Richard J H Smith

List of Publications by Year in Descending Order

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 Manual Technique for Isolation and Single-Cell RNA Sequencing Analysis of Cochlear Hair Cells and Supporting Cells. *Neuromethods*, **2022**, 131-149	0.4	0
549	AudioGene: refining the natural history of KCNQ4, GSDME, WFS1, and COCH-associated hearing loss.. *Human Genetics*, **2022**, 1	6.3	4
548	DVPred: a disease-specific prediction tool for variant pathogenicity classification for hearing loss.. *Human Genetics*, **2022**, 1	6.3	0
547	The voltage-gated Ca2+ channel subunit 2δ4 regulates locomotor behavior and sensorimotor gating in mice.. *PLoS ONE*, **2022**, 17, e0263197	3.7	1
546	The hearing-impaired patient: what the future holds.. *Human Genetics*, **2022**, 1	6.3	0
545	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants.. *Genetics in Medicine*, **2021**,	8.1	3
544	Rigid Video Laryngoscopy for Intubation in Severe Pierre Robin Sequence: A Retrospective Review. *Laryngoscope*, **2021**, 131, 1647-1651	3.6	1
543	Exome sequencing utility in defining the genetic landscape of hearing loss and novel-gene discovery in Iran. *Clinical Genetics*, **2021**, 100, 59-78	4	1
542	Pembrolizumab Induced Acute Persistent Airway Disease in a Patient with Recurrent Respiratory Papillomatosis (RRP). *Annals of Otology, Rhinology and Laryngology*, **2021**, 34894211021276	2.1	1
541	TSPEAR variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2417-2433	2.5	3
540	Functional characterization of 105 factor H variants associated with aHUS: lessons for variant classification. *Blood*, **2021**, 138, 2185-2201	2.2	3
539	Genetic Causes of Hearing Loss in a Large Cohort of Cochlear Implant Recipients. *Otolaryngology - Head and Neck Surgery*, **2021**, 1945998211021308	5.5	3
538	Clinical Pharmacogenetics Implementation Consortium Guideline for the Use of Aminoglycosides Based on MT-RNR1 Genotype. *Clinical Pharmacology and Therapeutics*, **2021**,	6.1	18
537	and Copy Number Variations in C3 Glomerulopathy and Immune Complex-Mediated Membranoproliferative Glomerulonephritis. *Frontiers in Genetics*, **2021**, 12, 670727	4.5	1
536	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. *Genetics in Medicine*, **2021**, 23, 2208-2212	8.1	0
535	Systematic and other reviews: criteria and complexities. *Journal of Laryngology and Otology*, **2021**, 135, 565-567	1.8	
534	Neonatal Lateral Epiglottic Defects. *Annals of Otology, Rhinology and Laryngology*, **2021**, 130, 311-313	2.1	0

533	What Is the Best Approach to Prevent Advanced-Stage Pressure Injuries After Pediatric Tracheotomy?. *Laryngoscope*, **2021**, 131, 1196-1197	3.6	
532	International Pediatric Otolaryngology Group (IPOG) management recommendations: Pediatric tracheostomy decannulation. *International Journal of Pediatric Otorhinolaryngology*, **2021**, 141, 110565	1.7	1
531	Targeted broad-based genetic testing by next-generation sequencing informs diagnosis and facilitates management in patients with kidney diseases. *Nephrology Dialysis Transplantation*, **2021**, 36, 295-305	4.3	20
530	Systemic Bevacizumab for Treatment of Respiratory Papillomatosis: International Consensus Statement. *Laryngoscope*, **2021**, 131, E1941-E1949	3.6	10
529	A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. *European Journal of Human Genetics*, **2021**, 29, 988-997	5.3	3
528	International Pediatric Otolaryngology Group (IPOG) survey: Efforts to avoid complications in home tracheostomy care. *International Journal of Pediatric Otorhinolaryngology*, **2021**, 141, 110563	1.7	2
527	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. *Human Genetics*, **2021**, 1	6.3	1
526	Selective Binding of Heparin/Heparan Sulfate Oligosaccharides to Factor H and Factor H-Related Proteins: Therapeutic Potential for C3 Glomerulopathies. *Frontiers in Immunology*, **2021**, 12, 676662	8.4	0
525	Monoclonal Gammopathy of Renal Significance Causes C3 Glomerulonephritis Via Monoclonal IgG Kappa Inhibition of Complement Factor H. *Kidney International Reports*, **2021**, 6, 2505-2509	4.1	0
524	COCH-related autosomal dominant nonsyndromic hearing loss: a phenotype-genotype study. *Human Genetics*, **2021**, 1	6.3	2
523	Improving Clinical Trials for Anticomplement Therapies in Complement-Mediated Glomerulopathies: Report of a Scientific Workshop Sponsored by the National Kidney Foundation. *American Journal of Kidney Diseases*, **2021**,	7.4	5
522	Genetic testing hearing loss: The challenge of non syndromic mimics. *International Journal of Pediatric Otorhinolaryngology*, **2021**, 150, 110872	1.7	1
521	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. *Human Genetics*, **2021**, 140, 915-931	6.3	5
520	Consortium of Otolaryngology Journal Editors: collegiality and contributions. *Journal of Laryngology and Otology*, **2020**, 134, 379-380	1.8	
519	Factor H Autoantibodies and Complement-Mediated Diseases. *Frontiers in Immunology*, **2020**, 11, 607211	8.4	5
518	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. *Human Genetics*, **2020**, 139, 1315-1323	6.3	8
517	Consortium of Otolaryngology Journal Editors-Collegiality and Contributions. *JAMA Otolaryngology - Head and Neck Surgery*, **2020**, 146, 521-522	3.9	1
516	Mutation of complement factor B causing massive fluid-phase dysregulation of the alternative complement pathway can result in atypical hemolytic uremic syndrome. *Kidney International*, **2020**, 98, 1265-1274	9.9	6

515	() c.991-15_991-13delTTC: Founder Mutation or Mutational Hotspot?. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	6
514	Novel loss-of-function mutations in COCH cause autosomal recessive nonsyndromic hearing loss. *Human Genetics*, **2020**, 139, 1565-1574	6.3	3
513	Initial experience from a renal genetics clinic demonstrates a distinct role in patient management. *Genetics in Medicine*, **2020**, 22, 1025-1035	8.1	20
512	Consortium of otolaryngology journal editors: Collegiality and contributions. *Operative Techniques in Otolaryngology - Head and Neck Surgery*, **2020**, 31, 71-73	0.4	
511	Hair Cell Transduction Efficiency of Single- and Dual-AAV Serotypes in Adult Murine Cochleae. *Molecular Therapy - Methods and Clinical Development*, **2020**, 17, 1167-1177	6.4	9
510	Genetic Testing for Congenital Bilateral Hearing Loss in the Context of Targeted Cytomegalovirus Screening. *Laryngoscope*, **2020**, 130, 2714-2718	3.6	9
509	Proteomic Analysis of Complement Proteins in Membranous Nephropathy. *Kidney International Reports*, **2020**, 5, 618-626	4.1	23
508	Insights into the pathophysiology of DFNA10 hearing loss associated with novel EYA4 variants. *Scientific Reports*, **2020**, 10, 6213	4.9	7
507	Is it Usher syndrome? Collaborative diagnosis and molecular genetics of patients with visual impairment and hearing loss. *Ophthalmic Genetics*, **2020**, 41, 151-158	1.2	4
506	C3(H2O) prevents rescue of complement-mediated C3 glomerulopathy in Cfh-/- Cfd-/- mice. *JCI Insight*, **2020**, 5,	9.9	5
505	Advanced practice providers and children's hospital-based pediatric otolarynology practices. *International Journal of Pediatric Otorhinolaryngology*, **2020**, 129, 109770	1.7	6
504	International Pediatric ORL Group (IPOG) Robin Sequence consensus recommendations. *International Journal of Pediatric Otorhinolaryngology*, **2020**, 130, 109855	1.7	3
503	International Pediatric Otolaryngology Group (IPOG) Consensus Recommendations: Congenital Cholesteatoma. *Otology and Neurotology*, **2020**, 41, 345-351	2.6	5
502	A recurrent missense variant in HARS2 results in variable sensorineural hearing loss in three unrelated families. *Journal of Human Genetics*, **2020**, 65, 305-311	4.3	4
501	Future directions for screening and treatment in congenital hearing loss. *Precision Clinical Medicine*, **2020**, 3, 175-186	6.7	9
500	International Pediatric Otolaryngology Group (IPOG) consensus recommendations: Management of suprastomal collapse in the pediatric population. *International Journal of Pediatric Otorhinolaryngology*, **2020**, 139, 110427	1.7	0
499	Minimal Change Disease With Nephrotic Syndrome Associated With Coronavirus Disease 2019 After Apolipoprotein L1 Risk Variant Kidney Transplant: A Case Report. *Transplantation Proceedings* , **2020**, 52, 2693-2697	1.1	9
498	International Pediatric Otolaryngology Group (IPOG): Consensus recommendations on the prenatal and perinatal management of anticipated airway obstruction. *International Journal of Pediatric Otorhinolaryngology*, **2020**, 138, 110281	1.7	6

497	International Pediatric Otolaryngology group (IPOG) consensus on the diagnosis and management of pediatric obstructive sleep apnea (OSA). *International Journal of Pediatric Otorhinolaryngology*, **2020**, 138, 110276	1.7	9
496	International Pediatric Otolaryngology Group (IPOG): Juvenile-onset recurrent respiratory papillomatosis consensus recommendations. *International Journal of Pediatric Otorhinolaryngology*, **2020**, 128, 109697	1.7	17
495	C3 glomerulopathy - understanding a rare complement-driven renal disease. *Nature Reviews Nephrology*, **2019**, 15, 129-143	14.9	109
494	Gene therapy for hearing loss. *Human Molecular Genetics*, **2019**, 28, R65-R79	5.6	39
493	A proposal for comprehensive newborn hearing screening to improve identification of deaf and hard-of-hearing children. *Genetics in Medicine*, **2019**, 21, 2614-2630	8.1	36
492	Age at diagnosis, but not HPV type, is strongly associated with clinical course in recurrent respiratory papillomatosis. *PLoS ONE*, **2019**, 14, e0216697	3.7	29
491	International Pediatric Otolaryngology Group (IPOG) consensus recommendations: Diagnosis, pre-operative, operative and post-operative pediatric choanal atresia care. *International Journal of Pediatric Otorhinolaryngology*, **2019**, 123, 151-155	1.7	9
490	Pediatric Tracheostomy Decannulation: 11-Year Experience. *Otolaryngology - Head and Neck Surgery*, **2019**, 161, 499-506	5.5	8
489	ClinGen expert clinical validity curation of 164 hearing loss gene-disease pairs. *Genetics in Medicine*, **2019**, 21, 2239-2247	8.1	39
488	Facteurs de risque de s□it□de la papillomatose respiratoire r□idivante juv□ile lors de la premi□e endoscopie. *Annales Francaises D□Oto-Rhino-Laryngologie Et De Pathologie Cervico-Faciale*, **2019**, 136, 23-27	0	
487	Insights into the Biology of Hearing and Deafness Revealed by Single-Cell RNA Sequencing. *Cell Reports*, **2019**, 26, 3160-3171.e3	10.6	49
486	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
485	Factor B and C4b2a Autoantibodies in C3 Glomerulopathy. *Frontiers in Immunology*, **2019**, 10, 668	8.4	1
484	Targeted Allele Suppression Prevents Progressive Hearing Loss in the Mature Murine Model of Human TMC1 Deafness. *Molecular Therapy*, **2019**, 27, 681-690	11.7	41
483	Structural Insights into Hearing Loss Genetics from Polarizable Protein Repacking. *Biophysical Journal*, **2019**, 117, 602-612	2.9	4
482	Brief Report of Variants Detected in Hereditary Hearing Loss Cases in Iran over a 3-Year Period. *Iranian Journal of Public Health*, **2019**, 48, 1910-1915	0.7	
481	C3 Glomerulopathy **2019**, 633-646		
480	Peer Reviewers-Making the Annals What It Is. *Annals of Otology, Rhinology and Laryngology*, **2019**, 128, 1097	2.1	0

479	Risk factors for severity of juvenile-onset recurrent respiratory papillomatosis at first endoscopy. *European Annals of Otorhinolaryngology, Head and Neck Diseases*, **2019**, 136, 25-28	2.2	6
478	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. *Genetics in Medicine*, **2019**, 21, 948-954	8.1	26
477	The Epidemiology of Deafness. *Cold Spring Harbor Perspectives in Medicine*, **2019**, 9,	5.4	35
476	Open Access-Is There a Predator at the Door?. *JAMA Otolaryngology - Head and Neck Surgery*, **2018**, 144, 289-290	3.9	
475	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
474	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
473	Open Access: Is There a Predator at the Door?. *Laryngoscope*, **2018**,	3.6	1
472	C3 glomerulonephritis secondary to mutations in factors H and I: rapid recurrence in deceased donor kidney transplant effectively treated with eculizumab. *Nephrology Dialysis Transplantation*, **2018**, 33, 2260-2265	4.3	11
471	Exonic mutations and exon skipping: Lessons learned from DFNA5. *Human Mutation*, **2018**, 39, 433-440	4.7	36
470	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. *Human Molecular Genetics*, **2018**, 27, 780-798	5.6	31
469	C3 glomerulopathy associated with monoclonal Ig ĩs a distinct subtype. *Kidney International*, **2018**, 94, 178-186	9.9	51
468	Old gene, new phenotype: splice-altering variants in cause recessive non-syndromic hearing impairment. *Journal of Medical Genetics*, **2018**, 55, 555-560	5.8	16
467	Deficiency of complement factor H-related proteins and autoantibody-positive hemolytic uremic syndrome in an infant with combined partial deficiencies and autoantibodies to complement factor H and ADAMTS13. *CKJ: Clinical Kidney Journal*, **2018**, 11, 791-796	4.5	2
466	Recurrent Atypical Hemolytic Uremic Syndrome in Children With Acute Lymphoblastic Leukemia Undergoing Maintenance Chemotherapy. *Journal of Pediatric Hematology/Oncology*, **2018**, 40, 560-562	1.2	3
465	C3 Glomerulopathy: Ten YearsNExperience at Mayo Clinic. *Mayo Clinic Proceedings*, **2018**, 93, 991-1008	6.4	45
464	Intracellular Regulome Variability Along the Organ of Corti: Evidence, Approaches, Challenges, and Perspective. *Frontiers in Genetics*, **2018**, 9, 156	4.5	15
463	The authors reply. *Kidney International*, **2018**, 94, 632-633	9.9	
462	ATYPICAL HEMOLYTIC UREMIC SYNDROME AND C3 GLOMERULOPATHY: CONCLUSIONS FROM A «KIDNEY DISEASE: IMPROVING GLOBAL OUTCOMES» (KDIGO) CONTROVERSIES CONFERENCE. *Nephrology (Saint-Petersburg)*, **2018**, 22, 18-39	0.4	

461	Variants in CIB2 cause DFNB48 and not USH1J. *Clinical Genetics*, **2018**, 93, 812-821	4	36
460	Genetic Abnormalities in Complement Regulating Proteins in C3 Glomerulopathy. *American Journal of Clinical Pathology*, **2018**, 150, S131-S131	1.9	
459	In Vivo Electrocochleography in Hybrid Cochlear Implant Users Implicates TMPRSS3 in Spiral Ganglion Function. *Scientific Reports*, **2018**, 8, 14165	4.9	11
458	Genetic Analysis of 400 Patients Refines Understanding and Implicates a New Gene in Atypical Hemolytic Uremic Syndrome. *Journal of the American Society of Nephrology: JASN*, **2018**, 29, 2809-2819	12.7	31
457	Genomic Landscape and Mutational Signatures of Deafness-Associated Genes. *American Journal of Human Genetics*, **2018**, 103, 484-497	11	116
456	Comprehensive Genetic Testing for Deafness from Fresh and Archived Dried Blood Spots. *Otolaryngology - Head and Neck Surgery*, **2018**, 159, 1058-1060	5.5	2
455	Grxcr2 is required for stereocilia morphogenesis in the cochlea. *PLoS ONE*, **2018**, 13, e0201713	3.7	7
454	A novel mutation in ACTG1 causing Baraitser-Winter syndrome with extremely variable expressivity in three generations. *Ophthalmic Genetics*, **2017**, 38, 152-156	1.2	12
453	IgG4-related disease in an adolescent with radiologic-pathologic correlation. *Radiology Case Reports* , **2017**, 12, 196-199	1	2
452	Congenital hearing loss. *Nature Reviews Disease Primers*, **2017**, 3, 16094	51.1	184
451	Genetic variants in the peripheral auditory system significantly affect adult cochlear implant performance. *Hearing Research*, **2017**, 348, 138-142	3.9	39
450	Complement C3-Targeted Therapy: Replacing Long-Held Assertions with Evidence-Based Discovery. *Trends in Immunology*, **2017**, 38, 383-394	14.4	20
449	C3 glomerulonephritis with a severe crescentic phenotype. *Pediatric Nephrology*, **2017**, 32, 1625-1633	3.2	13
448	Common Elements in Rare Kidney Diseases: Conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. *Kidney International*, **2017**, 92, 796-808	9.9	25
447	Screening of deafness-causing DNA variants that are common in patients of European ancestry using a microarray-based approach. *PLoS ONE*, **2017**, 12, e0169219	3.7	20
446	Characterization of C3 in C3 glomerulopathy. *Nephrology Dialysis Transplantation*, **2017**, 32, 459-465	4.3	21
445	C4 Nephritic Factors in C3 Glomerulopathy: A Case Series. *American Journal of Kidney Diseases*, **2017**, 70, 834-843	7.4	35
444	Intravenous rAAV2/9 injection for murine cochlear gene delivery. *Scientific Reports*, **2017**, 7, 9609	4.9	18

443	Late Reoccurrence of Collapsing FSGS After Transplantation of a Living-Related Kidney Bearing APOL 1 Risk Variants Without Disease Evident in Donor Supports the Second Hit Hypothesis. *Transplantation Direct*, **2017**, 3, e185	2.3	3
442	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. *EMBO Molecular Medicine*, **2017**, 9, 1711-1731	12	39
441	C3 Glomerulopathy **2017**, 1-14		1
440	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
439	C3 glomerulonephritis and autoimmune disease: more than a fortuitous association?. *Journal of Nephrology*, **2016**, 29, 203-209	4.8	15
438	Complement inhibition in C3 glomerulopathy. *Seminars in Immunology*, **2016**, 28, 241-9	10.7	32
437	Navigating genetic diagnostics in patients with hearing loss. *Current Opinion in Pediatrics*, **2016**, 28, 705-712	3.2	20
436	Ketogenic diet - A novel treatment for early epileptic encephalopathy due to PIGA deficiency. *Brain and Development*, **2016**, 38, 848-51	2.2	24
435	Audioprofile Surfaces: The 21st Century Audiogram. *Annals of Otology, Rhinology and Laryngology*, **2016**, 125, 361-8	2.1	6
434	Diagnosis of complement alternative pathway disorders. *Kidney International*, **2016**, 89, 278-88	9.9	50
433	Discontinuation of dialysis with eculizumab therapy in a pediatric patient with dense deposit disease. *Pediatric Nephrology*, **2016**, 31, 683-7	3.2	9
432	Comprehensive genetic testing in the clinical evaluation of 1119 patients with hearing loss. *Human Genetics*, **2016**, 135, 441-450	6.3	250
431	C4 Glomerulopathy: A Disease Entity Associated With ´C4d´Deposition. *American Journal of Kidney Diseases*, **2016**, 67, 949-53	7.4	17
430	Deafness **2016**, 197-201		
429	Mutations in Complement Factor H Impair Alternative Pathway Regulation on Mouse Glomerular Endothelial Cells in Vitro. *Journal of Biological Chemistry*, **2016**, 291, 4974-81	5.4	15
428	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
427	Heterogeneity of Hereditary Hearing Loss in Iran: a Comprehensive Review. *Archives of Iranian Medicine*, **2016**, 19, 720-728	2.4	16
426	Reducing the Cost of the Diagnostic Odyssey in Early Onset Epileptic Encephalopathies. *BioMed Research International*, **2016**, 2016, 6421039	3	36

425	International Pediatric ORL Group (IPOG) laryngomalacia consensus recommendations. *International Journal of Pediatric Otorhinolaryngology*, **2016**, 86, 256-61	1.7	34
424	Detection and Confirmation of Deafness-Causing Copy Number Variations in the STRC Gene by Massively Parallel Sequencing and Comparative Genomic Hybridization. *Annals of Otology, Rhinology and Laryngology*, **2016**, 125, 918-923	2.1	21
423	RNA Interference Prevents Autosomal-Dominant Hearing Loss. *American Journal of Human Genetics* , **2016**, 98, 1101-1113	11	66
422	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
421	Familial C3 glomerulonephritis caused by a novel CFHR5-CFHR2 fusion gene. *Molecular Immunology* , **2016**, 77, 89-96	4.3	34
420	Reconstruction of the pediatric midface following oncologic resection. *Journal of Reconstructive Microsurgery*, **2015**, 31, 336-42	2.5	10
419	Sensorineural Hearing Loss: A Changing Paradigm for Its Evaluation. *Otolaryngology - Head and Neck Surgery*, **2015**, 153, 843-850	5.5	15
418	Compstatin analog Cp40 inhibits complement dysregulation in vitro in C3 glomerulopathy. *Immunobiology*, **2015**, 220, 993-8	3.4	42
417	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
416	HOMER2, a stereociliary scaffolding protein, is essential for normal hearing in humans and mice. *PLoS Genetics*, **2015**, 11, e1005137	6	38
415	De novo mutation in X-linked hearing loss-associated POU3F4 in a sporadic case of congenital hearing loss. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 169S-76S	2.1	16
414	Novel PTPRQ mutations identified in three congenital hearing loss patients with various types of hearing loss. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 184S-92S	2.1	12
413	Mutations in LOXHD1 gene cause various types and severities of hearing loss. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 135S-41S	2.1	22
412	USH2 caused by GPR98 mutation diagnosed by massively parallel sequencing in advance of the occurrence of visual symptoms. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 123S-8S	2.1	8
411	The role of complement in C3 glomerulopathy. *Molecular Immunology*, **2015**, 67, 21-30	4.3	65
410	Atypical aHUS: State of the art. *Molecular Immunology*, **2015**, 67, 31-42	4.3	177
409	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. *Journal of Medical Genetics*, **2015**, 52, 823-9	5.8	72
408	Mutations in apoptosis-inducing factor cause X-linked recessive auditory neuropathy spectrum disorder. *Journal of Medical Genetics*, **2015**, 52, 523-31	5.8	62

407	Challenges and solutions for gene identification in the presence of familial locus heterogeneity. *European Journal of Human Genetics*, **2015**, 23, 1207-15	5.3	29
406	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 821-30	5.3	19
405	Mapping interactions between complement C3 and regulators using mutations in atypical hemolytic uremic syndrome. *Blood*, **2015**, 125, 2359-69	2.2	79
404	PDZD7 and hearing loss: More than just a modifier. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2957-65	2.5	45
403	C3 glomerulonephritis associated with complement factor B mutation. *American Journal of Kidney Diseases*, **2015**, 65, 520-1	7.4	10
402	Mutation of the nuclear lamin gene LMNB2 in progressive myoclonus epilepsy with early ataxia. *Human Molecular Genetics*, **2015**, 24, 4483-90	5.6	33
401	Superficial temporal artery and vein as recipient vessels for scalp and facial reconstruction: radiographic support for underused vessels. *Journal of Reconstructive Microsurgery*, **2015**, 31, 249-53	2.5	13
400	Hearing loss caused by a P2RX2 mutation identified in a MELAS family with a coexisting mitochondrial 3243AG mutation. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 177S-83S	2.1	13
399	Soluble c5b-9 as a biomarker for complement activation in atypical hemolytic uremic syndrome. *American Journal of Kidney Diseases*, **2015**, 65, 968-9	7.4	29
398	Overlap of ultrastructural findings in C3 glomerulonephritis and dense deposit disease. *Kidney International*, **2015**, 88, 1449-1450	9.9	4
397	TBC1D24 mutation causes autosomal-dominant nonsyndromic hearing loss. *Human Mutation*, **2014**, 35, 819-23	4.7	69
396	C3 glomerulopathy: the genetic and clinical findings in dense deposit disease and C3 glomerulonephritis. *Seminars in Thrombosis and Hemostasis*, **2014**, 40, 465-71	5.3	40
395	Defining the complement biomarker profile of C3 glomerulopathy. *Clinical Journal of the American Society of Nephrology: CJASN*, **2014**, 9, 1876-82	6.9	53
394	Cordova: web-based management of genetic variation data. *Bioinformatics*, **2014**, 30, 3438-9	7.2	2
393	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
392	Copy number variants are a common cause of non-syndromic hearing loss. *Genome Medicine*, **2014**, 6, 37	14.4	111
391	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
390	Preoperative evaluation of thyroglossal duct cysts: children versus adults--is there a difference?. *American Journal of Surgery*, **2014**, 207, 902-6	2.7	2

389	Generation of multiple fluid-phase C3b:plasma protein complexes during complement activation: possible implications in C3 glomerulopathies. *Journal of Immunology*, **2014**, 192, 1220-30	5.3	13
388	C4 dense-deposit disease. *New England Journal of Medicine*, **2014**, 370, 784-6	59.2	22
387	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
386	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
385	Cav3.2 T-type calcium channel is required for the NFAT-dependent Sox9 expression in tracheal cartilage. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, E1990-8	11.5	42
384	Rare variants in BMP2 and BMP4 found in otosclerosis patients reduce Smad signaling. *Otology and Neurotology*, **2014**, 35, 395-400	2.6	6
383	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
382	Streptococcal infection as possible trigger for dense deposit disease (C3 glomerulopathy). *European Journal of Pediatrics*, **2014**, 173, 767-72	4.1	22
381	Atypical haemolytic-uraemic syndrome due to heterozygous mutations of CFH/CFHR1-3 and complement factor H 479. *Blood Transfusion*, **2014**, 12, 111-3	3.6	6
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