Richard J H Smith

List of Publications by Citations

550 papers	27,553 citations	85 h-index	141 g-index
629 ext. papers	31,609 ext. citations	5.2 avg, IF	6.71 L-index

#	Paper	IF	Citations
550	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, 7227-32	11.5	1638
549	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	11	495
548	Sensorineural hearing loss in children. *Lancet, The*, **2005**, 365, 879-90	40	482
547	A candidate prostate cancer susceptibility gene at chromosome 17p. *Nature Genetics*, **2001**, 27, 172-80	36.3	469
546	C3 glomerulopathy: consensus report. *Kidney International*, **2013**, 84, 1079-89	9.9	398
545	GJB2 mutations and degree of hearing loss: a multicenter study. *American Journal of Human Genetics*, **2005**, 77, 945-57	11	372
544	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	7	335
543	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, 8090-5	11.5	325
542	Membranoproliferative glomerulonephritis type II (dense deposit disease): an update. *Journal of the American Society of Nephrology: JASN*, **2005**, 16, 1392-403	12.7	310
541	Mutations in the human alpha-tectorin gene cause autosomal dominant non-syndromic hearing impairment. *Nature Genetics*, **1998**, 19, 60-2	36.3	274
540	Localization of Usher syndrome type II to chromosome 1q. *Genomics*, **1990**, 7, 245-9	4.3	257
539	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, 21104-9	11.5	253
538	Comprehensive genetic testing in the clinical evaluation of 1119 patients with hearing loss. *Human Genetics*, **2016**, 135, 441-450	6.3	250
537	Nonsyndromic hearing impairment is associated with a mutation in DFNA5. *Nature Genetics*, **1998**, 20, 194-7	36.3	244
536	Pendred syndrome, DFNB4, and PDS/SLC26A4 identification of eight novel mutations and possible genotype-phenotype correlations. *Human Mutation*, **2001**, 17, 403-11	4.7	244
535	Eculizumab for dense deposit disease and C3 glomerulonephritis. *Clinical Journal of the American Society of Nephrology: CJASN*, **2012**, 7, 748-56	6.9	238
534	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, 1452-8	11	236

533	Mutations in COL11A2 cause non-syndromic hearing loss (DFNA13). *Nature Genetics*, **1999**, 23, 413-9	36.3	225
532	Mutations in alternative pathway complement proteins in American patients with atypical hemolytic uremic syndrome. *Human Mutation*, **2010**, 31, E1445-60	4.7	224
531	C3 glomerulonephritis: clinicopathological findings, complement abnormalities, glomerular proteomic profile, treatment, and follow-up. *Kidney International*, **2012**, 82, 465-73	9.9	215
530	Laryngomalacia and its treatment. *Laryngoscope*, **1999**, 109, 1770-5	3.6	214
529	Localization of two genes for Usher syndrome type I to chromosome 11. *Genomics*, **1992**, 14, 995-1002	4.3	203
528	The 1000 Genomes Project: data management and community access. *Nature Methods*, **2012**, 9, 459-62	21.6	202
527	New approaches to the treatment of dense deposit disease. *Journal of the American Society of Nephrology: JASN*, **2007**, 18, 2447-56	12.7	200
526	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, 278-92	11	190
525	Congenital hearing loss. *Nature Reviews Disease Primers*, **2017**, 3, 16094	51.1	184
524	Atypical aHUS: State of the art. *Molecular Immunology*, **2015**, 67, 31-42	4.3	177
523	Phenotypic manifestations of branchio-oto-renal syndrome. *American Journal of Medical Genetics Part A*, **1995**, 58, 365-70		168
522	Treatment of lymphangiomas with OK-432 (Picibanil) sclerotherapy: a prospective multi-institutional trial. *JAMA Otolaryngology*, **2002**, 128, 1137-44		167
521	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	12.7	165
520	The functional spectrum of low-frequency coding variation. *Genome Biology*, **2011**, 12, R84	18.3	161
519	Human male infertility caused by mutations in the CATSPER1 channel protein. *American Journal of Human Genetics*, **2009**, 84, 505-10	11	158
518	Transcriptional control of SLC26A4 is involved in Pendred syndrome and nonsyndromic enlargement of vestibular aqueduct (DFNB4). *American Journal of Human Genetics*, **2007**, 80, 1055-63	11	156
517	Frequency of Usher syndrome in two pediatric populations: Implications for genetic screening of deaf and hard of hearing children. *Genetics in Medicine*, **2010**, 12, 512-6	8.1	154
516	Defective complement control of factor H (Y402H) and FHL-1 in age-related macular degeneration. *Molecular Immunology*, **2007**, 44, 3398-406	4.3	154

515	Branchio-oto-renal syndrome: the mutation spectrum in EYA1 and its phenotypic consequences. *Human Mutation*, **2004**, 23, 582-9	4.7	152
514	Mutations in the transcriptional activator EYA4 cause late-onset deafness at the DFNA10 locus. *Human Molecular Genetics*, **2001**, 10, 195-200	5.6	150
513	Genomic structure and identification of novel mutations in usherin, the gene responsible for Usher syndrome type IIa. *American Journal of Human Genetics*, **2000**, 66, 1199-210	11	146
512	Prestin, a cochlear motor protein, is defective in non-syndromic hearing loss. *Human Molecular Genetics*, **2003**, 12, 1155-62	5.6	145
511	Membranoproliferative glomerulonephritis and C3 glomerulopathy: resolving the confusion. *Kidney International*, **2012**, 81, 434-41	9.9	144
510	Transcription factor SIX5 is mutated in patients with branchio-oto-renal syndrome. *American Journal of Human Genetics*, **2007**, 80, 800-4	11	144
509	Impact of tympanostomy tubes on child quality of life. *JAMA Otolaryngology*, **2000**, 126, 585-92		141
508	Glomeruli of Dense Deposit Disease contain components of the alternative and terminal complement pathway. *Kidney International*, **2009**, 75, 952-60	9.9	140
507	Mutational spectrum of the WFS1 gene in Wolfram syndrome, nonsyndromic hearing impairment, diabetes mellitus, and psychiatric disease. *Human Mutation*, **2003**, 22, 275-87	4.7	139
506	Causes of alternative pathway dysregulation in dense deposit disease. *Clinical Journal of the American Society of Nephrology: CJASN*, **2012**, 7, 265-74	6.9	136
505	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, 1278-87	12.7	132
504	Membranoproliferative glomerulonephritis secondary to monoclonal gammopathy. *Clinical Journal of the American Society of Nephrology: CJASN*, **2010**, 5, 770-82	6.9	130
503	Incidence of vocal fold paralysis in infants undergoing ligation of patent ductus arteriosus. *Annals of Thoracic Surgery*, **1996**, 61, 814-6	2.7	130
502	C3 glomerulonephritis associated with monoclonal gammopathy: a case series. *American Journal of Kidney Diseases*, **2013**, 62, 506-14	7.4	127
501	Atypical postinfectious glomerulonephritis is associated with abnormalities in the alternative pathway of complement. *Kidney International*, **2013**, 83, 293-9	9.9	124
500	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, 2163-75	6.6	124
499	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, 651-7	11	121
498	Utilizing ethnic-specific differences in minor allele frequency to recategorize reported pathogenic deafness variants. *American Journal of Human Genetics*, **2014**, 95, 445-53	11	118

497	Genomic Landscape and Mutational Signatures of Deafness-Associated Genes. *American Journal of Human Genetics*, **2018**, 103, 484-497	11	116
496	Proliferative glomerulonephritis secondary to dysfunction of the alternative pathway of complement. *Clinical Journal of the American Society of Nephrology: CJASN*, **2011**, 6, 1009-17	6.9	114
495	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, 1709-15	5.6	113
494	Copy number variants are a common cause of non-syndromic hearing loss. *Genome Medicine*, **2014**, 6, 37	14.4	111
493	Efficacy and safety of OK-432 immunotherapy of lymphatic malformations. *Laryngoscope*, **2009**, 119, 107-15	3.6	111
492	Function and expression pattern of nonsyndromic deafness genes. *Current Molecular Medicine*, **2009**, 9, 546-64	2.5	111
491	An update on the treatment of hemangiomas in children with interferon alfa-2a. *JAMA Otolaryngology*, **1999**, 125, 21-7		111
490	Genetic male infertility and mutation of CATSPER ion channels. *European Journal of Human Genetics* , **2010**, 18, 1178-84	5.3	110
489	Cystic fibrosis--an otolaryngologic perspective. *Otolaryngology - Head and Neck Surgery*, **1987**, 97, 356-60	5.5	110
488	C3 glomerulopathy - understanding a rare complement-driven renal disease. *Nature Reviews Nephrology*, **2019**, 15, 129-143	14.9	109
487	A classification scheme for paradoxical vocal cord motion. *Laryngoscope*, **1997**, 107, 1429-35	3.6	109
486	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, 328-37	11	108
485	Spastic diplegia and other motor disturbances in infants receiving interferon-alpha. *Laryngoscope*, **2004**, 114, 1231-6	3.6	107
484	Tracheal allograft reconstruction: the total North American and worldwide pediatric experiences. *Annals of Thoracic Surgery*, **1999**, 68, 1043-51; discussion 1052	2.7	106
483	New treatment options for lymphangioma in infants and children. *Annals of Otology, Rhinology and Laryngology*, **2002**, 111, 1066-75	2.1	101
482	Parental attitudes toward genetic testing for pediatric deafness. *American Journal of Human Genetics*, **2000**, 67, 1621-5	11	101
481	Clinical findings, pathology, and outcomes of C3GN after kidney transplantation. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 1110-7	12.7	100
480	Dense deposit disease associated with monoclonal gammopathy of undetermined significance. *American Journal of Kidney Diseases*, **2010**, 56, 977-82	7.4	100

479	Mutations in a novel gene, TMIE, are associated with hearing loss linked to the DFNB6 locus. *American Journal of Human Genetics*, **2002**, 71, 632-6	11	99
478	Pre-emptive eculizumab and plasmapheresis for renal transplant in atypical hemolytic uremic syndrome. *Clinical Journal of the American Society of Nephrology: CJASN*, **2011**, 6, 1488-94	6.9	98
477	Branchio-oto-renal syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1671-8	2.5	96
476	In vitro and in vivo suppression of GJB2 expression by RNA interference. *Human Molecular Genetics*, **2005**, 14, 1641-50	5.6	93
475	Clinical aspects of hereditary hearing loss. *Genetics in Medicine*, **2007**, 9, 393-408	8.1	91
474	Advancing genetic testing for deafness with genomic technology. *Journal of Medical Genetics*, **2013**, 50, 627-34	5.8	90
473	Temporal bone analysis of patients with presbycusis reveals high frequency of mitochondrial mutations. *Hearing Research*, **1997**, 110, 147-54	3.9	90
472	Treatment of lymphangiomas in children: an update of Picibanil (OK-432) sclerotherapy. *Otolaryngology - Head and Neck Surgery*, **1999**, 121, 381-7	5.5	90
471	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	5.3	89
470	A claudin-9-based ion permeability barrier is essential for hearing. *PLoS Genetics*, **2009**, 5, e1000610	6	88
469	Loss-of-function mutations of ILDR1 cause autosomal-recessive hearing impairment DFNB42. *American Journal of Human Genetics*, **2011**, 88, 127-37	11	87
468	Genotype-phenotype correlations for SLC26A4-related deafness. *Human Genetics*, **2007**, 122, 451-7	6.3	87
467	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	18.3	86
466	Massively Parallel Sequencing for Genetic Diagnosis of Hearing Loss: The New Standard of Care. *Otolaryngology - Head and Neck Surgery*, **2015**, 153, 175-82	5.5	83
465	Recent advances in the molecular genetics of epilepsy. *Journal of Medical Genetics*, **2013**, 50, 271-9	5.8	82
464	Carcinoembryonic antigen-related cell adhesion molecule 16 interacts with alpha-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, 4218-23	11.5	82
463	Allelic variants of complement genes associated with dense deposit disease. *Journal of the American Society of Nephrology: JASN*, **2011**, 22, 1551-9	12.7	81
462	Inactivation of NADPH oxidase organizer 1 results in severe imbalance. *Current Biology*, **2006**, 16, 208-13	6.3	81

461	Mapping interactions between complement C3 and regulators using mutations in atypical hemolytic uremic syndrome. *Blood*, **2015**, 125, 2359-69	2.2	79
460	A second gene for otosclerosis, OTSC2, maps to chromosome 7q34-36. *American Journal of Human Genetics*, **2001**, 68, 495-500	11	79
459	Connexin mutations and hearing loss. *Nature*, **1998**, 391, 32	50.4	77
458	Conductive hearing loss and otopathology in cleft palate patients. *Otolaryngology - Head and Neck Surgery*, **2006**, 134, 946-8	5.5	77
457	Sensorineural deafness and male infertility: a contiguous gene deletion syndrome. *Journal of Medical Genetics*, **2007**, 44, 233-40	5.8	75
456	Normal hearing in Splotch (Sp/+), the mouse homologue of Waardenburg syndrome type 1. *Nature Genetics*, **1992**, 2, 75-9	36.3	75
455	Eculizumab and recurrent C3 glomerulonephritis. *Pediatric Nephrology*, **2013**, 28, 1975-81	3.2	74
454	Complement factor B mutations in atypical hemolytic uremic syndrome-disease-relevant or benign?. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 2053-65	12.7	74
453	Cloning and characterization of SLC26A6, a novel member of the solute carrier 26 gene family. *Genomics*, **2001**, 72, 43-50	4.3	74
452	Linkage of a gene for dominant non-syndromic deafness to chromosome 19. *Human Molecular Genetics*, **1995**, 4, 1073-6	5.6	74
451	A gene for autosomal dominant nonsyndromic hereditary hearing impairment maps to 4p16.3. *Human Molecular Genetics*, **1995**, 4, 1967-72	5.6	74
450	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, 247-56	2.4	73
449	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. *Journal of Medical Genetics*, **2015**, 52, 823-9	5.8	72
448	Performance of cochlear implant recipients with GJB2-related deafness. *American Journal of Medical Genetics Part A*, **2002**, 109, 167-70		72
447	GJB2 mutations: passage through Iran. *American Journal of Medical Genetics, Part A*, **2005**, 133A, 132-7	2.5	72
446	Prediction of cochlear implant performance by genetic mutation: the spiral ganglion hypothesis. *Hearing Research*, **2012**, 292, 51-8	3.9	71
445	Soluble CR1 therapy improves complement regulation in C3 glomerulopathy. *Journal of the American Society of Nephrology: JASN*, **2013**, 24, 1820-9	12.7	71
444	Gipc3 mutations associated with audiogenic seizures and sensorineural hearing loss in mouse and human. *Nature Communications*, **2011**, 2, 201	17.4	71

443	Genetics: advances in genetic testing for deafness. *Current Opinion in Pediatrics*, **2012**, 24, 679-86	3.2	71
442	Dense deposit disease. *Molecular Immunology*, **2011**, 48, 1604-10	4.3	70
441	GJB2: the spectrum of deafness-causing allele variants and their phenotype. *Human Mutation*, **2004**, 24, 305-11	4.7	70
440	Mutations in the WFS1 gene that cause low-frequency sensorineural hearing loss are small non-inactivating mutations. *Human Genetics*, **2002**, 110, 389-94	6.3	70
439	TBC1D24 mutation causes autosomal-dominant nonsyndromic hearing loss. *Human Mutation*, **2014**, 35, 819-23	4.7	69
438	Pediatric exercise-induced laryngomalacia. *Annals of Otology, Rhinology and Laryngology*, **1996**, 105, 169-75	2.1	68
437	Temporal bone histopathology in connexin 26-related hearing loss. *Laryngoscope*, **2000**, 110, 269-75	3.6	68
436	Pelvic tilt. Intratester reliability of measuring the standing position and range of motion. *Physical Therapy*, **1985**, 65, 169-74	3.3	68
435	The coding polymorphism T263I in TGF-beta1 is associated with otosclerosis in two independent populations. *Human Molecular Genetics*, **2007**, 16, 2021-30	5.6	67
434	High-Throughput Genetic Testing for Thrombotic Microangiopathies and C3 Glomerulopathies. *Journal of the American Society of Nephrology: JASN*, **2016**, 27, 1245-53	12.7	66
433	Reducing the exome search space for mendelian diseases using genetic linkage analysis of exome genotypes. *Genome Biology*, **2011**, 12, R85	18.3	66
432	Mutations of the RDX gene cause nonsyndromic hearing loss at the DFNB24 locus. *Human Mutation*, **2007**, 28, 417-23	4.7	66
431	RNA Interference Prevents Autosomal-Dominant Hearing Loss. *American Journal of Human Genetics*, **2016**, 98, 1101-1113	11	66
430	The role of complement in C3 glomerulopathy. *Molecular Immunology*, **2015**, 67, 21-30	4.3	65
429	The mitochondrial TIM22 preprotein translocase is highly conserved throughout the eukaryotic kingdom. *FEBS Letters*, **1999**, 464, 41-7	3.8	64
428	SIX1 mutation screening in 247 branchio-oto-renal syndrome families: a recurrent missense mutation associated with BOR. *Human Mutation*, **2008**, 29, 565	4.7	63
427	Mutations in apoptosis-inducing factor cause X-linked recessive auditory neuropathy spectrum disorder. *Journal of Medical Genetics*, **2015**, 52, 523-31	5.8	62
426	DFNA8/12 caused by TECTA mutations is the most identified subtype of nonsyndromic autosomal dominant hearing loss. *Human Mutation*, **2011**, 32, 825-34	4.7	62

425	HspE7 treatment of pediatric recurrent respiratory papillomatosis: final results of an open-label trial. *Annals of Otology, Rhinology and Laryngology*, **2005**, 114, 730-7	2.1	62
424	A human recessive neurosensory nonsyndromic hearing impairment locus is potential homologue of murine deafness (dn) locus. *Human Molecular Genetics*, **1995**, 4, 2391-4	5.6	62
423	Recurrent respiratory papillomatosis. *Pediatric Clinics of North America*, **1996**, 43, 1385-401	3.6	61
422	Current management of laryngeal and laryngotracheoesophageal clefts. *Journal of Pediatric Surgery*, **1990**, 25, 855-60	2.6	61
421	Clinical features and outcomes of 98 children and adults with dense deposit disease. *Pediatric Nephrology*, **2012**, 27, 773-81	3.2	60
420	Partial ADAMTS13 deficiency in atypical hemolytic uremic syndrome. *Blood*, **2013**, 122, 1487-93	2.2	60
419	The effect of GJB2 allele variants on performance after cochlear implantation. *Laryngoscope*, **2003**, 113, 2135-40	3.6	60
418	Deafness in the genomics era. *Hearing Research*, **2011**, 282, 1-9	3.9	59
417	Causes of facial swelling in pediatric patients: correlation of clinical and radiologic findings. *Radiographics*, **2006**, 26, 157-71	5.4	59
416	Congenital oral synechiae. *Annals of Otology, Rhinology and Laryngology*, **1993**, 102, 186-97	2.1	59
415	A mutation in the Srrm4 gene causes alternative splicing defects and deafness in the Bronx waltzer mouse. *PLoS Genetics*, **2012**, 8, e1002966	6	58
414	Branchio-oto-renal syndrome (BOR): novel mutations in the EYA1 gene, and a review of the mutational genetics of BOR. *Human Mutation*, **2008**, 29, 537-44	4.7	58
413	Selective cochlear degeneration in mice lacking the F-box protein, Fbx2, a glycoprotein-specific ubiquitin ligase subunit. *Journal of Neuroscience*, **2007**, 27, 5163-71	6.6	58
412	Lymphatic malformations. *Lymphatic Research and Biology*, **2004**, 2, 25-31	2.3	58
411	Characterisation of DRASIC in the mouse inner ear. *Hearing Research*, **2004**, 190, 149-60	3.9	58
410	Localization of the gene for branchiootorenal syndrome to chromosome 8q. *Genomics*, **1992**, 14, 841-4	4.3	58
409	Advances in molecular and cellular therapies for hearing loss. *Molecular Therapy*, **2008**, 16, 224-36	11.7	57
408	Maternally inherited hearing impairment. *Clinical Genetics*, **2000**, 57, 409-14	4	57

407	Exercise-induced laryngomalacia. *Annals of Otology, Rhinology and Laryngology*, **1995**, 104, 537-41	2.1	56
406	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, 775-9	2.1	56
405	International Pediatric Otolaryngology Group (IPOG) consensus recommendations: Hearing loss in the pediatric patient. *International Journal of Pediatric Otorhinolaryngology*, **2016**, 90, 251-258	1.7	56
404	An integrated genetic and functional analysis of the role of type II transmembrane serine proteases (TMPRSSs) in hearing loss. *Human Mutation*, **2008**, 29, 130-41	4.7	55
403	Enhanced viral-mediated cochlear gene delivery in adult mice by combining canal fenestration with round window membrane inoculation. *Scientific Reports*, **2018**, 8, 2980	4.9	53
402	Defining the complement biomarker profile of C3 glomerulopathy. *Clinical Journal of the American Society of Nephrology: CJASN*, **2014**, 9, 1876-82	6.9	53
401	Treatment options for C3 glomerulopathy. *Current Opinion in Nephrology and Hypertension*, **2013**, 22, 231-7	3.5	53
400	Genetic heterogeneity of Usher syndrome: analysis of 151 families with Usher type I. *American Journal of Human Genetics*, **2000**, 67, 1569-74	11	53
399	A genome-wide analysis identifies genetic variants in the RELN gene associated with otosclerosis. *American Journal of Human Genetics*, **2009**, 84, 328-38	11	52
398	Pediatric Medial Subperiosteal Orbital Abscess: Medical Management Where Possible. *American Journal of Rhinology & Allergy*, **2004**, 18, 321-327		52
397	Pendred syndrome and DFNB4-mutation screening of SLC26A4 by denaturing high-performance liquid chromatography and the identification of eleven novel mutations **2004**, 124A, 1-9		52
396	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, 491-5	5.6	52
395	C3 glomerulopathy associated with monoclonal Ig is a distinct subtype. *Kidney International*, **2018**, 94, 178-186	9.9	51
394	Association of bone morphogenetic proteins with otosclerosis. *Journal of Bone and Mineral Research*, **2008**, 23, 507-16	6.3	51
393	Diagnosis of complement alternative pathway disorders. *Kidney International*, **2016**, 89, 278-88	9.9	50
392	A catechol-O-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, 14609-14	11.5	50
391	Insights into the Biology of Hearing and Deafness Revealed by Single-Cell RNA Sequencing. *Cell Reports*, **2019**, 26, 3160-3171.e3	10.6	49
390	The influence of mutations in the SLC26A4 gene on the temporal bone in a population with enlarged vestibular aqueduct. *JAMA Otolaryngology*, **2007**, 133, 162-8		49

389	A seventh locus for otosclerosis, OTSC7, maps to chromosome 6q13-16.1. *European Journal of Human Genetics*, **2007**, 15, 362-8	5.3	48
388	A comprehensive study to determine heterogeneity of autosomal recessive nonsyndromic hearing loss in Iran. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2485-92	2.5	47
387	New gene for autosomal recessive non-syndromic hearing loss maps to either chromosome 3q or 19p. *American Journal of Medical Genetics Part A*, **1997**, 71, 467-471		47
386	Case of progressive dysplasia concomitant with intralesional cidofovir administration for recurrent respiratory papillomatosis. *Annals of Otology, Rhinology and Laryngology*, **2005**, 114, 836-9	2.1	47
385	Deletion of and novel missense mutation in POU3F4 in 2 families segregating X-linked nonsyndromic deafness. *JAMA Otolaryngology*, **2005**, 131, 1057-63		47
384	Reflex laryngospasm induced by stimulation of distal esophageal afferents. *Laryngoscope*, **1994**, 104, 209-14	3.6	47
383	The role of connexins in human disease. *Ear and Hearing*, **2003**, 24, 314-23	3.4	46
382	Small-molecule factor B inhibitor for the treatment of complement-mediated diseases. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 7926-7931	11.5	45
381	C3 Glomerulopathy: Ten YearsNExperience at Mayo Clinic. *Mayo Clinic Proceedings*, **2018**, 93, 991-1008	6.4	45
380	PDZD7 and hearing loss: More than just a modifier. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2957-65	2.5	45
379	Screening for MYO15A gene mutations in autosomal recessive nonsyndromic, GJB2 negative Iranian deaf population. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1857-64	2.5	45
378	Linkage analysis of progressive hearing loss in five extended families maps the DFNA2 gene to a 1.25-Mb region on chromosome 1p. *Genomics*, **1997**, 41, 70-4	4.3	45
377	Mice lacking Dfna5 show a diverging number of cochlear fourth row outer hair cells. *Neurobiology of Disease*, **2005**, 19, 386-99	7.5	45
376	A gene for recessive nonsyndromic sensorineural deafness (DFNB18) maps to the chromosomal region 11p14-p15.1 containing the Usher syndrome type 1C gene. *Genomics*, **1998**, 50, 290-2	4.3	45
375	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	3.3	44
374	Nonsyndromic hearing loss. *Ear and Hearing*, **2003**, 24, 275-88	3.4	43
373	Refined localization of the branchiootorenal syndrome gene by linkage and haplotype analysis. *American Journal of Medical Genetics Part A*, **1994**, 51, 176-84		43
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