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	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. Nature Genetics, 2001, 29, 465-468.	21.4	1,555
2	SDH5 , a Gene Required for Flavination of Succinate Dehydrogenase, Is Mutated in Paraganglioma. Science, 2009, 325, 1139-1142.	12.6	682
3	Male pseudohermaphroditism due to a homozygous missense mutation of the luteinizing hormone receptor gene. Nature Genetics, 1995, 9, 160-164.	21.4	330
4	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	2.5	312
5	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
6	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
7	Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. Nature Genetics, 2010, 42, 160-164.	21.4	228
8	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
9	Usher syndrome: molecular links of pathogenesis, proteins and pathways. Human Molecular Genetics, 2006, 15, R262-R270.	2.9	219
10	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
11	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
12	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
13	GRM7 variants confer susceptibility to age-related hearing impairment. Human Molecular Genetics, 2009, 18, 785-796.	2.9	174
14	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
15	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
16	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
17	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
18	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

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19	SDHAF2 (PGL2-SDH5) and Hereditary Head and Neck Paraganglioma. Clinical Cancer Research, 2011, 17, 247-254.	7.0	137
20	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
21	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
22	Localizaion of the gene for dominant cystoid macular dystrophy on chromosome 7p. Human Molecular Genetics, 1994, 3, 299-302.	2.9	114
23	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
24	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	2.5	104
25	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
26	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
27	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
28	Gipc3 mutations associated with audiogenic seizures and sensorineural hearing loss in mouse and human. Nature Communications, 2011, 2, 201.	12.8	95
29	Ichthyosis Bullosa of Siemens Is Caused by Mutations in the Keratin 2e Gene. Journal of Investigative Dermatology, 1994, 103, 286-289.	0.7	94
30	Development of a genotyping microarray for Usher syndrome. Journal of Medical Genetics, 2006, 44, 153-160.	3.2	94
31	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
32	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
33	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
34	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
35	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
36	Antisense oligonucleotide-based treatment of retinitis pigmentosa caused by USH2A exon 13 mutations. Molecular Therapy, 2021, 29, 2441-2455.	8.2	75

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37	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
38	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
39	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
40	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
41	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
42	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	2.4	67
43	Mutations of LRTOMT, a fusion gene with alternative reading frames, cause nonsyndromic deafness in humans. Nature Genetics, 2008, 40, 1335-1340.	21.4	65
44	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
45	The Ciliopathy Protein CC2D2A Associates with NINL and Functions in RAB8-MICAL3-Regulated Vesicle Trafficking. PLoS Genetics, 2015, 11, e1005575.	3.5	64
46	Missense mutations inPOU4F3cause autosomal dominant hearing impairment DFNA15 and affect subcellular localization and DNA binding. Human Mutation, 2008, 29, 545-554.	2.5	62
47	CLRN1 Mutations Cause Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2011, 118, 1444-1448.	5.2	61
48	Structure and expression of histone H3.3 genes inDrosophila melanogasterandDrosophila hydei. Genome, 1995, 38, 586-600.	2.0	59
49	Chromatin organization in the male germ line of Drosophila hydei. Chromosoma, 1986, 94, 147-161.	2.2	58
50	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
51	Involvement ofDFNB59 mutations in autosomal recessive nonsyndromic hearing impairment. Human Mutation, 2007, 28, 718-723.	2.5	58
52	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
53	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
54	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. Genome Biology, 2015, 16, 293.	8.8	56

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55	Hereditary hearing loss; about the known and the unknown. Hearing Research, 2019, 376, 58-68.	2.0	56
56	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
57	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
58	X-linked mental retardation associated with cleft lip/palate maps to Xp11.3-q21.3. , 1999, 85, 216-220.		53
59	Usherin defects lead to early-onset retinal dysfunction in zebrafish. Experimental Eye Research, 2018, 173, 148-159.	2.6	53
60	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
61	Genetic Spectrum of Autosomal Recessive Non-Syndromic Hearing Loss in Pakistani Families. PLoS ONE, 2014, 9, e100146.	2.5	52
62	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
63	USH2A Mutation analysis in 70 Dutch families with Usher syndrome type II. Human Mutation, 2004, 24, 185-185.	2.5	50
64	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
65	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
66	GJB2 mutations in Turkish patients with ARNSHL: prevalence and two novel mutations. Hearing Research, 2005, 203, 88-93.	2.0	47
67	Y chromosomal fertility genes of Drosophila: a new type of eukaryotic genes. Genome, 1989, 31, 561-571.	2.0	46
68	Isolation and characterization of aDrosophila hydeihistone DNA repeat unit. Nucleic Acids Research, 1990, 18, 1573-1586.	14.5	46
69	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
70	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
71	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
72	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

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73	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
74	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
75	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
76	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
77	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
78	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
79	The Benign Concentric Annular Macular Dystrophy Locus Maps to 6p12.3-q16. , 2004, 45, 30.		40
80	Audiometric, Vestibular, and Genetic Aspects of a DFNA9 Family with a G88E COCH Mutation. Otology and Neurotology, 2005, 26, 926-933.	1.3	38
81	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
82	Progression of Low-Frequency Sensorineural Hearing Loss (DFNA6/14-WFS1). JAMA Otolaryngology, 2003, 129, 421.	1.2	36
83	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
84	MPP1 links the Usher protein network and the Crumbs protein complex in the retina. Human Molecular Genetics, 2007, 16, 1993-2003.	2.9	36
85	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
86	Evaluation of visual impairment in Usher syndrome 1b and Usher syndrome 2a. Acta Ophthalmologica, 2004, 82, 131-139.	0.3	35
87	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
88	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
89	Spermatogenesis of Drosophila hydei. International Review of Cytology, 1990, 123, 129-175.	6.2	33
90	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

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91	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
92	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
93	Heterozygous missense variants of LMX1A lead to nonsyndromic hearing impairment and vestibular dysfunction. Human Genetics, 2018, 137, 389-400.	3.8	32
94	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
95	Variable Clinical Features in Patients with CDH23 Mutations (USH1D-DFNB12). Otology and Neurotology, 2004, 25, 699-706.	1.3	29
96	BSCL2 mutations in two Dutch families with overlapping Silver syndrome-distal hereditary motor neuropathy. Neuromuscular Disorders, 2006, 16, 122-125.	0.6	29
97	Phenotype Description of a Novel DFNA9/COCH Mutation, I109T. Annals of Otology, Rhinology and Laryngology, 2007, 116, 349-357.	1.1	29
98	Phosphorylation of the Usher syndrome 1G protein SANS controls Magi2-mediated endocytosis. Human Molecular Genetics, 2014, 23, 3923-3942.	2.9	28
99	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
100	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
101	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
102	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
103	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
104	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
105	PTEN Mutation Analysis in Two Genetic Subtypes of High-Grade Oligodendroglial Tumors. Cancer Genetics and Cytogenetics, 2000, 119, 42-47.	1.0	23
106	NINL and DZANK1 Co-function in Vesicle Transport and Are Essential for Photoreceptor Development in Zebrafish. PLoS Genetics, 2015, 11, e1005574.	3.5	23
107	Similar Phenotypes Caused by Mutations in OTOG and OTOGL. Ear and Hearing, 2014, 35, e84-e91.	2.1	21
108	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

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109	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
110	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
111	Nonsyndromic Hearing Loss Caused by USH1G Mutations. Ear and Hearing, 2015, 36, 205-211.	2.1	20
112	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
113	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
114	Two Types of Polyadenated mRNAs are Synthesized from Drosophila Replication-Dependent Histone Genes. FEBS Journal, 1997, 244, 294-300.	0.2	18
115	Cochleovestibular and Ocular Features in a Dutch DFNA11 Family. Otology and Neurotology, 2006, 27, 323-331.	1.3	18
116	Clinical Characteristics of a Dutch DFNA9 Family with a Novel COCH Mutation, G87W. Audiology and Neuro-Otology, 2007, 12, 77-84.	1.3	18
117	The mitotic spindle protein SPAG5/Astrin connects to the Usher protein network postmitotically. Cilia, 2012, 1, 2.	1.8	18
118	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
119	Cochlear supporting cells require GAS2 for cytoskeletal architecture and hearing. Developmental Cell, 2021, 56, 1526-1540.e7.	7.0	18
120	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	2.4	18
121	Audiometric Characteristics of a Dutch DFNA10 Family With Mid-Frequency Hearing Impairment. Ear and Hearing, 2016, 37, 103-111.	2.1	17
122	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
123	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
124	X-linked nonspecific mental retardation (MRX) linkage studies in 25 unrelated families: The European XLMR consortium. , 1999, 85, 263-265.		16
125	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
126	Usher syndrome type IV: clinically and molecularly confirmed by novel ARSG variants. Human Genetics, 2022, 141, 1723-1738.	3.8	16

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127	Familial Aggregation of Pure Tone Hearing Thresholds in an Aging European Population. Otology and Neurotology, 2013, 34, 838-844.	1.3	15
128	Novel mutation in AAA domain of BCS1L causing Bjornstad syndrome. Journal of Human Genetics, 2013, 58, 819-821.	2.3	15
129	Spermatogenesis inDrosophila hydei: A genetic survey. Roux's Archives of Developmental Biology, 1990, 199, 251-280.	1.2	14
130	A Dutch Family With Hearing Loss Linked to the DFNA20/26 Locus. JAMA Otolaryngology, 2004, 130, 281.	1.2	14
131	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
132	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
133	Sequence variants of the DFNB31 gene among Usher syndrome patients of diverse origin. Molecular Vision, 2010, 16, 495-500.	1.1	13
134	Regulation and expression of the murine Pmp22 gene. Mammalian Genome, 1999, 10, 419-422.	2.2	12
135	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
136	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
137	Vestibular Impairment in a Dutch DFNA15 Family with an L289F Mutation in POU4F3. Audiology and Neuro-Otology, 2009, 14, 303-307.	1.3	11
138	Grxcr2 is required for stereocilia morphogenesis in the cochlea. PLoS ONE, 2018, 13, e0201713.	2.5	11
139	Naturally occurring testis-specific histone H3 antisense transcripts inDrosophila. Molecular Reproduction and Development, 1997, 48, 413-420.	2.0	10
140	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
141	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
142	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
143	Intrafamilial Variable Hearing Loss in TRPV4 Induced Spinal Muscular Atrophy. Annals of Otology, Rhinology and Laryngology, 2014, 123, 859-865.	1.1	8
144	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

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145	Vertical Corneal Striae in Families with Autosomal Dominant Hearing Loss: DFNA9/COCH. American Journal of Ophthalmology, 2007, 143, 847-852.e6.	3.3	7
146	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
147	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
148	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
149	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
150	Antisense Oligonucleotide Design and Evaluation of Splice-Modulating Properties Using Cell-Based Assays. Methods in Molecular Biology, 2018, 1828, 519-530.	0.9	5
151	Genotype-Phenotype Correlations of Pathogenic COCH Variants in DFNA9: A HuGE Systematic Review and Audiometric Meta-Analysis. Biomolecules, 2022, 12, 220.	4.0	5
152	Scrutinizing pathogenicity of the USH2A c.2276â€‰Gâ€‰>â€‰T; p.(Cys759Phe) variant. Npj Genomic Medicine, 2022, 7, .	3.8	5
153	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
154	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
155	Novel gene discovery for hearing loss and other routes to increased diagnostic rates. Human Genetics, 2022, 141, 383-386.	3.8	3
156	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
157	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
158	Positional Cloning of Deafness Genes. Methods in Molecular Biology, 2009, 493, 215-239.	0.9	1
159	Trends in genetic diagnostics of hereditary hearing loss. Journal of Laryngology and Otology, 2016, 130, S27-S27.	0.8	0
160	Genetic Analysis of Spermatogenesis in Drosophila Hydei: Male Sterile Mutations Affecting Nuclear Development During the Primary Spermatocyte Stage. , 1990, , 99-103.		0
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