Hannie Kremer

List of Publications by Year
in descending order

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161 papers	11,296 citations	34105 52 h-index	33894 99 g-index
170 all docs	170 docs citations	170 times ranked	13222 citing authors

#	Article	IF	Citations
1	Exploring the missing heritability in subjects with hearing loss, enlarged vestibular aqueducts, and a single or no pathogenic SLC26A4 variant. Human Genetics, 2022, 141, 465-484.	3.8	3
2	Novel gene discovery for hearing loss and other routes to increased diagnostic rates. Human Genetics, 2022, 141, 383-386.	3.8	3
3	Genotype-Phenotype Correlations of Pathogenic COCH Variants in DFNA9: A HuGE Systematic Review and Audiometric Meta-Analysis. Biomolecules, 2022, 12, 220.	4.0	5
4	Generation of Humanized Zebrafish Models for the In Vivo Assessment of Antisense Oligonucleotide-Based Splice Modulation Therapies. Methods in Molecular Biology, 2022, 2434, 281-299.	0.9	2
5	Usher syndrome type IV: clinically and molecularly confirmed by novel ARSG variants. Human Genetics, 2022, 141, 1723-1738.	3.8	16
6	Genetics of Hearing Impairment. Genes, 2022, 13, 852.	2.4	0
7	Scrutinizing pathogenicity of the USH2A c.2276â€‰Gâ€‰>â€‰T; p.(Cys759Phe) variant. Npj Genomic Medicine, 2022, 7, .	3.8	5
8	A RIPOR2 in-frame deletion is a frequent and highly penetrant cause of adult-onset hearing loss. Journal of Medical Genetics, 2021, 58, 96-104.	3.2	14
9	The Impact of Modern Technologies on Molecular Diagnostic Success Rates, with a Focus on Inherited Retinal Dystrophy and Hearing Loss. International Journal of Molecular Sciences, 2021, 22, 2943.	4.1	6
10	Cochlear supporting cells require GAS2 for cytoskeletal architecture and hearing. Developmental Cell, 2021, 56, 1526-1540.e7.	7.0	18
11	Variants in USP48 encoding ubiquitin hydrolase are associated with autosomal dominant non-syndromic hereditary hearing loss. Human Molecular Genetics, 2021, 30, 1785-1796.	2.9	6
12	Molecular Inversion Probe-Based Sequencing of USH2A Exons and Splice Sites as a Cost-Effective Screening Tool in USH2 and arRP Cases. International Journal of Molecular Sciences, 2021, 22, 6419.	4.1	8
13	AON-based degradation of c.151C>T mutant COCH transcripts associated with dominantly inherited hearing impairment DFNA9. Molecular Therapy - Nucleic Acids, 2021, 24, 274-283.	5.1	9
14	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	2.4	18
15	Antisense oligonucleotide-based treatment of retinitis pigmentosa caused by USH2A exon 13 mutations. Molecular Therapy, 2021, 29, 2441-2455.	8.2	75
16	Zebrafish as a Model to Evaluate a CRISPR/Cas9-Based Exon Excision Approach as a Future Treatment Option for EYS-Associated Retinitis Pigmentosa. International Journal of Molecular Sciences, 2021, 22, 9154.	4.1	6
17	Efficient Generation of Knock-In Zebrafish Models for Inherited Disorders Using CRISPR-Cas9 Ribonucleoprotein Complexes. International Journal of Molecular Sciences, 2021, 22, 9429.	4.1	10
18	A Novel COCH Mutation Affects the vWFA2 Domain and Leads to a Relatively Mild DFNA9 Phenotype. Otology and Neurotology, 2021, 42, e399-e407.	1.3	2

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19	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	6.2	75
20	De novo and inherited loss-of-function variants of ATP2B2 are associated with rapidly progressive hearing impairment. Human Genetics, 2019, 138, 61-72.	3.8	27
21	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	2.5	104
22	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	2.4	67
23	Hereditary hearing loss; about the known and the unknown. Hearing Research, 2019, 376, 58-68.	2.0	56
24	Variants affecting diverse domains of MEPE are associated with two distinct bone disorders, a craniofacial bone defect and otosclerosis. Genetics in Medicine, 2019, 21, 1199-1208.	2.4	17
25	Poor Splice-Site Recognition in a Humanized Zebrafish Knockin Model for the Recurrent Deep-Intronic c.7595-2144A>G Mutation inUSH2A. Zebrafish, 2018, 15, 597-609.	1.1	21
26	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	2.5	312
27	Antisense Oligonucleotide Design and Evaluation of Splice-Modulating Properties Using Cell-Based Assays. Methods in Molecular Biology, 2018, 1828, 519-530.	0.9	5
28	Grxcr2 is required for stereocilia morphogenesis in the cochlea. PLoS ONE, 2018, 13, e0201713.	2.5	11
29	Usherin defects lead to early-onset retinal dysfunction in zebrafish. Experimental Eye Research, 2018, 173, 148-159.	2.6	53
30	Heterozygous missense variants of LMX1A lead to nonsyndromic hearing impairment and vestibular dysfunction. Human Genetics, 2018, 137, 389-400.	3.8	32
31	MPZL2, Encoding the Epithelial Junctional Protein Myelin Protein Zero-like 2, Is Essential for Hearing in Man and Mouse. American Journal of Human Genetics, 2018, 103, 74-88.	6.2	34
32	Homozygous variants in KIAA1549, encoding a ciliary protein, are associated with autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2018, 55, 705-712.	3.2	13
33	A homozygousFITM2mutation causes a deafness-dystonia syndrome with motor regression and signs of ichthyosis and sensory neuropathy. DMM Disease Models and Mechanisms, 2017, 10, 105-118.	2.4	16
34	Broadening the phenotype of DFNB28: Mutations in TRIOBP are associated with moderate, stable hereditary hearing impairment. Hearing Research, 2017, 347, 56-62.	2.0	17
35	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.	2.8	90
36	Audiometric Characteristics of a Dutch DFNA10 Family With Mid-Frequency Hearing Impairment. Ear and Hearing, 2016, 37, 103-111.	2.1	17

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37	Trends in genetic diagnostics of hereditary hearing loss. Journal of Laryngology and Otology, 2016, 130, S27-S27.	0.8	0
38	Antisense Oligonucleotide-based Splice Correction for USH2A-associated Retinal Degeneration Caused by a Frequent Deep-intronic Mutation. Molecular Therapy - Nucleic Acids, 2016, 5, e381.	5.1	104
39	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
40	A combination of two truncating mutations in USH2A causes more severe and progressive hearing impairment in Usher syndrome type IIa. Hearing Research, 2016, 339, 60-68.	2.0	43
41	Novel and recurrent CIB2 variants, associated with nonsyndromic deafness, do not affect calcium buffering and localization in hair cells. European Journal of Human Genetics, 2016, 24, 542-549.	2.8	28
42	A Novel Locus Harbouring a Functional CD164 Nonsense Mutation Identified in a Large Danish Family with Nonsyndromic Hearing Impairment. PLoS Genetics, 2015, 11, e1005386.	3.5	18
43	The Ciliopathy Protein CC2D2A Associates with NINL and Functions in RAB8-MICAL3-Regulated Vesicle Trafficking. PLoS Genetics, 2015, 11, e1005575.	3.5	64
44	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. Genome Biology, 2015, 16, 293.	8.8	56
45	Nonsyndromic Hearing Loss Caused by USH1G Mutations. Ear and Hearing, 2015, 36, 205-211.	2.1	20
46	Allelic Mutations of KITLG, Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. American Journal of Human Genetics, 2015, 97, 647-660.	6.2	55
47	Progressive hearing loss and vestibular dysfunction caused by a homozygous nonsense mutation in CLIC5. European Journal of Human Genetics, 2015, 23, 189-194.	2.8	49
48	NINL and DZANK1 Co-function in Vesicle Transport and Are Essential for Photoreceptor Development in Zebrafish. PLoS Genetics, 2015, 11, e1005574.	3.5	23
49	Genetic Spectrum of Autosomal Recessive Non-Syndromic Hearing Loss in Pakistani Families. PLoS ONE, 2014, 9, e100146.	2.5	52
50	A canonical splice site mutation in GIPC3 causes sensorineural hearing loss in a large Pakistani family. Journal of Human Genetics, 2014, 59, 683-686.	2.3	4
51	Similar Phenotypes Caused by Mutations in OTOG and OTOGL. Ear and Hearing, 2014, 35, e84-e91.	2.1	21
52	Phosphorylation of the Usher syndrome 1G protein SANS controls Magi2-mediated endocytosis. Human Molecular Genetics, 2014, 23, 3923-3942.	2.9	28
53	Intrafamilial Variable Hearing Loss in TRPV4 Induced Spinal Muscular Atrophy. Annals of Otology, Rhinology and Laryngology, 2014, 123, 859-865.	1.1	8
54	Disruption of the Basal Body Protein POC1B Results in Autosomal-Recessive Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 95, 131-142.	6.2	65

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55	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.	2.5	303
56	Interfering with UDP-GlcNAc Metabolism and Heparan Sulfate Expression Using a Sugar Analogue Reduces Angiogenesis. ACS Chemical Biology, 2013, 8, 2331-2338.	3.4	32
57	Active Transport and Diffusion Barriers Restrict Joubert Syndrome-Associated ARL13B/ARL-13 to an Inv-like Ciliary Membrane Subdomain. PLoS Genetics, 2013, 9, e1003977.	3.5	91
58	Familial Aggregation of Pure Tone Hearing Thresholds in an Aging European Population. Otology and Neurotology, 2013, 34, 838-844.	1.3	15
59	Novel mutation in AAA domain of BCS1L causing Bjornstad syndrome. Journal of Human Genetics, 2013, 58, 819-821.	2.3	15
60	ZNF408 is mutated in familial exudative vitreoretinopathy and is crucial for the development of zebrafish retinal vasculature. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9856-9861.	7.1	144
61	A Mutation in CABP2 , Expressed in Cochlear Hair Cells, Causes Autosomal-Recessive Hearing Impairment. American Journal of Human Genetics, 2012, 91, 636-645.	6.2	96
62	Mutations in OTOGL , Encoding the Inner Ear Protein Otogelin-like, Cause Moderate Sensorineural Hearing Loss. American Journal of Human Genetics, 2012, 91, 872-882.	6.2	97
63	Mutations of the Gene Encoding Otogelin Are a Cause of Autosomal-Recessive Nonsyndromic Moderate Hearing Impairment. American Journal of Human Genetics, 2012, 91, 883-889.	6.2	69
64	The mitotic spindle protein SPAG5/Astrin connects to the Usher protein network postmitotically. Cilia, 2012, 1, 2.	1.8	18
65	CLRN1 Mutations Cause Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2011, 118, 1444-1448.	5.2	61
66	Direct interaction of the Usher syndrome 1G protein SANS and myomegalin in the retina. Biochimica Et Biophysica Acta - Molecular Cell Research, 2011, 1813, 1883-1892.	4.1	43
67	Next-Generation Sequencing Identifies Mutations of SMPX, which Encodes the Small Muscle Protein, X-Linked, as a Cause of Progressive Hearing Impairment. American Journal of Human Genetics, 2011, 88, 628-634.	6.2	88
68	Genotypeâ€“Phenotype Correlation in DFNB8/10 Families with TMPRSS3 Mutations. JARO - Journal of the Association for Research in Otolaryngology, 2011, 12, 753-766.	1.8	69
69	DFNA8/12 caused by TECTA mutations is the most identified subtype of nonsyndromic autosomal dominant hearing loss. Human Mutation, 2011, 32, 825-834.	2.5	73
70	Progressive Sensorineural Hearing Loss and Normal Vestibular Function in a Dutch DFNB7/11 Family with a Novel Mutation in <i>TMC1</i>. Audiology and Neuro-Otology, 2011, 16, 93-105.	1.3	36
71	SDHAF2 (PGL2-SDH5) and Hereditary Head and Neck Paraganglioma. Clinical Cancer Research, 2011, 17, 247-254.	7.0	137
72	Gipc3 mutations associated with audiogenic seizures and sensorineural hearing loss in mouse and human. Nature Communications, 2011, 2, 201.	12.8	95

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73	Autosomal Recessive Mental Retardation, Deafness, Ankylosis, and Mild Hypophosphatemia Associated with a NovelANKHMutation in a Consanguineous Family. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E189-E198.	3.6	42
74	Homozygosity Mapping Reveals Mutations of GRXCR1 as a Cause of Autosomal-Recessive Nonsyndromic Hearing Impairment. American Journal of Human Genetics, 2010, 86, 138-147.	6.2	58
75	Mutations in TPRN Cause a Progressive Form of Autosomal-Recessive Nonsyndromic Hearing Loss. American Journal of Human Genetics, 2010, 86, 479-484.	6.2	56
76	Mutations in PTPRQ Are a Cause of Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB84 and Associated with Vestibular Dysfunction. American Journal of Human Genetics, 2010, 86, 604-610.	6.2	72
77	Dominant Mutations in KBTBD13, a Member of the BTB/Kelch Family, Cause Nemaline Myopathy with Cores. American Journal of Human Genetics, 2010, 87, 842-847.	6.2	143
78	Multiple enhancers located in a 1-Mb region upstream of POU3F4 promote expression during inner ear development and may be required for hearing. Human Genetics, 2010, 128, 411-419.	3.8	35
79	Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. Nature Genetics, 2010, 42, 160-164.	21.4	228
80	Association of Whirlin with Ca_v1.3 (Î±_{1D}) Channels in Photoreceptors, Defining a Novel Member of the Usher Protein Network. , 2010, 51, 2338.		52
81	Sequence variants of the DFNB31 gene among Usher syndrome patients of diverse origin. Molecular Vision, 2010, 16, 495-500.	1.1	13
82	SDH5 , a Gene Required for Flavination of Succinate Dehydrogenase, Is Mutated in Paraganglioma. Science, 2009, 325, 1139-1142.	12.6	682
83	Vestibular Impairment in a Dutch DFNA15 Family with an L289F Mutation in POU4F3. Audiology and Neuro-Otology, 2009, 14, 303-307.	1.3	11
84	Audiometric and Vestibular Features in a Second Dutch DFNA20/26 Family with a Novel Mutation inACTG1. Annals of Otology, Rhinology and Laryngology, 2009, 118, 382-390.	1.1	20
85	Usher syndrome and Leber congenital amaurosis are molecularly linked via a novel isoform of the centrosomal ninein-like protein. Human Molecular Genetics, 2009, 18, 51-64.	2.9	43
86	GRM7 variants confer susceptibility to age-related hearing impairment. Human Molecular Genetics, 2009, 18, 785-796.	2.9	174
87	Mild and Variable Audiometric and Vestibular Features in a Third DFNA15 Family with a Novel Mutation in POU4F3. Annals of Otology, Rhinology and Laryngology, 2009, 118, 313-320.	1.1	12
88	Positional Cloning of Deafness Genes. Methods in Molecular Biology, 2009, 493, 215-239.	0.9	1
89	Occupational Noise, Smoking, and a High Body Mass Index are Risk Factors for Age-related Hearing Impairment and Moderate Alcohol Consumption is Protective: A European Population-based Multicenter Study. JARO - Journal of the Association for Research in Otolaryngology, 2008, 9, 264-276.	1.8	214
90	Missense mutations inPOU4F3cause autosomal dominant hearing impairment DFNA15 and affect subcellular localization and DNA binding. Human Mutation, 2008, 29, 545-554.	2.5	62

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91	Genome-wide SNP-Based Linkage Scan Identifies a Locus on 8q24 for an Age-Related Hearing Impairment Trait. American Journal of Human Genetics, 2008, 83, 401-407.	6.2	54
92	Mid-frequency DFNA8/12 hearing loss caused by a synonymous TECTA mutation that affects an exonic splice enhancer. European Journal of Human Genetics, 2008, 16, 1430-1436.	2.8	33
93	Mutations of LRTOMT, a fusion gene with alternative reading frames, cause nonsyndromic deafness in humans. Nature Genetics, 2008, 40, 1335-1340.	21.4	65
94	A novel TECTA mutation confirms the recognizable phenotype among autosomal recessive hearing impairment families. International Journal of Pediatric Otorhinolaryngology, 2008, 72, 249-255.	1.0	43
95	Mutations of ESRRB Encoding Estrogen-Related Receptor Beta Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB35. American Journal of Human Genetics, 2008, 82, 125-138.	6.2	127
96	A novel Usher protein network at the periciliary reloading point between molecular transport machineries in vertebrate photoreceptor cells. Human Molecular Genetics, 2008, 17, 71-86.	2.9	224
97	The grainyhead like 2 gene (GRHL2), alias TFCP2L3, is associated with age-related hearing impairment. Human Molecular Genetics, 2008, 17, 159-169.	2.9	121
98	Audiometric Characteristics of a Dutch Family Linked to DFNA15 With a Novel Mutation (p.L289F) in POU4F3. JAMA Otolaryngology, 2008, 134, 294.	1.2	26
99	MPP1 links the Usher protein network and the Crumbs protein complex in the retina. Human Molecular Genetics, 2007, 16, 1993-2003.	2.9	36
100	Phenotype Description of a Novel DFNA9/COCH Mutation, I109T. Annals of Otology, Rhinology and Laryngology, 2007, 116, 349-357.	1.1	29
101	Clinical Characteristics of a Dutch DFNA9 Family with a Novel COCH Mutation, G87W. Audiology and Neuro-Otology, 2007, 12, 77-84.	1.3	18
102	Vertical Corneal Striae in Families with Autosomal Dominant Hearing Loss: DFNA9/COCH. American Journal of Ophthalmology, 2007, 143, 847-852.e6.	3.3	7
103	MYO15A (DFNB3) mutations in Turkish hearing loss families and functional modeling of a novel motor domain mutation. American Journal of Medical Genetics, Part A, 2007, 143A, 2382-2389.	1.2	45
104	Involvement ofDFNB59 mutations in autosomal recessive nonsyndromic hearing impairment. Human Mutation, 2007, 28, 718-723.	2.5	58
105	The contribution of GJB2 (Connexin 26) 35delG to age-related hearing impairment and noise-induced hearing loss. Otology and Neurotology, 2007, 28, 970-5.	1.3	37
106	Comparison of 12 Reference Genes for Normalization of Gene Expression Levels in Epstein-Barr Virus-Transformed Lymphoblastoid Cell Lines and Fibroblasts. Molecular Diagnosis and Therapy, 2006, 10, 197-204.	3.8	51
107	BSCL2 mutations in two Dutch families with overlapping Silver syndrome-distal hereditary motor neuropathy. Neuromuscular Disorders, 2006, 16, 122-125.	0.6	29
108	Cochleovestibular and Ocular Features in a Dutch DFNA11 Family. Otology and Neurotology, 2006, 27, 323-331.	1.3	18

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109	A Novel TECTA Mutation in a Dutch DFNA8/12 Family Confirms Genotypeâ€“Phenotype Correlation. JARO - Journal of the Association for Research in Otolaryngology, 2006, 7, 173-181.	1.8	44
110	Mutations in thelipoma HMGIC fusion partner-like 5 (LHFPL5)gene cause autosomal recessive nonsyndromic hearing loss. Human Mutation, 2006, 27, 633-639.	2.5	58
111	Identification of a novel COCH mutation, G87W, causing autosomal dominant hearing impairment (DFNA9). American Journal of Medical Genetics, Part A, 2006, 140A, 1791-1794.	1.2	26
112	The DFNB31 gene product whirlin connects to the Usher protein network in the cochlea and retina by direct association with USH2A and VLGR1. Human Molecular Genetics, 2006, 15, 751-765.	2.9	162
113	Usher syndrome: molecular links of pathogenesis, proteins and pathways. Human Molecular Genetics, 2006, 15, R262-R270.	2.9	219
114	Development of a genotyping microarray for Usher syndrome. Journal of Medical Genetics, 2006, 44, 153-160.	3.2	94
115	Cochlin immunostaining of inner ear pathologic deposits and proteomic analysis in DFNA9 deafness and vestibular dysfunction. Human Molecular Genetics, 2006, 15, 1071-1085.	2.9	100
116	Audiometric, Vestibular, and Genetic Aspects of a DFNA9 Family with a G88E COCH Mutation. Otology and Neurotology, 2005, 26, 926-933.	1.3	38
117	Vestibular Deterioration Precedes Hearing Deterioration in the P51S COCH Mutation (DFNA9): An Analysis in 74 Mutation Carriers. Otology and Neurotology, 2005, 26, 918-925.	1.3	42
118	Phenotype Determination Guides Swift Genotyping of a DFNA2/KCNQ4 Family With a Hot Spot Mutation (W276S). Otology and Neurotology, 2005, 26, 52-58.	1.3	31
119	Fine mapping of autosomal dominant nonsyndromic hearing impairmentDFNA21 to chromosome 6p24.1-22.3. American Journal of Medical Genetics, Part A, 2005, 137A, 41-46.	1.2	5
120	Scaffold protein harmonin (USH1C) provides molecular links between Usher syndrome type 1 and type 2. Human Molecular Genetics, 2005, 14, 3933-3943.	2.9	164
121	Hearing impairment in Dutch patients with connexin 26 (GJB2) and connexin 30 (GJB6) mutations. International Journal of Pediatric Otorhinolaryngology, 2005, 69, 165-174.	1.0	36
122	GJB2 mutations in Turkish patients with ARNSHL: prevalence and two novel mutations. Hearing Research, 2005, 203, 88-93.	2.0	47
123	The Benign Concentric Annular Macular Dystrophy Locus Maps to 6p12.3-q16. , 2004, 45, 30.		40
124	Longitudinal Phenotypic Analysis in Patients with Connexin 26 (GJB2) (DFNB1) and Connexin 30 (GJB6) Mutations. Annals of Otology, Rhinology and Laryngology, 2004, 113, 587-593.	1.1	8
125	A Novel Mutation Identified in the DFNA5 Gene in a Dutch Family: A Clinical and Genetic Evaluation. Audiology and Neuro-Otology, 2004, 9, 34-46.	1.3	74
126	Evaluation of visual impairment in Usher syndrome 1b and Usher syndrome 2a. Acta Ophthalmologica, 2004, 82, 131-139.	0.3	35

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127	Identification and molecular modelling of a mutation in the motor head domain of myosin VIIA in a family with autosomal dominant hearing impairment (DFNA11). Human Genetics, 2004, 115, 149-156.	3.8	45
128	USH2A Mutation analysis in 70 Dutch families with Usher syndrome type II. Human Mutation, 2004, 24, 185-185.	2.5	50
129	Identification of 51 Novel Exons of the Usher Syndrome Type 2A (USH2A) Gene That Encode Multiple Conserved Functional Domains and That Are Mutated in Patients with Usher Syndrome Type II. American Journal of Human Genetics, 2004, 74, 738-744.	6.2	176
130	Variable Clinical Features in Patients with CDH23 Mutations (USH1D-DFNB12). Otology and Neurotology, 2004, 25, 699-706.	1.3	29
131	A Dutch Family With Hearing Loss Linked to the DFNA20/26 Locus. JAMA Otolaryngology, 2004, 130, 281.	1.2	14
132	Mutations in the calcium-binding motifs of CDH23 and the 35delG mutation in GJB2 cause hearing loss in one family. Human Genetics, 2003, 112, 156-163.	3.8	43
133	Progression of Low-Frequency Sensorineural Hearing Loss (DFNA6/14-WFS1). JAMA Otolaryngology, 2003, 129, 421.	1.2	36
134	Mutations in the WFS1 gene that cause low-frequency sensorineural hearing loss are small non-inactivating mutations. Human Genetics, 2002, 110, 389-394.	3.8	81
135	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. Nature Genetics, 2001, 29, 465-468.	21.4	1,555
136	PTEN Mutation Analysis in Two Genetic Subtypes of High-Grade Oligodendroglial Tumors. Cancer Genetics and Cytogenetics, 2000, 119, 42-47.	1.0	23
137	Fine mapping of the neurally expressed gene SOX14 to human 3q23, relative to three congenital diseases. Human Genetics, 2000, 106, 432-439.	3.8	19
138	Novel PTEN mutations in patients with Cowden disease: absence of clear genotypeâ€“phenotype correlations. European Journal of Human Genetics, 1999, 7, 267-273.	2.8	302
139	Regulation and expression of the murine Pmp22 gene. Mammalian Genome, 1999, 10, 419-422.	2.2	12
140	X-linked mental retardation associated with cleft lip/palate maps to Xp11.3-q21.3. , 1999, 85, 216-220.		53
141	X-linked nonspecific mental retardation (MRX) linkage studies in 25 unrelated families: The European XLMR consortium. , 1999, 85, 263-265.		16
142	Four families (MRX43, MRX44, MRX45, MRX52) with nonspecific X-linked mental retardation: Clinical and psychometric data and results of linkage analysis. , 1999, 85, 290-304.		26
143	X-linked mental retardation: Evidence for a recent mutation in a five-generation family (MRX65) linked to the pericentromeric region. , 1999, 85, 305-308.		11
144	The Gene for Hypotrichosis of Marie Unna Maps between D8S258 and D8S298: Exclusion of the hr Gene by cDNA and Genomic Sequencing. American Journal of Human Genetics, 1999, 65, 413-419.	6.2	49

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145	An Atypical Form of Bullous Congenital Ichthyosiform Erythroderma is Caused by a Mutation in the L12 Linker Region of Keratin 1. Journal of Investigative Dermatology, 1998, 111, 1224-1226.	0.7	23
146	Two Types of Polyadenated mRNAs are Synthesized from Drosophila Replication-Dependent Histone Genes. FEBS Journal, 1997, 244, 294-300.	0.2	18
147	Naturally occurring testis-specific histone H3 antisense transcripts inDrosophila. Molecular Reproduction and Development, 1997, 48, 413-420.	2.0	10
148	Further Delineation of the Critical Region for Noonan Syndrome on the Long Arm of Chromosome 12. European Journal of Human Genetics, 1997, 5, 336-337.	2.8	21
149	A gene for nonspecific X-linked mental retardation (MRX41) is located in the distal segment of Xq28. American Journal of Medical Genetics Part A, 1996, 64, 131-133.	2.4	19
150	Male pseudohermaphroditism due to a homozygous missense mutation of the luteinizing hormone receptor gene. Nature Genetics, 1995, 9, 160-164.	21.4	330
151	Structure and expression of histone H3.3 genes inDrosophila melanogasterandDrosophila hydei. Genome, 1995, 38, 586-600.	2.0	59
152	Localizaion of the gene for dominant cystoid macular dystrophy on chromosome 7p. Human Molecular Genetics, 1994, 3, 299-302.	2.9	114
153	Genetic Linkage of the Keratin Type II Gene Cluster with Ichthyosis Bullosa of Siemens and with Autosomal Dominant Ichthyosis Exfoliativa. Journal of Investigative Dermatology, 1994, 103, 282-285.	0.7	32
154	Ichthyosis Bullosa of Siemens Is Caused by Mutations in the Keratin 2e Gene. Journal of Investigative Dermatology, 1994, 103, 286-289.	0.7	94
155	Cosegregation of missense mutations of the luteinizing hormone receptor gene with familial male-limited precocious puberty. Human Molecular Genetics, 1993, 2, 1779-1783.	2.9	165
156	Spermatogenesis inDrosophila hydei: A genetic survey. Roux's Archives of Developmental Biology, 1990, 199, 251-280.	1.2	14
157	Spermatogenesis of Drosophila hydei. International Review of Cytology, 1990, 123, 129-175.	6.2	33
158	Isolation and characterization of aDrosophila hydeihistone DNA repeat unit. Nucleic Acids Research, 1990, 18, 1573-1586.	14.5	46
159	Genetic Analysis of Spermatogenesis in Drosophila Hydei: Male Sterile Mutations Affecting Nuclear Development During the Primary Spermatocyte Stage. , 1990, , 99-103.		0
160	Y chromosomal fertility genes of Drosophila: a new type of eukaryotic genes. Genome, 1989, 31, 561-571.	2.0	46
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