4	104
40	Effects of Keratin 14 Ablation on the Clinical and Cellular Phenotype in a Kindred with Recessive Epidermolysis Bullosa Simplex. Journal of Investigative Dermatology, 1996, 107, 764-769.	0.3	103
41	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
42	SMN genotypes producing less SMN protein increase susceptibility to and severity of sporadic ALS. Neurology, 2005, 65, 820-825.	1.5	94
43	Genome-wide association analysis of anti-TNF drug response in patients with rheumatoid arthritis. Annals of the Rheumatic Diseases, 2013, 72, 1375-1381.	0.5	94
44	Prenatal diagnostic testing of the Noonan syndrome genes in fetuses with abnormal ultrasound findings. European Journal of Human Genetics, 2013, 21, 936-942.	1.4	94
45	The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands. European Journal of Human Genetics, 2017, 25, 308-314.	1.4	90
46	Clinical exome sequencing for cerebellar ataxia and spastic paraplegia uncovers novel geneâ€“disease associations and unanticipated rare disorders. European Journal of Human Genetics, 2016, 24, 1460-1466.	1.4	89
47	Allele sharing on chromosome 11q13 in sibs with asthma and atopy. Lancet, The, 1993, 342, 936.	6.3	86
48	Massively parallel sequencing of ataxia genes after array-based enrichment. Human Mutation, 2010, 31, 494-499.	1.1	86
49	Mutations in C8orf37, Encoding a Ciliary Protein, are Associated with Autosomal-Recessive Retinal Dystrophies with Early Macular Involvement. American Journal of Human Genetics, 2012, 90, 102-109.	2.6	82
50	Paraplegin mutations in sporadic adult-onset upper motor neuron syndromes. Neurology, 2008, 71, 1500-1505.	1.5	81
51	Association of the dopamine transporter (SLC6A3/DAT1) gene 9â€“6 haplotype with adult ADHD. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1576-1579.	1.1	78
52	Persistent failures in gene repair. Nature Biotechnology, 2001, 19, 305-306.	9.4	77
53	The Association Between HTR2C Gene Polymorphisms and the Metabolic Syndrome in Patients With Schizophrenia. Journal of Clinical Psychopharmacology, 2007, 27, 338-343.	0.7	76
54	HTR2C Gene Polymorphisms and the Metabolic Syndrome in Patients With Schizophrenia. Journal of Clinical Psychopharmacology, 2009, 29, 16-20.	0.7	74

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55	Genotype-phenotype correlations in MYCN-related Feingold syndrome. Human Mutation, 2008, 29, 1125-1132.	1.1	72
56	Clinical and Molecular Evaluation of Probands and Family Members with Familial Exudative Vitreoretinopathy. , 2009, 50, 4379.		68
57	Hereditary myokymia and paroxysmal ataxia linked to chromosome 12 is responsive to acetazolamide.. Journal of Neurology, Neurosurgery and Psychiatry, 1995, 59, 400-405.	0.9	67
58	Two cases of maternal uniparental disomy 14 with a phenotype overlapping with the Prader-Willi phenotype. , 1999, 84, 76-79.		64
59	Best practice guidelines for molecular analysis in spinal muscular atrophy. European Journal of Human Genetics, 2001, 9, 484-491.	1.4	63
60	Glycogen storage disease type Ia: recent experience with mutation analysis, a summary of mutations reported in the literature and a newly developed diagnostic flowchart. European Journal of Pediatrics, 2000, 159, 322-330.	1.3	62
61	An association study of 45 folateâ€related genes in spina bifida: Involvement of cubilin (CUBN) and tRNA aspartic acid methyltransferase 1 (TRDMT1). Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 216-226.	1.6	62
62	Partial Revertant Mosaicism of Keratin 14 in a Patient with Recessive Epidermolysis Bullosa Simplex11Part of this paper was presented at the 2nd Scientific Meeting of the Dutch Society for Experimental Dermatology, Lunteren, the Netherlands, February 8, 2001 and on the 30th Annual Meeting of the European Society for Dermatological Research, Berlin, September 22, 2000.. Journal of Investigative Dermatology, 2002, 118, 626-630.	0.3	61
63	Validation Study of Existing Gene Expression Signatures for Anti-TNF Treatment in Patients with Rheumatoid Arthritis. PLoS ONE, 2012, 7, e33199.	1.1	61
64	Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome. European Journal of Human Genetics, 2015, 23, 317-324.	1.4	61
65	242 Breakpoints in the 200-kb deletion-prone P20 region of the DMD gene are widely spread. Genomics, 1991, 10, 631-639.	1.3	60
66	Maternal uniparental disomy for chromosome 14 in a boy with a normal karyotype. Journal of Medical Genetics, 1999, 36, 782-785.	1.5	60
67	SMA carrier testing â€“ validation of hemizygous SMN exon 7 deletion test for the identification of proximal spinal muscular atrophy carriers and patients with a single allele deletion. European Journal of Human Genetics, 2000, 8, 79-86.	1.4	59
68	Autosomal Recessive Inheritance of GLUT1 Deficiency Syndrome. Neuropediatrics, 2009, 40, 207-210.	0.3	57
69	Autosomal Recessive Cerebellar Ataxia Type 3 Due toANO10Mutations. JAMA Neurology, 2014, 71, 1305.	4.5	57
70	Genetic comorbidities in Parkinson's disease. Human Molecular Genetics, 2014, 23, 831-841.	1.4	57
71	Mutation analysis of the entire keratin 5 and 14 genes in patients with epidermolysis bullosa simplex and identification of novel mutations. Human Mutation, 2003, 21, 447-447.	1.1	56
72	Three novel KCNA1 mutations in episodic ataxia type I families. Human Genetics, 1998, 102, 464-466.	1.8	54

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73	A novel translation re-initiation mechanism for the p63 gene revealed by amino-terminal truncating mutations in Rapp-Hodgkin/Hay-Wells-like syndromes. Human Molecular Genetics, 2008, 17, 1968-1977.	1.4	53
74	Molecular evidence that AEC syndrome and Rapp-Hodgkin syndrome are variable expression of a single genetic disorder. Clinical Genetics, 2004, 66, 79-80.	1.0	52
75	Early Assessment of Thiopurine Metabolites Identifies Patients at Risk of Thiopurine-induced Leukopenia in Inflammatory Bowel Disease. Journal of Crohn's and Colitis, 2017, 11, 175-184.	0.6	52
76	Recommendations for quality improvement in genetic testing for cystic fibrosis European Concerted Action on Cystic Fibrosis. European Journal of Human Genetics, 2000, 8, S2-S12.	1.4	51
77	Cerebellar ataxia and congenital disorder of glycosylation Ia (CDG-Ia) with normal routine CDG screening. Journal of Neurology, 2007, 254, 1356-1358.	1.8	50
78	Spectrum of p63 mutations in a selected patient cohort affected with ankyloblepharonâ€ectodermal defectsâ€cleft lip/palate syndrome (AEC). American Journal of Medical Genetics, Part A, 2009, 149A, 1948-1951.	0.7	49
79	Human Uteroglobin Gene: Structure, Subchromosomal Localization, and Polymorphism. DNA and Cell Biology, 1997, 16, 73-83.	0.9	48
80	Autosomal recessive oculopharyngodistal myopathy: a distinct phenotypical, histological, and genetic entity. Journal of Neurology, Neurosurgery and Psychiatry, 2004, 75, 1499-1501.	0.9	48
81	Pharmacogenetics of anti-TNF treatment in patients with rheumatoid arthritis. Pharmacogenomics, 2007, 8, 761-773.	0.6	48
82	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	1.4	48
83	Spastin mutations in sporadic adult-onset upper motor neuron syndromes. Annals of Neurology, 2005, 58, 865-869.	2.8	47
84	The association between HTR2C polymorphisms and obesity in psychiatric patients using antipsychotics: a cross-sectional study. Pharmacogenomics Journal, 2007, 7, 318-324.	0.9	46
85	Mutation Frequency of Cystic Fibrosis Transmembrane Regulator is not Increased in Oligozoospermic Male Candidates For Intracytoplasmic Sperm Injection. Fertility and Sterility, 1998, 69, 899-903.	0.5	45
86	Hereditary spastic paraplegia due to SPAST mutations in 151 Dutch patients: new clinical aspects and 27 novel mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 1073-1078.	0.9	45
87	The clinical and molecular genetic approach to Duchenne and Becker muscular dystrophy: an updated protocol.. Journal of Medical Genetics, 1997, 34, 805-812.	1.5	44
88	The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 666-673.	0.9	43
89	Mechanisms of Natural Gene Therapy in Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2014, 134, 2097-2104.	0.3	40
90	Autosomal recessive cerebellar ataxias: the current state of affairs. Journal of Medical Genetics, 2011, 48, 651-659.	1.5	39

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91	Partial 3q duplication syndrome and assignment of D3S5 to 3q25â€“3q28. Human Genetics, 1991, 87, 151-154.	1.8	38
92	The tumour necrosis factor receptor superfamily member 1b 676T>G polymorphism in relation to response to infliximab and adalimumab treatment and disease severity in rheumatoid arthritis. Annals of the Rheumatic Diseases, 2008, 67, 1174-1177.	0.5	38
93	A homozygous FKRP start codon mutation is associated with Walkerâ€“Warburg syndrome, the severe end of the clinical spectrum. Clinical Genetics, 2010, 78, 275-281.	1.0	38
94	Complete association of loss of heterozygosity of chromosomes 13 and 17 in osteosarcoma. Cancer Genetics and Cytogenetics, 1991, 53, 45-55.	1.0	37
95	Pure adult-onset Spastic Paraplegia caused by a novel mutation in the KIAA0196 (SPG8) gene. Journal of Neurology, 2013, 260, 1765-1769.	1.8	37
96	Stepwise ABC system for classification of any type of genetic variant. European Journal of Human Genetics, 2022, 30, 150-159.	1.4	37
97	The inversa type of recessive dystrophic epidermolysis bullosa is caused by specific arginine and glycine substitutions in type VII collagen. Journal of Medical Genetics, 2011, 48, 160-167.	1.5	35
98	Early prediction of thiopurineâ€induced hepatotoxicity in inflammatory bowel disease. Alimentary Pharmacology and Therapeutics, 2017, 45, 391-402.	1.9	35
99	Exempting Homologous Pseudogene Sequences from Polymerase Chain Reaction Amplification Allows Genomic Keratin 14 Hotspot Mutation Analysis. Journal of Investigative Dermatology, 2000, 114, 616-619.	0.3	34
100	Validation of the determination of Î”F508 mutations of the cystic fibrosis gene in over 11000 mouthwashes. Human Genetics, 1996, 97, 334-336.	1.8	33
101	Gene expression profiling in rheumatoid arthritis: current concepts and future directions. Annals of the Rheumatic Diseases, 2008, 67, 1663-1669.	0.5	33
102	Haplotype identity between individuals who share a CFTR mutation allele "identical by descent": demonstration of the usefulness of the haplotype-sharing concept for gene mapping in real populations. Human Genetics, 1996, 98, 304-309.	1.8	32
103	Comprehensive and accurate mutation scanning of theCFTR gene by two-dimensional DNA electrophoresis. Human Mutation, 1996, 8, 160-167.	1.1	31
104	Risk factors for thiopurineâ€induced myelosuppression and infections in inflammatory bowel disease patients with a normal <scp>TPMT</scp> genotype. Alimentary Pharmacology and Therapeutics, 2017, 46, 953-963.	1.9	31
105	Mutational scanning of large genes by extensive PCR multiplexing and two-dimensional electrophoresis: application to the RB1 gene. Human Molecular Genetics, 1996, 5, 755-761.	1.4	30
106	GLUT1 Deficiency With Delayed Myelination Responding to Ketogenic Diet. Pediatric Neurology, 2007, 37, 130-133.	1.0	30
107	MR Spectroscopy and Serial Magnetic Resonance Imaging in a Patient with Mitochondrial Cystic Leukoencephalopathy due to Complex I Deficiency andNDUFV1Mutations and Mild Clinical Course. Neuropediatrics, 2008, 39, 172-175.	0.3	30
108	BSCL2 mutations in two Dutch families with overlapping Silver syndrome-distal hereditary motor neuropathy. Neuromuscular Disorders, 2006, 16, 122-125.	0.3	29

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109	Molecular Diagnosis of Usher Syndrome: Application of Two Different Next Generation Sequencing-Based Procedures. PLoS ONE, 2012, 7, e43799.	1.1	29
110	Occurrence of deletion of a COL2A1 allele as the mutation in Stickler syndrome shows that a collagen type II dosage effect underlies this syndrome. Human Mutation, 2002, 20, 236-236.	1.1	28
111	Susceptibility loci for pigmentation and melanoma in relation to Parkinson's disease. Neurobiology of Aging, 2014, 35, 1512.e5-1512.e10.	1.5	28
112	A novel SLC2A1 mutation linking hemiplegic migraine with alternating hemiplegia of childhood. Cephalalgia, 2015, 35, 10-15.	1.8	28
113	Two novel germline mutations of the retinoblastoma gene (RB1) that show incomplete penetrance, one splice site and one missense. Journal of Medical Genetics, 2000, 37, 6e-6.	1.5	27
114	Long-term follow-up of patients with recessive dystrophic epidermolysis bullosa in the Netherlands: Expansion of the mutation database and unusual phenotypeâ€“genotype correlations. Journal of Dermatological Science, 2009, 56, 9-18.	1.0	27
115	A BRCA1 founder mutation, identified with haplotype analysis, allowing genotype/phenotype determination and predictive testing. European Journal of Cancer, 1997, 33, 2390-2392.	1.3	26
116	Natural Exon Skipping Sets the Stage for Exon Skipping as Therapy for Dystrophic Epidermolysis Bullosa. Molecular Therapy - Nucleic Acids, 2019, 18, 465-475.	2.3	26
117	A straightforward approach to isolate DNA sequences with potential linkage to the retinoblastoma locus. Human Genetics, 1986, 74, 249-255.	1.8	25
118	Translating Sanger-Based Routine DNA Diagnostics into Generic Massive Parallel Ion Semiconductor Sequencing. Clinical Chemistry, 2015, 61, 154-162.	1.5	25
119	A Provisional Transcript Map of the Spinal Muscular Atrophy (SMA) Critical Region. European Journal of Human Genetics, 1995, 3, 87-95.	1.4	25
120	Prevalence of carriers of the most common medium-chain acyl-CoA dehydrogenase (MCAD) deficiency mutation (G985A) in The Netherlands. Human Genetics, 1996, 98, 1-2.	1.8	24
121	Genetic Variants in Toll-Like Receptors Are Not Associated with Rheumatoid Arthritis Susceptibility or Anti-Tumour Necrosis Factor Treatment Outcome. PLoS ONE, 2010, 5, e14326.	1.1	24
122	Homozygous acute intermittent porphyria in a 7-year-old boy with massive excretions of porphyrins and porphyrin precursors. Journal of Inherited Metabolic Disease, 2004, 27, 19-27.	1.7	23
123	Clinical exome sequencing in daily practice: 1,000 patients and beyond. Genome Medicine, 2014, 6, 2.	3.6	23
124	More Dose-dependent Side Effects with Mercaptopurine over Azathioprine in IBD Treatment Due to Relatively Higher Dosing. Inflammatory Bowel Diseases, 2017, 23, 1873-1881.	0.9	23
125	Rapidly deteriorating course in Dutch hereditary spastic paraplegia type 11 patients. European Journal of Human Genetics, 2013, 21, 1312-1315.	1.4	22
126	Diagnostic Exome Sequencing in Persons With Severe Intellectual Disability. Obstetrical and Gynecological Survey, 2013, 68, 191-193.	0.2	22

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127	Prenatal diagnosis in a family with X-linked chronic granulomatous disease with the use of the polymerase chain reaction. Prenatal Diagnosis, 1992, 12, 773-777.	1.1	21
128	Expanding the clinical spectrum ofMYCN-related Feingold syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2254-2256.	0.7	21
129	Consanguinity sans reproche. Human Genetics, 1991, 86, 295-296.	1.8	19
130	Infantile spinal muscular atrophy variant with congenital fractures in a female neonate: evidence for autosomal recessive inheritance. Journal of Medical Genetics, 2002, 39, 74-77.	1.5	19
131	Meta-analysis identified the TNFA -308G > A promoter polymorphism as a risk factor for disease severity in patients with rheumatoid arthritis. Arthritis Research and Therapy, 2012, 14, R264.	1.6	19
132	Noonan Syndrome: Comparing Mutation-Positive with Mutation-Negative Dutch Patients. Molecular Syndromology, 2013, 4, 227-234.	0.3	19
133	Genotype-Guided Thiopurine Dosing Does not Lead to Additional Costs in Patients With Inflammatory Bowel Disease. Journal of Crohn's and Colitis, 2019, 13, 838-845.	0.6	19
134	Epidermolysis bullosa simplex with mottled pigmentation: Clinical aspects and confirmation of the P24L mutation in theKRT5 gene in further patients. , 1999, 86, 376-379.		18
135	Implications of intragenic marker homozygosity and haplotype sharing in a rare autosomal recessive disorder: the example of the collagen type XVII (COL17A1) locus in generalised atrophic benign epidermolysis bullosa. Human Genetics, 1997, 100, 230-235.	1.8	17
136	Milder phenotypes of glucose transporter type 1 deficiency syndrome. Developmental Medicine and Child Neurology, 2011, 53, 664-668.	1.1	17
137	ATL1 and REEP1 mutations in hereditary and sporadic upper motor neuron syndromes. Journal of Neurology, 2013, 260, 869-875.	1.8	17
138	Dystrophic Epidermolysis Bullosa Inversa with COL7A1 Mutations and Absence of GDA-J/F3 Protein. Pediatric Dermatology, 2003, 20, 243-248.	0.5	16
139	EEC syndrome, Arg227GlnTP63 mutation and micturition difficulties: Is there a genotypeâ€“phenotype correlation?. American Journal of Medical Genetics, Part A, 2007, 143A, 1114-1119.	0.7	16
140	Chromosomal sublocalization of the 2;13 translocation breakpoint in alveolar rhabdomyosarcoma. Genes Chromosomes and Cancer, 1992, 4, 241-249.	1.5	15
141	A case of neuromuscular mimicry. Neuromuscular Disorders, 2006, 16, 510-513.	0.3	14
142	Somatic mosaicism for theCOL7A1mutation p.Gly2034Arg in the unaffected mother of a patient with dystrophic epidermolysis bullosa pruriginosa. British Journal of Dermatology, 2015, 172, 778-781.	1.4	14
143	European registration process for Clinical Laboratory Geneticists in genetic healthcare. European Journal of Human Genetics, 2017, 25, 515-519.	1.4	13
144	Localization at a subband level of polymorphic 13q14 DNA probes for diagnosis of hereditary retinoblastoma and Wilson disease. Human Genetics, 1987, 77, 335-337.	1.8	12

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145	Apparent SMA I unlinked to 5q.. Journal of Medical Genetics, 1994, 31, 242-244.	1.5	12
146	Refinement by linkage analysis in two large families of the candidate region of the third locus (SCA3) for autosomal dominant cerebellar ataxia type I. Human Genetics, 1995, 96, 691-694.	1.8	12
147	Relative frequencies of cystic fibrosis mutations in The Netherlands as an illustration of significant regional variation in a small country. Human Genetics, 1998, 102, 587-590.	1.8	12
148	Regarding the rights and duties of Clinical Laboratory Geneticists in genetic healthcare systems; results of a survey in over 50 countries. European Journal of Human Genetics, 2019, 27, 1168-1174.	1.4	12
149	Close linkage of the Wilsons's disease locus to D13S12 in the chromosomal region 13q21 and not to ESD in 13q14. Human Genetics, 1990, 85, 560-562.	1.8	11
150	Holoprosencephaly, bilateral cleft lip and palate and ectrodactyly: another case and follow up. Clinical Dysmorphology, 2003, 12, 221-225.	0.1	11
151	Interlaboratory Diagnostic Validation of Conformation-Sensitive Capillary Electrophoresis for Mutation Scanning. Clinical Chemistry, 2010, 56, 593-602.	1.5	11
152	Identification of a novel mutation (867delA) in the glucose-6-phosphatase gene in two siblings with glycogen storage disease type Ia with different phenotypes. Human Mutation, 2000, 15, 381-381.	1.1	10
153	Extended gene analysis can increase specificity of neonatal screening for cystic fibrosis. Acta Paediatrica, International Journal of Paediatrics, 2006, 95, 1424-1428.	0.7	10
154	Developing National Guidance on Genetic Testing for Breast Cancer Predisposition: The Role of Economic Evidence?. Genetic Testing and Molecular Biomarkers, 2012, 16, 580-591.	0.3	10
155	Patientsâ€™ beliefs about medicine are associated with early thiopurine discontinuation in patients with inflammatory bowel diseases. European Journal of Gastroenterology and Hepatology, 2018, 30, 167-173.	0.8	10
156	Physical localisation of the chromosomal marker D13S31 places the Wilson disease locus at the junction of bands q14.3 and q21.1 of chromosome 13. Human Genetics, 1993, 91, 504-506.	1.8	9
157	The glutathione transferase Mu null genotype leads to lower 6-MMPR levels in patients treated with azathioprine but not with mercaptopurine. Pharmacogenomics Journal, 2018, 18, 160-166.	0.9	9
158	Validation of the determination of Î”F508 mutations of the cystic fibrosis gene in over 11000 mouthwashes. Human Genetics, 1996, 97, 334-336.	1.8	9
159	An anonymous single copy genomic clone at 13q12â€“13q13 identifies three RFLPs [HGM8] assignment no. D13S11]. Nucleic Acids Research, 1986, 14, 3148-3148.	6.5	8
160	Linkage and apparent heterogeneity in proximal spinal muscular atrophies. Neuromuscular Disorders, 1993, 3, 327-333.	0.3	8
161	Confirmation of clinical diagnosis in requests for prenatal prediction of SMA type I.. Journal of Neurology, Neurosurgery and Psychiatry, 1993, 56, 319-321.	0.9	8
162	Glycogen storage disease type Ia: Four novel mutations (175delGG, R170X, G266V and V338F) identified. Human Mutation, 1999, 13, 173-173.	1.1	8

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163	A Deletion Hybrid Breakpoint Map of the Chromosomal Region 13q14-q21 Orders 19 Genetic Markers in 10 Intervals. European Journal of Human Genetics, 1994, 2, 59-65.	1.4	8
164	Allelic Imbalance Analysis Using a Single-Nucleotide Polymorphism Microarray for the Detection of Bladder Cancer Recurrence. Clinical Cancer Research, 2008, 14, 8198-8204.	3.2	7
165	Design and Validation of a Conformation Sensitive Capillary Electrophoresis-Based Mutation Scanning System and Automated Data Analysis of the More than 15 kbp-Spanning Coding Sequence of the SACS Gene. Journal of Molecular Diagnostics, 2009, 11, 514-523.	1.2	7
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