## **Christine** Petit

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Defective myosin VIIA gene responsible for Usher syndrome type IB. Nature, 1995, 374, 60-61.	27.8	1,101
2	A novel mutation in the potassium channel gene KVLQT1 causes the Jervell and Lange-Nielsen cardioauditory syndrome. Nature Genetics, 1997, 15, 186-189.	21.4	844
3	KCNQ4, a Novel Potassium Channel Expressed in Sensory Outer Hair Cells, Is Mutated in Dominant Deafness. Cell, 1999, 96, 437-446.	28.9	783
4	Loss-of-function mutations in FGFR1 cause autosomal dominant Kallmann syndrome. Nature Genetics, 2003, 33, 463-465.	21.4	764
5	A human homologue of the Drosophila eyes absent gene underlies Branchio-Oto-Renal (BOR) syndrome and identifies a novel gene family. Nature Genetics, 1997, 15, 157-164.	21.4	628
6	Otoferlin, Defective in a Human Deafness Form, Is Essential for Exocytosis at the Auditory Ribbon Synapse. Cell, 2006, 127, 277-289.	28.9	554
7	A mutation in OTOF, encoding otoferlin, a FER-1-like protein, causes DFNB9, a nonsyndromic form of deafness. Nature Genetics, 1999, 21, 363-369.	21.4	481
8	A defect in harmonin, a PDZ domain-containing protein expressed in the inner ear sensory hair cells, underlies Usher syndrome type 1C. Nature Genetics, 2000, 26, 51-55.	21.4	449
9	Clinical features of the prevalent form of childhood deafness, DFNB1, due to a connexin-26 gene defect: implications for genetic counselling. Lancet, The, 1999, 353, 1298-1303.	13.7	412
10	Targeted Ablation of Connexin26 in the Inner Ear Epithelial Gap Junction Network Causes Hearing Impairment and Cell Death. Current Biology, 2002, 12, 1106-1111.	3.9	409
11	Myosin VIIa, harmonin and cadherin 23, three Usher I gene products that cooperate to shape the sensory hair cell bundle. EMBO Journal, 2002, 21, 6689-6699.	7.8	392
12	Kallmann Syndrome: Mutations in the Genes Encoding Prokineticin-2 and Prokineticin Receptor-2. PLoS Genetics, 2006, 2, e175.	3.5	391
13	The autosomal recessive isolated deafness, DFNB2, and the Usher 1B syndrome are allelic defects of the myosin-VIIA gene. Nature Genetics, 1997, 16, 191-193.	21.4	387
14	SIX1 mutations cause branchio-oto-renal syndrome by disruption of EYA1-SIX1-DNA complexes. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 8090-8095.	7.1	374
15	Connexin30 (Gjb6)-deficiency causes severe hearing impairment and lack of endocochlear potential. Human Molecular Genetics, 2003, 12, 13-21.	2.9	322
16	Ciliary proteins link basal body polarization to planar cell polarity regulation. Nature Genetics, 2008, 40, 69-77.	21.4	306
17	A cluster of sulfatase genes on Xp22.3: Mutations in chondrodysplasia punctata (CDPX) and implications for warfarin embryopathy. Cell, 1995, 81, 15-25.	28.9	303
18	Defects in whirlin, a PDZ domain molecule involved in stereocilia elongation, cause deafness in the whirler mouse and families with DFNB31. Nature Genetics, 2003, 34, 421-428.	21.4	293

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19	Connexin 26 gene linked to a dominant deafness. Nature, 1998, 393, 319-320.	27.8	291
20	Molecular Genetics of Hearing Loss. Annual Review of Genetics, 2001, 35, 589-645.	7.6	290
21	Prevalence and Evolutionary Origins of the del(GJB6-D13S1830) Mutation in the DFNB1 Locus in Hearing-Impaired Subjects: a Multicenter Study. American Journal of Human Genetics, 2003, 73, 1452-1458.	6.2	269
22	A non–syndromic form of neurosensory, recessive deafness maps to the pericentromeric region of chromosome 13q. Nature Genetics, 1994, 6, 24-28.	21.4	262
23	Usher syndrome type I G (USH1G) is caused by mutations in the gene encoding SANS, a protein that associates with the USH1C protein, harmonin. Human Molecular Genetics, 2003, 12, 463-471.	2.9	262
24	Mutations in the gene encoding pejvakin, a newly identified protein of the afferent auditory pathway, cause DFNB59 auditory neuropathy. Nature Genetics, 2006, 38, 770-778.	21.4	262
25	USHERSYNDROME: From Genetics to Pathogenesis. Annual Review of Genomics and Human Genetics, 2001, 2, 271-297.	6.2	248
26	Genes responsible for human hereditary deafness: symphony of a thousand. Nature Genetics, 1996, 14, 385-391.	21.4	245
27	Interactions in the network of Usher syndrome type 1 proteins. Human Molecular Genetics, 2005, 14, 347-356.	2.9	231
28	Abnormal pattern detected in fragile-X patients by pulsed-field gel electrophoresis. Nature, 1991, 349, 624-626.	27.8	217
29	Heterogeneity in the mutations responsible for X chromosome-linked Kallmann syndrome. Human Molecular Genetics, 1993, 2, 373-377.	2.9	206
30	SLC26A4 gene is frequently involved in nonsyndromic hearing impairment with enlarged vestibular aqueduct in Caucasian populations. European Journal of Human Genetics, 2006, 14, 773-779.	2.8	204
31	Anosmin-1, Defective in the X-Linked Form of Kallmann Syndrome, Promotes Axonal Branch Formation from Olfactory Bulb Output Neurons. Cell, 2002, 109, 217-228.	28.9	201
32	Human adenylate kinase 2 deficiency causes a profound hematopoietic defect associated with sensorineural deafness. Nature Genetics, 2009, 41, 106-111.	21.4	198
33	How the Genetics of Deafness Illuminates Auditory Physiology. Annual Review of Physiology, 2011, 73, 311-334.	13.1	195
34	A core cochlear phenotype in USH1 mouse mutants implicates fibrous links of the hair bundle in its cohesion, orientation and differential growth. Development (Cambridge), 2008, 135, 1427-1437.	2.5	193
35	Molecular Characterization of the Ankle-Link Complex in Cochlear Hair Cells and Its Role in the Hair Bundle Functioning. Journal of Neuroscience, 2007, 27, 6478-6488.	3.6	190
36	Prestin, a cochlear motor protein, is defective in non-syndromic hearing loss. Human Molecular Genetics, 2003, 12, 1155-1162.	2.9	173

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37	Usher I syndrome: unravelling the mechanisms that underlie the cohesion of the growing hair bundle in inner ear sensory cells. Journal of Cell Science, 2005, 118, 4593-4603.	2.0	172
38	MyRIP, a novel Rab effector, enables myosin VIIa recruitment to retinal melanosomes. EMBO Reports, 2002, 3, 463-470.	4.5	171
39	Usherin, the defective protein in Usher syndrome type IIA, is likely to be a component of interstereocilia ankle links in the inner ear sensory cells. Human Molecular Genetics, 2005, 14, 3921-3932.	2.9	166
40	Myosin XVa and whirlin, two deafness gene products required for hair bundle growth, are located at the stereocilia tips and interact directly. Human Molecular Genetics, 2005, 14, 401-410.	2.9	166
41	Transcription Factor SIX5 Is Mutated in Patients with Branchio-Oto-Renal Syndrome. American Journal of Human Genetics, 2007, 80, 800-804.	6.2	164
42	Otoancorin, an inner ear protein restricted to the interface between the apical surface of sensory epithelia and their overlying acellular gels, is defective in autosomal recessive deafness DFNB22. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 6240-6245.	7.1	163
43	Dual AAV-mediated gene therapy restores hearing in a DFNB9 mouse model. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 4496-4501.	7.1	162
44	OTOF Encodes Multiple Long and Short Isoforms: Genetic Evidence That the Long Ones Underlie Recessive Deafness DFNB9. American Journal of Human Genetics, 2000, 67, 591-600.	6.2	160
45	Mutations in a new gene encoding a protein of the hair bundle cause non-syndromic deafness at the DFNB16 locus. Nature Genetics, 2001, 29, 345-349.	21.4	159
46	The Auditory Hair Cell Ribbon Synapse: From Assembly to Function. Annual Review of Neuroscience, 2012, 35, 509-528.	10.7	158
47	Hypervulnerability to Sound Exposure through Impaired Adaptive Proliferation of Peroxisomes. Cell, 2015, 163, 894-906.	28.9	158
48	Linking genes underlying deafness to hair-bundle development and function. Nature Neuroscience, 2009, 12, 703-710.	14.8	156
49	Rab27A and its effector MyRIP link secretory granules to F-actin and control their motion towards release sites. Journal of Cell Biology, 2003, 163, 559-570.	5.2	154
50	Connexin30 deficiency causes instrastrial fluid-blood barrier disruption within the cochlear stria vascularis. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 6229-6234.	7.1	154
51	Cadherin 23 is a component of the transient lateral links in the developing hair bundles of cochlear sensory cells. Developmental Biology, 2005, 280, 281-294.	2.0	151
52	Involvement of the Rab27 Binding Protein Slac2c/MyRIP in Insulin Exocytosis. Molecular Biology of the Cell, 2003, 14, 4103-4113.	2.1	146
53	Localization of Usher 1 proteins to the photoreceptor calyceal processes, which are absent from mice. Journal of Cell Biology, 2012, 199, 381-399.	5.2	145
54	Human Usher 1B/mouse shaker-1: the retinal phenotype discrepancy explained by the presence/absence of myosin VIIA in the photoreceptor cells. Human Molecular Genetics, 1996, 5, 1171-1178.	2.9	144

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55	Anosmin-1 is a regionally restricted component of basement membranes and interstitial matrices during organogenesis: Implications for the developmental anomalies of X chromosome-linked Kallmann syndrome. Developmental Dynamics, 1999, 215, 26-44.	1.8	143
56	Targeted disruption of Otog results in deafness and severe imbalance. Nature Genetics, 2000, 24, 139-143.	21.4	141
57	A human gene responsible for neurosensory, non-syndromic recessive deafness is a candidate homologue of the mouse sh-1 gene. Human Molecular Genetics, 1994, 3, 989-993.	2.9	134
58	A proposed new contiguous gene syndrome on 8q consists of Branchio-Oto-Renal (BOR) syndrome, Duane syndrome, a dominant form of hydrocephalus and trapeze aplasia; implications for the mapping of the BOR gene. Human Molecular Genetics, 1994, 3, 1859-1866.	2.9	121
59	Usher type 1G protein sans is a critical component of the tip-link complex, a structure controlling actin polymerization in stereocilia. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 5825-5830.	7.1	120
60	Structure of the X–linked Kallmann syndrome gene and its homologous pseudogene on the Y chromosome. Nature Genetics, 1992, 2, 305-310.	21.4	115
61	An abnormal terminal X-Y interchange accounts for most but not all cases of human XX maleness. Cell, 1987, 49, 595-602.	28.9	114
62	Stereocilin-deficient mice reveal the origin of cochlear waveform distortions. Nature, 2008, 456, 255-258.	27.8	114
63	Otoferlin Is Critical for a Highly Sensitive and Linear Calcium-Dependent Exocytosis at Vestibular Hair Cell Ribbon Synapses. Journal of Neuroscience, 2009, 29, 10474-10487.	3.6	113
64	Connexin 26 Gene Mutations in Congenitally Deaf Children. JAMA Otolaryngology, 2001, 127, 927.	1.2	111
65	Mutations in the alternatively spliced exons of USH1C cause non-syndromic recessive deafness. Human Genetics, 2002, 111, 26-30.	3.8	111
66	Otoferlin acts as a Ca2+ sensor for vesicle fusion and vesicle pool replenishment at auditory hair cell ribbon synapses. ELife, 2017, 6, .	6.0	108
67	Control of Exocytosis by Synaptotagmins and Otoferlin in Auditory Hair Cells. Journal of Neuroscience, 2010, 30, 13281-13290.	3.6	106
68	Eya1 expression in the developing ear and kidney: Towards the understanding of the pathogenesis of branchio-oto-renal (BOR) syndrome. , 1998, 213, 486-499.		103
69	A newly identified locus for Usher syndrome type I, USH1E, maps to chromosome 21q21. Human Molecular Genetics, 1997, 6, 27-31.	2.9	101
70	Expression of myosin VIIA during mouse embryogenesis. Anatomy and Embryology, 1997, 196, 159-170.	1.5	101
71	Local gene therapy durably restores vestibular function in a mouse model of Usher syndrome type 1G. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 9695-9700.	7.1	101
72	BOR and BO Syndromes Are Allelic Defects of EYA1. European Journal of Human Genetics, 1997, 5, 242-246.	2.8	99

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73	Stereocilin connects outer hair cell stereocilia to one another and to the tectorial membrane. Journal of Comparative Neurology, 2011, 519, 194-210.	1.6	98
74	Clarin-1 gene transfer rescues auditory synaptopathy in model of Usher syndrome. Journal of Clinical Investigation, 2018, 128, 3382-3401.	8.2	97
75	Cadherin-23, myosin VIIa and harmonin, encoded by Usher syndrome type I genes, form a ternary complex and interact with membrane phospholipids. Human Molecular Genetics, 2010, 19, 3557-3565.	2.9	94
76	Probing the Functional Equivalence of Otoferlin and Synaptotagmin 1 in Exocytosis. Journal of Neuroscience, 2011, 31, 4886-4895.	3.6	94
77	A gene responsible for a dominant form of neurosensory non-syndromic deafness maps to the NSRD1 recessive deafness gene interval. Human Molecular Genetics, 1994, 3, 2219-2222.	2.9	93
78	GJB2 and GJB6 Mutations. JAMA Otolaryngology, 2005, 131, 481.	1.2	93
79	Complete exon sequencing of all known Usher syndrome genes greatly improves molecular diagnosis. Orphanet Journal of Rare Diseases, 2011, 6, 21.	2.7	93
80	Mutations in apoptosis-inducing factor cause X-linked recessive auditory neuropathy spectrum disorder. Journal of Medical Genetics, 2015, 52, 523-531.	3.2	92
81	Neurogenetics. Current Opinion in Neurobiology, 2013, 23, 1-2.	4.2	86
82	Whole mitochondrial genome screening in maternally inherited non-syndromic hearing impairment using a microarray resequencing mitochondrial DNA chip. European Journal of Human Genetics, 2007, 15, 1145-1155.	2.8	85
83	Auditory Distortions: Origins and Functions. Physiological Reviews, 2013, 93, 1563-1619.	28.8	84
84	Myosin VI is required for the proper maturation and function of inner hair cell ribbon synapses. Human Molecular Genetics, 2009, 18, 4615-4628.	2.9	81
85	Calcium- and Otoferlin-Dependent Exocytosis by Immature Outer Hair Cells. Journal of Neuroscience, 2008, 28, 1798-1803.	3.6	80
86	Novel gene function revealed by mouse mutagenesis screens for models of age-related disease. Nature Communications, 2016, 7, 12444.	12.8	79
87	A novel locus for Usher syndrome type II, USH2B, maps to chromosome 3 at p23–24.2. European Journal of Human Genetics, 1999, 7, 363-367.	2.8	77
88	Harmonin-b, an actin-binding scaffold protein, is involved in the adaptation of mechanoelectrical transduction by sensory hair cells. Pflugers Archiv European Journal of Physiology, 2009, 459, 115-130.	2.8	77
89	An innovative strategy for the molecular diagnosis of Usher syndrome identifies causal biallelic mutations in 93% of European patients. European Journal of Human Genetics, 2016, 24, 1730-1738.	2.8	77
90	Differential Distribution of Harmonin Isoforms and Their Possible Role in Usher-1 Protein Complexes in Mammalian Photoreceptor Cells. , 2003, 44, 5006.		76

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91	Unconventional myosin VIIa and vezatin, two proteins crucial forListeriaentry into epithelial cells. Journal of Cell Science, 2004, 117, 2121-2130.	2.0	75
92	Temperature-sensitive auditory neuropathy associated with an otoferlin mutation: Deafening fever!. Biochemical and Biophysical Research Communications, 2010, 394, 737-742.	2.1	74
93	From deafness genes to hearing mechanisms: harmony and counterpoint. Trends in Molecular Medicine, 2006, 12, 57-64.	6.7	73
94	Mapping of DFNB12, a gene for a non-syndromal autosomal recessive deafness, to chromosome 10q21-22. Human Molecular Genetics, 1996, 5, 1061-1064.	2.9	72
95	Myosin-I nomenclature. Journal of Cell Biology, 2001, 155, 703-704.	5.2	71
96	Expression of the connexin43- and connexin45-encoding genes in the developing and mature mouse inner ear. Cell and Tissue Research, 2004, 316, 15-22.	2.9	68
97	Chapter 8 Mouse Models for Human Hereditary Deafness. Current Topics in Developmental Biology, 2008, 84, 385-429.	2.2	68
98	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. EMBO Molecular Medicine, 2017, 9, 1711-1731.	6.9	66
99	Hearing Is Normal without Connexin30. Journal of Neuroscience, 2013, 33, 430-434.	3.6	65
100	Characterisation of DRASIC in the mouse inner ear. Hearing Research, 2004, 190, 149-160.	2.0	64
101	A novel locus for Usher syndrome typeÂl, USH1G, maps to chromosome 17q24–25. Human Genetics, 2002, 110, 348-350.	3.8	63
102	Pejvakin-mediated pexophagy protects auditory hair cells against noise-induced damage. Proceedings of the United States of America, 2019, 116, 8010-8017.	7.1	63
103	αII-βV spectrin bridges the plasma membrane and cortical lattice in the lateral wall of the auditory outer hair cells. Journal of Cell Science, 2008, 121, 3347-3356.	2.0	62
104	The <scp>CD</scp> 2 isoform of protocadherinâ€15 is an essential component of the tipâ€link complex in mature auditory hair cells. EMBO Molecular Medicine, 2014, 6, 984-992.	6.9	62
105	Cloning of the Genes Encoding Two Murine and Human Cochlear Unconventional Type I Myosins. Genomics, 1997, 40, 332-341.	2.9	61
106	Townes-Brocks syndrome: Detection of aSALL1 mutation hot spot and evidence for a position effect in one patient. Human Mutation, 1999, 14, 377-386.	2.5	61
107	Shroom2, a myosin-VIIa- and actin-binding protein, directly interacts with ZO-1 at tight junctions. Journal of Cell Science, 2007, 120, 2838-2850.	2.0	60
108	A specific promoter of the sensory cells of the inner ear defined by transgenesis. Human Molecular Genetics, 2001, 10, 1581-1589.	2.9	59

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109	Highly homologous loci on the X and Y chromosomes are hot–spots for ectopic recombinations leading to XX maleness. Nature Genetics, 1994, 7, 414-419.	21.4	58
110	A mouse model for human deafness DFNB22 reveals that hearing impairment is due to a loss of inner hair cell stimulation. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 19351-19356.	7.1	57
111	A Human Pseudoautosomal Gene Encodes the ANT3 ADP/ATP Translocase and Escapes X-Inactivation. Genomics, 1993, 16, 26-33.	2.9	55
112	Unconventional Myosin VIIA Is a Novel A-kinase-anchoring Protein. Journal of Biological Chemistry, 2000, 275, 29654-29659.	3.4	55
113	Deafness heterogeneity in a Druze isolate from the Middle East: novel OTOF and PDS mutations, low prevalence of GJB2 35delG mutation and indication for a new DFNB locus. European Journal of Human Genetics, 2000, 8, 437-442.	2.8	54
114	Mechanotransduction is required for establishing and maintaining mature inner hair cells and regulating efferent innervation. Nature Communications, 2018, 9, 4015.	12.8	54
115	Spatiotemporal expression of otogelin in the developing and adult mouse inner ear. Hearing Research, 2001, 158, 151-159.	2.0	53
116	Defect in the gene encoding the EAR/EPTP domain-containing protein TSPEAR causes DFNB98 profound deafness. Human Molecular Genetics, 2012, 21, 3835-3844.	2.9	53
117	Class III myosins shape the auditory hair bundles by limiting microvilli and stereocilia growth. Journal of Cell Biology, 2016, 212, 231-244.	5.2	51
118	Two large French pedigrees with non syndromic sensorineural deafness and the mitochondrial DNA T7511C mutation: evidence for a modulatory factor. European Journal of Human Genetics, 2002, 10, 851-856.	2.8	50
119	Exocytotic Machineries of Vestibular Type I and Cochlear Ribbon Synapses Display Similar Intrinsic Otoferlin-Dependent Ca2+ Sensitivity But a Different Coupling to Ca2+ Channels. Journal of Neuroscience, 2014, 34, 10853-10869.	3.6	50
120	From DFNB2 to Usher syndrome: Variable expressivity of the same disease. American Journal of Medical Genetics Part A, 2001, 101, 181-183.	2.4	49
121	Cadherins as Targets for Genetic Diseases. Cold Spring Harbor Perspectives in Biology, 2010, 2, a003095-a003095.	5.5	49
122	Hair-Bundle Links: Genetics as the Gateway to Function. Cold Spring Harbor Perspectives in Medicine, 2019, 9, a033142.	6.2	49
123	Clinical evidence of the nonpathogenic nature of the M34T variant in the connexin 26 gene. European Journal of Human Genetics, 2004, 12, 279-284.	2.8	48
124	The giant spectrin βV couples the molecular motors to phototransduction and Usher syndrome type I proteins along their trafficking route. Human Molecular Genetics, 2013, 22, 3773-3788.	2.9	48
125	Usher syndrome type 1–associated cadherins shape the photoreceptor outer segment. Journal of Cell Biology, 2017, 216, 1849-1864.	5.2	47
126	Exome Sequencing and Linkage Analysis Identified Tenascin-C (TNC) as a Novel Causative Gene in Nonsyndromic Hearing Loss. PLoS ONE, 2013, 8, e69549.	2.5	46

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127	Screening mutations of OTOFgene in Chinese patients with auditory neuropathy, including a familial case of temperature-sensitive auditory neuropathy. BMC Medical Genetics, 2010, 11, 79.	2.1	45
128	Fundus autofluorescence and optical coherence tomography in relation to visual function in Usher syndrome type 1 and 2. Vision Research, 2012, 75, 60-70.	1.4	45
129	The retinal phenotype of Usher syndrome: Pathophysiological insights from animal models. Comptes Rendus - Biologies, 2014, 337, 167-177.	0.2	44
130	Characterization of the two zebrafish orthologues of the KAL-1 gene underlying X chromosome-linked Kallmann syndrome. Mechanisms of Development, 2000, 90, 89-94.	1.7	43
131	EPS8, encoding an actin-binding protein of cochlear hair cell stereocilia, is a new causal gene for autosomal recessive profound deafness. Orphanet Journal of Rare Diseases, 2014, 9, 55.	2.7	43
132	Characterization of the Chicken and Quail Homologues of the Human Gene Responsible for the X-Linked Kallmann Syndrome. Genomics, 1993, 17, 516-518.	2.9	42
133	Twister mutant mice are defective for otogelin, a component specific to inner ear acellular membranes. Mammalian Genome, 2000, 11, 961-966.	2.2	42
134	Large deletion of theGJB6gene in deaf patients heterozygous for theGJB2gene mutation: Genotypic and phenotypic analysis. , 2004, 127A, 263-267.		42
135	Identification of three novel mutations in the USH1C gene and detection of thirty-one polymorphisms used for haplotype analysis. Human Mutation, 2001, 17, 34-41.	2.5	41
136	Cochlear outer hair cells undergo an apical circumference remodeling constrained by the hair bundle shape. Development (Cambridge), 2010, 137, 1373-1383.	2.5	41
137	Diversity of the causal genes in hearing impaired Algerian individuals identified by whole exome sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 189-196.	1.2	40
138	A 45,X male with an X;Y translocation: implications for the mapping of the genes responsible for the mapping of the genes responsible for Turner syndrome and X-linked chondrodysplasia punctata. Human Molecular Genetics, 1993, 2, 1853-1856.	2.9	39
139	Vezatin, an integral membrane protein of adherens junctions, is required for the sound resilience of cochlear hair cells. EMBO Molecular Medicine, 2009, 1, 125-138.	6.9	39
140	An interstitial deletion in Xp22.3 in a family with X-linked recessive chondrodysplasia punctata and short stature. Human Genetics, 1990, 85, 247-50.	3.8	38
141	A sensorineural progressive autosomal recessive form of isolated deafness, DFNB13, maps to chromosome 7q34-q36. European Journal of Human Genetics, 1998, 6, 245-250.	2.8	38
142	CLINICAL PRESENTATION AND DISEASE COURSE OF USHER SYNDROME BECAUSE OF MUTATIONS IN MYO7A OR USH2A. Retina, 2017, 37, 1581-1590.	1.7	36
143	Consortin, a trans-Golgi network cargo receptor for the plasma membrane targeting and recycling of connexins. Human Molecular Genetics, 2010, 19, 262-275.	2.9	35
144	Mutations in CDC14A , Encoding a Protein Phosphatase Involved in Hair Cell Ciliogenesis, Cause Autosomal-Recessive Severe to Profound Deafness. American Journal of Human Genetics, 2016, 98, 1266-1270.	6.2	35

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145	Auditory cortex interneuron development requires cadherins operating hair-cell mechanoelectrical transduction. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 7765-7774.	7.1	35
146	Otogelin, otogelin-like, and stereocilin form links connecting outer hair cell stereocilia to each other and the tectorial membrane. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 25948-25957.	7.1	35
147	Different Ca <sub>V</sub> 1.3 Channel Isoforms Control Distinct Components of the Synaptic Vesicle Cycle in Auditory Inner Hair Cells. Journal of Neuroscience, 2017, 37, 2960-2975.	3.6	34
148	DFNB31, a recessive form of sensorineural hearing loss, maps to chromosome 9q32-34. European Journal of Human Genetics, 2002, 10, 210-212.	2.8	33
149	PHR1, an integral membrane protein of the inner ear sensory cells, directly interacts with myosin 1c and myosin VIIa. Journal of Cell Science, 2005, 118, 2891-2899.	2.0	33
150	The tip-link molecular complex of the auditory mechano-electrical transduction machinery. Hearing Research, 2015, 330, 10-17.	2.0	33
151	Genes Involved in the Development and Physiology of Both the Peripheral and Central Auditory Systems. Annual Review of Neuroscience, 2019, 42, 67-86.	10.7	33
152	Cells of adult brain germinal zone have properties akin to hair cells and can be used to replace inner ear sensory cells after damage. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 21000-21005.	7.1	32
153	Down-expression of <i>P2RX2</i> , <i>KCNQ5</i> , <i>ERBB3</i> and <i>SOCS3</i> through DNA hypermethylation in elderly women with presbycusis. Biomarkers, 2018, 23, 347-356.	1.9	31
154	Fdp, a New Fibrocyte-derived Protein Related toMIA/CD-RAP, Has an in VitroEffect on the Early Differentiation of the Inner Ear Mesenchyme. Journal of Biological Chemistry, 2000, 275, 40036-40041.	3.4	30
155	Vezatin, a protein associated to adherens junctions, is required for mouse blastocyst morphogenesis. Developmental Biology, 2005, 287, 180-191.	2.0	30
156	Spectrin βV adaptive mutations and changes in subcellular location correlate with emergence of hair cell electromotility in mammalians. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 2054-2059.	7.1	29
157	Cone degeneration is triggered by the absence of USH1 proteins but prevented by antioxidant treatments. Scientific Reports, 2018, 8, 1968.	3.3	29
158	Ultrarare heterozygous pathogenic variants of genes causing dominant forms of early-onset deafness underlie severe presbycusis. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 31278-31289.	7.1	29
159	Characterization and Chromosomal Assignment of a Human cDNA Encoding a Protein Related to the Murine 102-kDa Cadherin-Associated Protein (α-Catenin). Genomics, 1993, 15, 13-20.	2.9	28
160	A Subtracted cDNA Library from the Zebrafish (Danio rerio) Embryonic Inner Ear. Genome Research, 2002, 12, 1007-1011.	5.5	28
161	Non-syndromic recessive deafness in Jordan: mapping of a new locus to chromosome 9q34.3 and prevalence of DFNB1 mutations. European Journal of Human Genetics, 2002, 10, 391-394.	2.8	28
162	Anosmin-1 immunoreactivity during embryogenesis in a primitive eutherian mammal. Developmental Brain Research, 2003, 140, 157-167.	1.7	28

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163	Coupling of the mechanotransduction machinery and stereocilia F-actin polymerization in the cochlear hair bundles. Bioarchitecture, 2011, 1, 169-174.	1.5	28
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