Richard J H Smith

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	From The Cover: A common haplotype in the complement regulatory gene factor H (HF1/CFH) predisposes individuals to age-related macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 7227-7232.	3.3	1,867
2	Sensorineural hearing loss in children. Lancet, The, 2005, 365, 879-890.	6.3	641
3	Usher Syndrome 1D and Nonsyndromic Autosomal Recessive Deafness DFNB12 Are Caused by Allelic Mutations of the Novel Cadherin-Like Gene CDH23. American Journal of Human Genetics, 2001, 68, 26-37.	2.6	549
4	C3 glomerulopathy: consensus report. Kidney International, 2013, 84, 1079-1089.	2.6	505
5	A candidate prostate cancer susceptibility gene at chromosome 17p. Nature Genetics, 2001, 27, 172-180.	9.4	504
6	GJB2 Mutations and Degree of Hearing Loss: A Multicenter Study. American Journal of Human Genetics, 2005, 77, 945-957.	2.6	455
7	Forty-six genes causing nonsyndromic hearing impairment: Which ones should be analyzed in DNA diagnostics?. Mutation Research - Reviews in Mutation Research, 2009, 681, 189-196.	2.4	386
8	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.	3.3	374
9	Comprehensive genetic testing in the clinical evaluation of 1119 patients with hearing loss. Human Genetics, 2016, 135, 441-450.	1.8	373
10	Membranoproliferative Glomerulonephritis Type II (Dense Deposit Disease): An Update. Journal of the American Society of Nephrology: JASN, 2005, 16, 1392-1403.	3.0	354
11	Congenital hearing loss. Nature Reviews Disease Primers, 2017, 3, 16094.	18.1	328
12	Nonsyndromic hearing impairment is associated with a mutation in DFNA5. Nature Genetics, 1998, 20, 194-197.	9.4	323
13	Mutations in the human α-tectorin gene cause autosomal dominant non-syndromic hearing impairment. Nature Genetics, 1998, 19, 60-62.	9.4	323
14	The 1000 Genomes Project: data management and community access. Nature Methods, 2012, 9, 459-462.	9.0	308
15	Eculizumab for Dense Deposit Disease and C3 Glomerulonephritis. Clinical Journal of the American Society of Nephrology: CJASN, 2012, 7, 748-756.	2.2	295
16	Comprehensive genetic testing for hereditary hearing loss using massively parallel sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 21104-21109.	3.3	294
17	Mutations in COL11A2 cause non-syndromic hearing loss (DFNA13). Nature Genetics, 1999, 23, 413-419.	9.4	285
18	Laryngomalacia and Its Treatment. Laryngoscope, 1999, 109, 1770-1775.	1.1	273

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19	Localization of Usher syndrome type II to chromosome 1q. Genomics, 1990, 7, 245-249.	1.3	272
20	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.	2.6	269
21	Mutations in alternative pathway complement proteins in American patients with atypical hemolytic uremic syndrome. Human Mutation, 2010, 31, E1445-E1460.	1.1	268
22	Pendred syndrome, DFNB4, andPDS/SLC26A4 identification of eight novel mutations and possible genotype-phenotype correlations. Human Mutation, 2001, 17, 403-411.	1.1	267
23	C3 glomerulonephritis: clinicopathological findings, complement abnormalities, glomerular proteomic profile, treatment, and follow-up. Kidney International, 2012, 82, 465-473.	2.6	264
24	Impairment of SLC17A8 Encoding Vesicular Glutamate Transporter-3, VGLUT3, Underlies Nonsyndromic Deafness DFNA25 and Inner Hair Cell Dysfunction in Null Mice. American Journal of Human Genetics, 2008, 83, 278-292.	2.6	237
25	Atypical aHUS: State of the art. Molecular Immunology, 2015, 67, 31-42.	1.0	236
26	New Approaches to the Treatment of Dense Deposit Disease. Journal of the American Society of Nephrology: JASN, 2007, 18, 2447-2456.	3.0	231
27	C3 glomerulopathy — understanding a rare complement-driven renal disease. Nature Reviews Nephrology, 2019, 15, 129-143.	4.1	223
28	Localization of two genes for usher syndrome type I to chromosome 11. Genomics, 1992, 14, 995-1002.	1.3	216
29	Genomic Landscape and Mutational Signatures of Deafness-Associated Genes. American Journal of Human Genetics, 2018, 103, 484-497.	2.6	214
30	Mutations in the transcriptional activator EYA4 cause late-onset deafness at the DFNA10 locus. Human Molecular Genetics, 2001, 10, 195-200.	1.4	210
31	Mayo Clinic/Renal Pathology Society Consensus Report on Pathologic Classification, Diagnosis, and Reporting of GN. Journal of the American Society of Nephrology: JASN, 2016, 27, 1278-1287.	3.0	210
32	Human Male Infertility Caused by Mutations in the CATSPER1 Channel Protein. American Journal of Human Genetics, 2009, 84, 505-510.	2.6	206
33	Comprehensive Genetic Analysis of Complement and Coagulation Genes in Atypical Hemolytic Uremic Syndrome. Journal of the American Society of Nephrology: JASN, 2014, 25, 55-64.	3.0	201
34	Treatment of Lymphangiomas With OK-432 (Picibanil) Sclerotherapy. JAMA Otolaryngology, 2002, 128, 1137.	1.5	199
35	Frequency of Usher syndrome in two pediatric populations: Implications for genetic screening of deaf and hard of hearing children. Genetics in Medicine, 2010, 12, 512-516.	1.1	198
36	Phenotypic manifestations of branchiootorenal syndrome. American Journal of Medical Genetics Part A, 1995, 58, 365-370.	2.4	195

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37	Branchio-oto-renal syndrome: The mutation spectrum inEYA1and its phenotypic consequences. Human Mutation, 2004, 23, 582-589.	1.1	194
38	Transcriptional Control of SLC26A4 Is Involved in Pendred Syndrome and Nonsyndromic Enlargement of Vestibular Aqueduct (DFNB4). American Journal of Human Genetics, 2007, 80, 1055-1063.	2.6	184
39	Defective complement control of Factor H (Y402H) and FHL-1 in age-related macular degeneration. Molecular Immunology, 2007, 44, 3398-3406.	1.0	181
40	Impact of Tympanostomy Tubes on Child Quality of Life. JAMA Otolaryngology, 2000, 126, 585.	1.5	180
41	Glomeruli of Dense Deposit Disease contain components of the alternative and terminal complement pathway. Kidney International, 2009, 75, 952-960.	2.6	178
42	Membranoproliferative glomerulonephritis and C3 glomerulopathy: resolving the confusion. Kidney International, 2012, 81, 434-441.	2.6	175
43	Prestin, a cochlear motor protein, is defective in non-syndromic hearing loss. Human Molecular Genetics, 2003, 12, 1155-1162.	1.4	173
44	The functional spectrum of low-frequency coding variation. Genome Biology, 2011, 12, R84.	13.9	173
45	Genomic Structure and Identification of Novel Mutations in Usherin, the Gene Responsible for Usher Syndrome Type IIa. American Journal of Human Genetics, 2000, 66, 1199-1210.	2.6	168
46	Causes of Alternative Pathway Dysregulation in Dense Deposit Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2012, 7, 265-274.	2.2	166
47	Transcription Factor SIX5 Is Mutated in Patients with Branchio-Oto-Renal Syndrome. American Journal of Human Genetics, 2007, 80, 800-804.	2.6	164
48	Atypical postinfectious glomerulonephritis is associated with abnormalities in the alternative pathway of complement. Kidney International, 2013, 83, 293-299.	2.6	161
49	Mutational spectrum of theWFS1 gene in Wolfram syndrome, nonsyndromic hearing impairment, diabetes mellitus, and psychiatric disease. Human Mutation, 2003, 22, 275-287.	1.1	160
50	A Forward Genetics Screen in Mice Identifies Recessive Deafness Traits and Reveals That Pejvakin Is Essential for Outer Hair Cell Function. Journal of Neuroscience, 2007, 27, 2163-2175.	1.7	159
51	Membranoproliferative Glomerulonephritis Secondary to Monoclonal Gammopathy. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 770-782.	2.2	156
52	Function and Expression Pattern of Nonsyndromic Deafness Genes. Current Molecular Medicine, 2009, 9, 546-564.	0.6	151
53	C3 Glomerulonephritis Associated With Monoclonal Gammopathy: A Case Series. American Journal of Kidney Diseases, 2013, 62, 506-514.	2.1	150
54	Incidence of vocal fold paralysis in infants undergoing ligation of patent ductus arteriosus. Annals of Thoracic Surgery, 1996, 61, 814-816.	0.7	146

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55	Mutations of KCNJ10 Together with Mutations of SLC26A4 Cause Digenic Nonsyndromic Hearing Loss Associated with Enlarged Vestibular Aqueduct Syndrome. American Journal of Human Genetics, 2009, 84, 651-657.	2.6	144
56	Functional differences of the PDS gene product are associated with phenotypic variation in patients with Pendred syndrome and non-syndromic hearing loss (DFNB4). Human Molecular Genetics, 2000, 9, 1709-1715.	1.4	139
57	Genetic male infertility and mutation of CATSPER ion channels. European Journal of Human Genetics, 2010, 18, 1178-1184.	1.4	139
58	New Treatment Options for Lymphangioma in Infants and Children. Annals of Otology, Rhinology and Laryngology, 2002, 111, 1066-1075.	0.6	137
59	Copy number variants are a common cause of non-syndromic hearing loss. Genome Medicine, 2014, 6, 37.	3.6	137
60	Utilizing Ethnic-Specific Differences in Minor Allele Frequency to Recategorize Reported Pathogenic Deafness Variants. American Journal of Human Genetics, 2014, 95, 445-453.	2.6	137
61	Insights into the Biology of Hearing and Deafness Revealed by Single-Cell RNA Sequencing. Cell Reports, 2019, 26, 3160-3171.e3.	2.9	137
62	An Update on the Treatment of Hemangiomas in Children With Interferon Alfa-2a. JAMA Otolaryngology, 1999, 125, 21.	1.5	134
63	Efficacy and safety of OKâ€432 immunotherapy of lymphatic malformations. Laryngoscope, 2009, 119, 107-115.	1.1	134
64	Branchio-oto-renal syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1671-1678.	0.7	133
65	Proliferative Glomerulonephritis Secondary to Dysfunction of the Alternative Pathway of Complement. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 1009-1017.	2.2	133
66	Statistical Validation of Rare Complement Variants Provides Insights into the Molecular Basis of Atypical Hemolytic Uremic Syndrome and C3 Glomerulopathy. Journal of Immunology, 2018, 200, 2464-2478.	0.4	130
67	Mutations in LOXHD1, an Evolutionarily Conserved Stereociliary Protein, Disrupt Hair Cell Function in Mice and Cause Progressive Hearing Loss in Humans. American Journal of Human Genetics, 2009, 85, 328-337.	2.6	129
68	Cystic Fibrosis—An Otolaryngologic Perspective. Otolaryngology - Head and Neck Surgery, 1987, 97, 356-360.	1.1	128
69	Parental Attitudes toward Genetic Testing for Pediatric Deafness. American Journal of Human Genetics, 2000, 67, 1621-1625.	2.6	126
70	Clinical Findings, Pathology, and Outcomes of C3GN after Kidney Transplantation. Journal of the American Society of Nephrology: JASN, 2014, 25, 1110-1117.	3.0	126
71	A Classification Scheme for Paradoxical Vocal Cord Motion. Laryngoscope, 1997, 107, 1429-1435.	1.1	124
72	Carcinoembryonic antigen-related cell adhesion molecule 16 interacts with α-tectorin and is mutated in autosomal dominant hearing loss (DFNA4). Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 4218-4223.	3.3	123

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73	Spastic Diplegia and Other Motor Disturbances in Infants Receiving Interferon-Alpha. Laryngoscope, 2004, 114, 1231-1236.	1.1	122
74	Mutations in a Novel Gene, TMIE, Are Associated with Hearing Loss Linked to the DFNB6 Locus. American Journal of Human Genetics, 2002, 71, 632-636.	2.6	120
75	Clinical aspects of hereditary hearing loss. Genetics in Medicine, 2007, 9, 393-408.	1.1	120
76	Tracheal allograft reconstruction: the total North American and worldwide pediatric experiences. Annals of Thoracic Surgery, 1999, 68, 1043-1051.	0.7	119
77	Small-molecule factor B inhibitor for the treatment of complement-mediated diseases. Proceedings of the United States of America, 2019, 116, 7926-7931.	3.3	116
78	Massively Parallel Sequencing for Genetic Diagnosis of Hearing Loss. Otolaryngology - Head and Neck Surgery, 2015, 153, 175-182.	1.1	113
79	In vitro and in vivo suppression of GJB2 expression by RNA interference. Human Molecular Genetics, 2005, 14, 1641-1650.	1.4	112
80	Mapping interactions between complement C3 and regulators using mutations in atypical hemolytic uremic syndrome. Blood, 2015, 125, 2359-2369.	0.6	112
81	Pre-emptive Eculizumab and Plasmapheresis for Renal Transplant in Atypical Hemolytic Uremic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 1488-1494.	2.2	111
82	Recent advances in the molecular genetics of epilepsy. Journal of Medical Genetics, 2013, 50, 271-279.	1.5	111
83	Conductive Hearing Loss and Otopathology in Cleft Palate Patients. Otolaryngology - Head and Neck Surgery, 2006, 134, 946-948.	1.1	108
84	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	2.6	108
85	Treatment of Lymphangiomas in Children: An Update of Picibanil (OK-432) Sclerotherapy. Otolaryngology - Head and Neck Surgery, 1999, 121, 381-387.	1.1	107
86	Dense Deposit Disease Associated With Monoclonal Gammopathy of Undetermined Significance. American Journal of Kidney Diseases, 2010, 56, 977-982.	2.1	107
87	Complement Factor B Mutations in Atypical Hemolytic Uremic Syndrome—Disease-Relevant or Benign?. Journal of the American Society of Nephrology: JASN, 2014, 25, 2053-2065.	3.0	107
88	Prediction of cochlear implant performance by genetic mutation: The spiral ganglion hypothesis. Hearing Research, 2012, 292, 51-58.	0.9	104
89	Advancing genetic testing for deafness with genomic technology. Journal of Medical Genetics, 2013, 50, 627-634.	1.5	104
90	Temporal bone analysis of patients with presbycusis reveals high frequency of mitochondrial mutations. Hearing Research, 1997, 110, 147-154.	0.9	102

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91	A Claudin-9–Based Ion Permeability Barrier Is Essential for Hearing. PLoS Genetics, 2009, 5, e1000610.	1.5	102
92	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	13.9	101
93	Connexin mutations and hearing loss. Nature, 1998, 391, 32-32.	13.7	98
94	Inactivation of NADPH oxidase organizer 1 Results in Severe Imbalance. Current Biology, 2006, 16, 208-213.	1.8	98
95	Sensorineural deafness and male infertility: a contiguous gene deletion syndrome. Journal of Medical Genetics, 2007, 44, 233-240.	1.5	98
96	Genotype–phenotype correlations for SLC26A4-related deafness. Human Genetics, 2007, 122, 451-457.	1.8	97
97	Gipc3 mutations associated with audiogenic seizures and sensorineural hearing loss in mouse and human. Nature Communications, 2011, 2, 201.	5.8	95
98	RNA Interference Prevents Autosomal-Dominant Hearing Loss. American Journal of Human Genetics, 2016, 98, 1101-1113.	2.6	95
99	Mutations in apoptosis-inducing factor cause X-linked recessive auditory neuropathy spectrum disorder. Journal of Medical Genetics, 2015, 52, 523-531.	1.5	92
100	Enhanced viral-mediated cochlear gene delivery in adult mice by combining canal fenestration with round window membrane inoculation. Scientific Reports, 2018, 8, 2980.	1.6	92
101	A Second Gene for Otosclerosis, OTSC2, Maps to Chromosome 7q34-36. American Journal of Human Genetics, 2001, 68, 495-500.	2.6	91
102	Allelic Variants of Complement Genes Associated with Dense Deposit Disease. Journal of the American Society of Nephrology: JASN, 2011, 22, 1551-1559.	3.0	90
103	Genetics. Current Opinion in Pediatrics, 2012, 24, 679-686.	1.0	89
104	High-Throughput Genetic Testing for Thrombotic Microangiopathies and C3 Glomerulopathies. Journal of the American Society of Nephrology: JASN, 2016, 27, 1245-1253.	3.0	89
105	International Pediatric Otolaryngology Group (IPOG) consensus recommendations: Hearing loss in the pediatric patient. International Journal of Pediatric Otorhinolaryngology, 2016, 90, 251-258.	0.4	88
106	Normal hearing in Splotch (Sp/+), the mouse homologue of Waardenburg syndrome type 1. Nature Genetics, 1992, 2, 75-79.	9.4	87
107	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. Journal of Medical Genetics, 2015, 52, 823-829.	1.5	87
108	Linkage of a gene for dominant non-syndromic deafness to chromosome 19. Human Molecular Genetics, 1995, 4, 1073-1076.	1.4	86

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109	The WFS1 gene, responsible for low frequency sensorineural hearing loss and Wolfram syndrome, is expressed in a variety of inner ear cells. Histochemistry and Cell Biology, 2003, 119, 247-256.	0.8	85
110	Cloning and Characterization of SLC26A6, a Novel Member of the Solute Carrier 26 Gene Family. Genomics, 2001, 72, 43-50.	1.3	84
111	A gene for autosomal dominant nonsyndromic hereditary hearing impairment maps to 4p16.3. Human Molecular Genetics, 1995, 4, 1967-1972.	1.4	83
112	Eculizumab and recurrent C3 glomerulonephritis. Pediatric Nephrology, 2013, 28, 1975-1981.	0.9	82
113	C3 Glomerulopathy: Ten Years' Experience at Mayo Clinic. Mayo Clinic Proceedings, 2018, 93, 991-1008.	1.4	82
114	Mutations in the WFS1 gene that cause low-frequency sensorineural hearing loss are small non-inactivating mutations. Human Genetics, 2002, 110, 389-394.	1.8	81
115	<i>SIX1</i> mutation screening in 247 branchio-oto-renal syndrome families: a recurrent missense mutation associated with BOR. Human Mutation, 2008, 29, 565-565.	1.1	81
116	Temporal Bone Histopathology in Connexin 26-Related Hearing Loss. Laryngoscope, 2000, 110, 269-269.	1.1	80
117	Dense deposit disease. Molecular Immunology, 2011, 48, 1604-1610.	1.0	80
118	Soluble CR1 Therapy Improves Complement Regulation in C3 Glomerulopathy. Journal of the American Society of Nephrology: JASN, 2013, 24, 1820-1829.	3.0	80
119	Branchio-oto-renal syndrome (BOR): novel mutations in the <i>EYA1</i> gene, and a review of the mutational genetics of BOR. Human Mutation, 2008, 29, 537-544.	1.1	79
120	Pelvic Tilt. Physical Therapy, 1985, 65, 169-174.	1.1	78
121	RECURRENT RESPIRATORY PAPILLOMATOSIS. Pediatric Clinics of North America, 1996, 43, 1385-1401.	0.9	78
122	Performance of cochlear implant recipients withGJB2-related deafness. American Journal of Medical Genetics Part A, 2002, 109, 167-170.	2.4	78
123	<i>TBC1D24</i> Mutation Causes Autosomal-Dominant Nonsyndromic Hearing Loss. Human Mutation, 2014, 35, 819-823.	1.1	78
124	The role of complement in C3 glomerulopathy. Molecular Immunology, 2015, 67, 21-30.	1.0	78
125	Gene therapy for hearing loss. Human Molecular Genetics, 2019, 28, R65-R79.	1.4	78
126	The Epidemiology of Deafness. Cold Spring Harbor Perspectives in Medicine, 2019, 9, a033258.	2.9	78

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127	Pediatric Exercise-Induced Laryngomalacia. Annals of Otology, Rhinology and Laryngology, 1996, 105, 169-175.	0.6	77
128	GJB2 mutations: Passage through Iran. American Journal of Medical Genetics, Part A, 2005, 133A, 132-137.	0.7	77
129	Mutations of theRDXgene cause nonsyndromic hearing loss at theDFNB24locus. Human Mutation, 2007, 28, 417-423.	1.1	77
130	A Mutation in the Srrm4 Gene Causes Alternative Splicing Defects and Deafness in the Bronx Waltzer Mouse. PLoS Genetics, 2012, 8, e1002966.	1.5	77
131	Clinical features and outcomes of 98 children and adults with dense deposit disease. Pediatric Nephrology, 2012, 27, 773-781.	0.9	77
132	C3 glomerulopathy associated with monoclonal IgÂis a distinct subtype. Kidney International, 2018, 94, 178-186.	2.6	77
133	Polymerase Chain Reaction Amplification of Herpes Simplex Viral Dna from the Geniculate Ganglion of a Patient with Bell's Palsy. Annals of Otology, Rhinology and Laryngology, 1994, 103, 775-779.	0.6	75
134	The mitochondrial TIM22 preprotein translocase is highly conserved throughout the eukaryotic kingdom. FEBS Letters, 1999, 464, 41-47.	1.3	75
135	Maternally inherited hearing impairment. Clinical Genetics, 2000, 57, 409-414.	1.0	75
136	The coding polymorphism T263I in TGF-β1 is associated with otosclerosis in two independent populations. Human Molecular Genetics, 2007, 16, 2021-2030.	1.4	75
137	Deafness in the genomics era. Hearing Research, 2011, 282, 1-9.	0.9	74
138	Diagnosis of complement alternative pathway disorders. Kidney International, 2016, 89, 278-288.	2.6	74
139	Congenital Oral Synechiae. Annals of Otology, Rhinology and Laryngology, 1993, 102, 186-197.	0.6	73
140	The Effect of GJB2 Allele Variants on Performance After Cochlear Implantation. Laryngoscope, 2003, 113, 2135-2140.	1.1	73
141	DFNA8/12 caused by TECTA mutations is the most identified subtype of nonsyndromic autosomal dominant hearing loss. Human Mutation, 2011, 32, 825-834.	1.1	73
142	Current management of laryngeal and laryngotracheoesophageal clefts. Journal of Pediatric Surgery, 1990, 25, 855-860.	0.8	72
143	GJB2: The spectrum of deafness-causing allele variants and their phenotype. Human Mutation, 2004, 24, 305-311.	1.1	72
144	HspE7 Treatment of Pediatric Recurrent Respiratory Papillomatosis: Final Results of an Open-Label Trial. Annals of Otology, Rhinology and Laryngology, 2005, 114, 730-737.	0.6	72

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145	Causes of Facial Swelling in Pediatric Patients: Correlation of Clinical and Radiologic Findings. Radiographics, 2006, 26, 157-171.	1.4	72
146	Reducing the exome search space for Mendelian diseases using genetic linkage analysis of exome genotypes. Genome Biology, 2011, 12, R85.	13.9	72
147	Partial ADAMTS13 deficiency in atypical hemolytic uremic syndrome. Blood, 2013, 122, 1487-1493.	0.6	72
148	Defining the Complement Biomarker Profile of C3 Glomerulopathy. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 1876-1882.	2.2	72
149	Exercise-Induced Laryngomalacia. Annals of Otology, Rhinology and Laryngology, 1995, 104, 537-541.	0.6	71
150	Selective Cochlear Degeneration in Mice Lacking the F-Box Protein, Fbx2, a Glycoprotein-Specific Ubiquitin Ligase Subunit. Journal of Neuroscience, 2007, 27, 5163-5171.	1.7	70
151	An integrated genetic and functional analysis of the role of type II transmembrane serine proteases (TMPRSSs) in hearing loss. Human Mutation, 2008, 29, 130-141.	1.1	70
152	International Pediatric ORL Group (IPOG) laryngomalacia consensus recommendations. International Journal of Pediatric Otorhinolaryngology, 2016, 86, 256-261.	0.4	70
153	A human recessive neurosensory nonsyndromic hearing impairment locus is a potential homologue of the murine deafness (dn) locus. Human Molecular Genetics, 1995, 4, 2391-2394.	1.4	69
154	Genetic variants in the peripheral auditory system significantly affect adult cochlear implant performance. Hearing Research, 2017, 348, 138-142.	0.9	68
155	Reflex Laryngospasm Induced by Stimulation of Distal Esophageal Afferents. Laryngoscope, 1994, 101, 209???214.	1.1	67
156	A Comparative Study of Eya1 and Eya4 Protein Function and Its Implication in Branchio-oto-renal Syndrome and DFNA10. JARO - Journal of the Association for Research in Otolaryngology, 2004, 5, 295-304.	0.9	67
157	ClinGen expert clinical validity curation of 164 hearing loss gene–disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	1.1	67
158	Advances in Molecular and Cellular Therapies for Hearing Loss. Molecular Therapy, 2008, 16, 224-236.	3.7	66
159	A Genome-wide Analysis Identifies Genetic Variants in the RELN Gene Associated with Otosclerosis. American Journal of Human Genetics, 2009, 84, 328-338.	2.6	66
160	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. EMBO Molecular Medicine, 2017, 9, 1711-1731.	3.3	66
161	Targeted Allele Suppression Prevents Progressive Hearing Loss in the Mature Murine Model of Human TMC1 Deafness. Molecular Therapy, 2019, 27, 681-690.	3.7	66
162	Value-Based Pathology: <i>A Cost-Benefit Analysis of the Examination of Routine and Nonroutine Tonsil and Adenoid Specimens</i> . American Journal of Clinical Pathology, 1997, 108, 158-165.	0.4	64

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163	Lymphatic Malformations. Lymphatic Research and Biology, 2004, 2, 25-31.	0.5	64
164	Characterisation of DRASIC in the mouse inner ear. Hearing Research, 2004, 190, 149-160.	0.9	64
165	Localization of the gene for branchiootorenal syndrome to chromosome 8q. Genomics, 1992, 14, 841-844.	1.3	63
166	Genetic Heterogeneity of Usher Syndrome: Analysis of 151 Families with Usher Type I. American Journal of Human Genetics, 2000, 67, 1569-1574.	2.6	63
167	A proposal for comprehensive newborn hearing screening to improve identification of deaf and hard-of-hearing children. Genetics in Medicine, 2019, 21, 2614-2630.	1.1	63
168	Case of Progressive Dysplasia Concomitant with Intralesional Cidofovir Administration for Recurrent Respiratory Papillomatosis. Annals of Otology, Rhinology and Laryngology, 2005, 114, 836-839.	0.6	62
169	A catechol- <i>O</i> -methyltransferase that is essential for auditory function in mice and humans. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 14609-14614.	3.3	62
170	Pediatric Medial Subperiosteal Orbital Abscess: Medical Management Where Possible. American Journal of Rhinology & Allergy, 2004, 18, 321-327.	2.3	61
171	Pendred syndrome and DFNB4-mutation screening ofSLC26A4by denaturing high-performance liquid chromatography and the identification of eleven novel mutations. , 2004, 124A, 1-9.		61
172	The Influence of Mutations in the SLC26A4 Gene on the Temporal Bone in a Population With Enlarged Vestibular Aqueduct. JAMA Otolaryngology, 2007, 133, 162.	1.5	61
173	Association of Bone Morphogenetic Proteins With Otosclerosis. Journal of Bone and Mineral Research, 2008, 23, 507-516.	3.1	58
174	The Role of Connexins in Human Disease. Ear and Hearing, 2003, 24, 314-323.	1.0	57
175	Lymphangioma. An otolaryngologic perspective. International Journal of Pediatric Otorhinolaryngology, 1987, 14, 133-140.	0.4	56
176	Autosomal dominant branchio-oto-renal syndrome—localization of a disease gene to chromosome 8q by linkage in a Dutch family. Human Molecular Genetics, 1992, 1, 491-495.	1.4	56
177	Recurrent respiratory papillomatosis: pathogenesis to treatment. Current Opinion in Otolaryngology and Head and Neck Surgery, 2005, 13, 354-359.	0.8	56
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