

# Richard J H Smith

## List of Publications by Year in descending order

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605  
papers

34,924  
citations

3116

95  
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7427

157  
g-index

629  
all docs

629  
docs citations

629  
times ranked

24101  
citing authors

#	ARTICLE	IF	CITATIONS
1	Pembrolizumab Induced Acute Persistent Airway Disease in a Patient with Recurrent Respiratory Papillomatosis (RRP). <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2022, 131, 331-336.	0.6	3
2	Genetic Causes of Hearing Loss in a Large Cohort of Cochlear Implant Recipients. <i>Otolaryngology - Head and Neck Surgery</i> , 2022, 166, 734-737.	1.1	17
3	Clinical Pharmacogenetics Implementation Consortium Guideline for the Use of Aminoglycosides Based on <i>MTâ€RNR1</i> Genotype. <i>Clinical Pharmacology and Therapeutics</i> , 2022, 111, 366-372.	2.3	50
4	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. <i>Human Genetics</i> , 2022, 141, 853-863.	1.8	7
5	COCH-related autosomal dominant nonsyndromic hearing loss: a phenotypeâ€“genotype study. <i>Human Genetics</i> , 2022, 141, 889-901.	1.8	7
6	Improving Clinical Trials for Anticomplement Therapies in Complement-Mediated Glomerulopathies: Report of a Scientific Workshop Sponsored by the National Kidney Foundation. <i>American Journal of Kidney Diseases</i> , 2022, 79, 570-581.	2.1	15
7	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. <i>Genetics in Medicine</i> , 2022, 24, 130-145.	1.1	45
8	AudioGene: refining the natural history of <i>KCNQ4</i> , <i>GSDME</i> , <i>WFS1</i> , and <i>COCH</i> -associated hearing loss. <i>Human Genetics</i> , 2022, , 1.	1.8	6
9	DVPred: a disease-specific prediction tool for variant pathogenicity classification for hearing loss. <i>Human Genetics</i> , 2022, 141, 401-411.	1.8	6
10	The voltage-gated Ca <sup>2+</sup> channel subunit $\hat{1}\pm 2\hat{1}$ -4 regulates locomotor behavior and sensorimotor gating in mice. <i>PLoS ONE</i> , 2022, 17, e0263197.	1.1	5
11	The hearing-impaired patient: what the future holds. <i>Human Genetics</i> , 2022, 141, 307-310.	1.8	1
12	Complement Factor I Variants in Complement-Mediated Renal Diseases. <i>Frontiers in Immunology</i> , 2022, 13, .	2.2	1
13	Results from a nationwide retrospective cohort measure the impact of C3 and soluble C5b-9 levels on kidney outcomes in C3 glomerulopathy. <i>Kidney International</i> , 2022, 102, 904-916.	2.6	12
14	<scp>C3</scp> glomerulopathy: Understanding an ultraâ€“rare complementâ€“mediated renal disease. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2022, 190, 344-357.	0.7	15
15	Rigid Video Laryngoscopy for Intubation in Severe Pierre Robin Sequence: A Retrospective Review. <i>Laryngoscope</i> , 2021, 131, 1647-1651.	1.1	4
16	Neonatal Lateral Epiglottic Defects. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2021, 130, 311-313.	0.6	1
17	Consortium of Otolaryngology Journal Editors: Collegiality and Contributions. <i>Journal of Voice</i> , 2021, 35, 170-171.	0.6	0
18	What Is the Best Approach to Prevent Advancedâ€“Stage Pressure Injuries After Pediatric Tracheotomy?. <i>Laryngoscope</i> , 2021, 131, 1196-1197.	1.1	0

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19	International Pediatric Otolaryngology Group (IPOG) management recommendations: Pediatric tracheostomy decannulation. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2021, 141, 110565.	0.4	9
20	Targeted broad-based genetic testing by next-generation sequencing informs diagnosis and facilitates management in patients with kidney diseases. <i>Nephrology Dialysis Transplantation</i> , 2021, 36, 295-305.	0.4	34
21	Systemic Bevacizumab for Treatment of Respiratory Papillomatosis: International Consensus Statement. <i>Laryngoscope</i> , 2021, 131, E1941-E1949.	1.1	24
22	A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. <i>European Journal of Human Genetics</i> , 2021, 29, 988-997.	1.4	8
23	International Pediatric Otolaryngology Group (IPOG) survey: Efforts to avoid complications in home tracheostomy care. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2021, 141, 110563.	0.4	5
24	Exome sequencing utility in defining the genetic landscape of hearing loss and novel gene discovery in Iran. <i>Clinical Genetics</i> , 2021, 100, 59-78.	1.0	4
25	Systematic and Other Reviews: Criteria and Complexities. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2021, 130, 649-652.	0.6	3
26	Systematic and Other Reviews: Criteria and Complexities. <i>Laryngoscope</i> , 2021, 131, 1443-1445.	1.1	1
27	<sc><i>TSPEAR</i></sc> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2417-2433.	0.7	10
28	MO136RELATIONSHIP BETWEEN UPCR AND EGFR IN C3 GLOMERULOPATHY. <i>Nephrology Dialysis Transplantation</i> , 2021, 36, .	0.4	0
29	Systematic and Other Reviews: Criteria and Complexities. <i>Journal of Neurological Surgery, Part B: Skull Base</i> , 2021, 82, 273-276.	0.4	0
30	Systematic and Other Reviews: Criteria and Complexities. <i>American Journal of Rhinology and Allergy</i> , 2021, 35, 412-416.	1.0	0
31	Functional characterization of 105 factor H variants associated with aHUS: lessons for variant classification. <i>Blood</i> , 2021, 138, 2185-2201.	0.6	29
32	CFH and CFHR Copy Number Variations in C3 Glomerulopathy and Immune Complex-Mediated Membranoproliferative Glomerulonephritis. <i>Frontiers in Genetics</i> , 2021, 12, 670727.	1.1	11
33	Systematic and other reviews: Criteria and complexities. <i>Head and Neck</i> , 2021, 43, 1979-1982.	0.9	1
34	Systematic and other reviews: Criteria and complexities. <i>Journal of Otolaryngology - Head and Neck Surgery</i> , 2021, 50, 41.	0.9	0
35	Systematic and Other Reviews: Criteria and Complexities. <i>Journal of Voice</i> , 2021, 35, 509-511.	0.6	0
36	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. <i>Genetics in Medicine</i> , 2021, 23, 2208-2212.	1.1	18

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37	Systematic and other reviews: criteria and complexities. Journal of Laryngology and Otology, 2021, 135, 565-567.	0.4	0
38	Systematic and Other Reviews: Criteria and Complexities. Ear, Nose and Throat Journal, 2021, 100, 403-406.	0.4	0
39	Systematic and other reviews: Criteria and complexities. World Journal of Otorhinolaryngology - Head and Neck Surgery, 2021, 7, 236-239.	0.7	3
40	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.	2.2	4
41	Systematic and other reviews: Criteria and complexities. International Journal of Pediatric Otorhinolaryngology, 2021, 147, 110640.	0.4	2
42	Systematic and other reviews: Criteria and complexities. American Journal of Otolaryngology - Head and Neck Medicine and Surgery, 2021, 42, 102957.	0.6	0
43	Monoclonal Gammopathy of Renal Significance Causes C3 Glomerulonephritis Via Monoclonal IgG Kappa Inhibition of Complement Factor H. Kidney International Reports, 2021, 6, 2505-2509.	0.4	4
44	Genetic testing hearing loss: The challenge of non syndromic mimics. International Journal of Pediatric Otorhinolaryngology, 2021, 150, 110872.	0.4	6
45	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human Genetics, 2021, 140, 915-931.	1.8	16
46	Sequential genetic testing of living-related donors for inherited renal disease to promote informed choice and enhance safety of living donation. Transplant International, 2021, 34, 2696-2705.	0.8	7
47	International Pediatric Otolaryngology Group (IPOG): Juvenile-onset recurrent respiratory papillomatosis consensus recommendations. International Journal of Pediatric Otorhinolaryngology, 2020, 128, 109697.	0.4	21
48	Advanced practice providers and children's hospital-based pediatric otolaryngology practices. International Journal of Pediatric Otorhinolaryngology, 2020, 129, 109770.	0.4	8
49	International Pediatric ORL Group (IPOG) Robin Sequence consensus recommendations. International Journal of Pediatric Otorhinolaryngology, 2020, 130, 109855.	0.4	10
50	International Pediatric Otolaryngology Group (IPOG) Consensus Recommendations: Congenital Cholesteatoma. Otology and Neurotology, 2020, 41, 345-351.	0.7	13
51	A recurrent missense variant in HARS2 results in variable sensorineural hearing loss in three unrelated families. Journal of Human Genetics, 2020, 65, 305-311.	1.1	5
52	Future directions for screening and treatment in congenital hearing loss. Precision Clinical Medicine, 2020, 3, 175-186.	1.3	20
53	International Pediatric Otolaryngology Group (IPOG) consensus recommendations: Management of suprastomal collapse in the pediatric population. International Journal of Pediatric Otorhinolaryngology, 2020, 139, 110427.	0.4	4
54	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.	0.3	14

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55	Consortium of Otolaryngology Journal Editors: Collegiality and contributions. International Journal of Pediatric Otorhinolaryngology, 2020, 134, 109959.	0.4	0
56	Consortium of Otolaryngology Journal Editors: Collegiality and contributions. International Forum of Allergy and Rhinology, 2020, 10, 698-699.	1.5	1
57	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.	0.4	18
58	Consortium of Otolaryngology Journal Editors: Collegiality and Contributions. American Journal of Rhinology and Allergy, 2020, 34, 321-323.	1.0	0
59	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.	0.4	38
60	Consortium of Otolaryngology Journal Editors: Collegiality and Contributions. Otolaryngology - Head and Neck Surgery, 2020, 163, 1067-1069.	1.1	0
61	Consortium of Otolaryngology Journal Editors: collegiality and contributions. Journal of Laryngology and Otology, 2020, 134, 379-380.	0.4	0
62	Factor H Autoantibodies and Complement-Mediated Diseases. Frontiers in Immunology, 2020, 11, 607211.	2.2	20
63	Consortium of otolaryngology journal editors: Collegiality and contributions. Clinical Otolaryngology, 2020, 45, 313-315.	0.6	0
64	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. Human Genetics, 2020, 139, 1315-1323.	1.8	12
65	Consortium of Otolaryngology Journal Editorsâ€™ Collegiality and Contributions. JAMA Otolaryngology - Head and Neck Surgery, 2020, 146, 521.	1.2	1
66	Consortium of Otolaryngology Journal Editors: Collegiality and Contributions. Laryngoscope, 2020, 130, 1357-1358.	1.1	0
67	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.	2.6	10
68	DFNA5 (GSDME) c.991-15_991-13delTTC: Founder Mutation or Mutational Hotspot?. International Journal of Molecular Sciences, 2020, 21, 3951.	1.8	8
69	Novel loss-of-function mutations in COCH cause autosomal recessive nonsyndromic hearing loss. Human Genetics, 2020, 139, 1565-1574.	1.8	13
70	Consortium of Otolaryngology Journal Editors: Collegiality and Contributions. Ear, Nose and Throat Journal, 2020, 100, 014556132091288.	0.4	0
71	Initial experience from a renal genetics clinic demonstrates a distinct role in patient management. Genetics in Medicine, 2020, 22, 1025-1035.	1.1	45
72	Consortium of Otolaryngology Journal Editors: Collegiality and Contributions. Annals of Otology, Rhinology and Laryngology, 2020, 129, 533-535.	0.6	0

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73	Consortium of otolaryngology journal editors: Collegiality and contributions. Operative Techniques in Otolaryngology - Head and Neck Surgery, 2020, 31, 71-73.	0.1	0
74	Hair Cell Transduction Efficiency of Single- and Dual-AAV Serotypes in Adult Murine Cochleae. Molecular Therapy - Methods and Clinical Development, 2020, 17, 1167-1177.	1.8	27
75	Consortium of Otolaryngology Journal Editors: Collegiality and contributions. American Journal of Otolaryngology - Head and Neck Medicine and Surgery, 2020, 41, 102430.	0.6	0
76	Genetic Testing for Congenital Bilateral Hearing Loss in the Context of Targeted Cytomegalovirus Screening. Laryngoscope, 2020, 130, 2714-2718.	1.1	12
77	Proteomic Analysis of Complement Proteins in Membranous Nephropathy. Kidney International Reports, 2020, 5, 618-626.	0.4	51
78	Insights into the pathophysiology of DFNA10 hearing loss associated with novel EYA4 variants. Scientific Reports, 2020, 10, 6213.	1.6	15
79	Is it Usher syndrome? Collaborative diagnosis and molecular genetics of patients with visual impairment and hearing loss. Ophthalmic Genetics, 2020, 41, 151-158.	0.5	7
80	C3(H2O) prevents rescue of complement-mediated C3 glomerulopathy in Cfh <sup>-/-</sup> Cfd <sup>-/-</sup> mice. JCI Insight, 2020, 5, .	2.3	13
81	Structural Insights into Hearing Loss Genetics from Polarizable Protein Repacking. Biophysical Journal, 2019, 117, 602-612.	0.2	12
82	C3 glomerulopathy â€” understanding a rare complement-driven renal disease. Nature Reviews Nephrology, 2019, 15, 129-143.	4.1	223
83	Gene therapy for hearing loss. Human Molecular Genetics, 2019, 28, R65-R79.	1.4	78
84	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
85	Age at diagnosis, but not HPV type, is strongly associated with clinical course in recurrent respiratory papillomatosis. PLoS ONE, 2019, 14, e0216697.	1.1	43
86	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.	0.4	23
87	Pediatric Tracheostomy Decannulation: 11â€”Year Experience. Otolaryngology - Head and Neck Surgery, 2019, 161, 499-506.	1.1	18
88	ClinGen expert clinical validity curation of 164 hearing loss geneâ€”disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	1.1	67
89	Insights into the Biology of Hearing and Deafness Revealed by Single-Cell RNA Sequencing. Cell Reports, 2019, 26, 3160-3171.e3.	2.9	137
90	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.	3.3	116

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91	Factor B and C4b2a Autoantibodies in C3 Glomerulopathy. <i>Frontiers in Immunology</i> , 2019, 10, 668.	2.2	4
92	Targeted Allele Suppression Prevents Progressive Hearing Loss in the Mature Murine Model of Human TMC1 Deafness. <i>Molecular Therapy</i> , 2019, 27, 681-690.	3.7	66
93	C3 Glomerulopathy. , 2019, , 633-646.		0
94	Peer Reviewersâ€™ Making the Annals What It Is. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2019, 128, 1097-1097.	0.6	2
95	Risk factors for severity of juvenile-onset recurrent respiratory papillomatosis at first endoscopy. <i>European Annals of Otorhinolaryngology, Head and Neck Diseases</i> , 2019, 136, 25-28.	0.4	9
96	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. <i>Genetics in Medicine</i> , 2019, 21, 948-954.	1.1	36
97	The Epidemiology of Deafness. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2019, 9, a033258.	2.9	78
98	Brief Report of Variants Detected in Hereditary Hearing Loss Cases in Iran over a 3-Year Period. <i>Iranian Journal of Public Health</i> , 2019, 48, 1910-1915.	0.3	1
99	Open access: is there a predator at the door?. <i>Journal of Laryngology and Otolaryngology</i> , 2018, 132, 189-190.	0.4	0
100	Open Access: Is There a Predator at the Door?. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2018, 127, 137-138.	0.6	1
101	Open Accessâ€™ Is There a Predator at the Door?. <i>JAMA Otolaryngology - Head and Neck Surgery</i> , 2018, 144, 289.	1.2	0
102	Statistical Validation of Rare Complement Variants Provides Insights into the Molecular Basis of Atypical Hemolytic Uremic Syndrome and C3 Glomerulopathy. <i>Journal of Immunology</i> , 2018, 200, 2464-2478.	0.4	130
103	Enhanced viral-mediated cochlear gene delivery in adult mice by combining canal fenestration with round window membrane inoculation. <i>Scientific Reports</i> , 2018, 8, 2980.	1.6	92
104	Open Access: Is There a Predator at the Door?. <i>Otology and Neurotology</i> , 2018, 39, 271-272.	0.7	0
105	C3 glomerulonephritis secondary to mutations in factors H and I: rapid recurrence in deceased donor kidney transplant effectively treated with eculizumab. <i>Nephrology Dialysis Transplantation</i> , 2018, 33, 2260-2265.	0.4	17
106	Open Access: Is There a Predator at the Door?. <i>Journal of Voice</i> , 2018, 32, 1-2.	0.6	2
107	Open access: is there a predator at the door?. <i>International Forum of Allergy and Rhinology</i> , 2018, 8, 81-82.	1.5	2
108	Exonic mutations and exon skipping: Lessons learned from <i>DFNA5</i>. <i>Human Mutation</i> , 2018, 39, 433-440.	1.1	44

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109	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. <i>Human Molecular Genetics</i> , 2018, 27, 780-798.	1.4	49
110	C3 glomerulopathy associated with monoclonal IgA is a distinct subtype. <i>Kidney International</i> , 2018, 94, 178-186.	2.6	77
111	Open access: Is there a predator at the door?. <i>Laryngoscope</i> , 2018, 128, 1255-1256.	1.1	0
112	Old gene, new phenotype: splice-altering variants in <i>CEACAM16</i> cause recessive non-syndromic hearing impairment. <i>Journal of Medical Genetics</i> , 2018, 55, 555-560.	1.5	48
113	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. <i>Clinical Kidney Journal</i> , 2018, 11, 791-796.	1.4	3
114	Open Access: Is There a Predator at the Door?. <i>Journal of Neurological Surgery, Part B: Skull Base</i> , 2018, 79, 115-116.	0.4	1
115	Recurrent Atypical Hemolytic Uremic Syndrome in Children With Acute Lymphoblastic Leukemia Undergoing Maintenance Chemotherapy. <i>Journal of Pediatric Hematology/Oncology</i> , 2018, 40, 560-562.	0.3	7
116	Variants in <i>CIB2</i> cause DFNB48 and not USH1J. <i>Clinical Genetics</i> , 2018, 93, 812-821.	1.0	46
117	Open Access: Is There a Predator at the Door?. <i>Ear, Nose and Throat Journal</i> , 2018, 97, 10-12.	0.4	14
118	Genetic Abnormalities in Complement Regulating Proteins in C3 Glomerulopathy. <i>American Journal of Clinical Pathology</i> , 2018, 150, S131-S131.	0.4	0
119	In Vivo Electrocochleography in Hybrid Cochlear Implant Users Implicates <i>TMPRSS3</i> in Spiral Ganglion Function. <i>Scientific Reports</i> , 2018, 8, 14165.	1.6	25
120	Genetic Analysis of 400 Patients Refines Understanding and Implicates a New Gene in Atypical Hemolytic Uremic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2809-2819.	3.0	50
121	Genomic Landscape and Mutational Signatures of Deafness-Associated Genes. <i>American Journal of Human Genetics</i> , 2018, 103, 484-497.	2.6	214
122	Comprehensive Genetic Testing for Deafness from Fresh and Archived Dried Blood Spots. <i>Otolaryngology - Head and Neck Surgery</i> , 2018, 159, 1058-1060.	1.1	3
123	<i>Grxcr2</i> is required for stereocilia morphogenesis in the cochlea. <i>PLoS ONE</i> , 2018, 13, e0201713.	1.1	11
124	C3 Glomerulopathy: Ten Years' Experience at Mayo Clinic. <i>Mayo Clinic Proceedings</i> , 2018, 93, 991-1008.	1.4	82
125	Intracellular Regulome Variability Along the Organ of Corti: Evidence, Approaches, Challenges, and Perspective. <i>Frontiers in Genetics</i> , 2018, 9, 156.	1.1	17
126	The authors reply. <i>Kidney International</i> , 2018, 94, 632-633.	2.6	0

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127	Open Access: Is There a Predator at the Door?. Otolaryngology - Head and Neck Surgery, 2018, 158, 401-402.	1.1	4
128	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.	0.1	0
129	Characterization of C3 in C3 glomerulopathy. Nephrology Dialysis Transplantation, 2017, 32, gfw290.	0.4	29
130	A novel mutation in <i>ACTG1</i> causing Baraitser-Winter syndrome with extremely variable expressivity in three generations. Ophthalmic Genetics, 2017, 38, 152-156.	0.5	16
131	IgG4-related disease in an adolescent with radiologic-pathologic correlation. Radiology Case Reports, 2017, 12, 196-199.	0.2	5
132	Congenital hearing loss. Nature Reviews Disease Primers, 2017, 3, 16094.	18.1	328
133	Genetic variants in the peripheral auditory system significantly affect adult cochlear implant performance. Hearing Research, 2017, 348, 138-142.	0.9	68
134	Complement C3-Targeted Therapy: Replacing Long-Held Assertions with Evidence-Based Discovery. Trends in Immunology, 2017, 38, 383-394.	2.9	31
135	C3 glomerulonephritis with a severe crescentic phenotype. Pediatric Nephrology, 2017, 32, 1625-1633.	0.9	15
136	Common Elements in Rare Kidney Diseases: Conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney International, 2017, 92, 796-808.	2.6	40
137	C4 Nephritic Factors in C3 Glomerulopathy: A Case Series. American Journal of Kidney Diseases, 2017, 70, 834-843.	2.1	45
138	Intravenous rAAV2/9 injection for murine cochlear gene delivery. Scientific Reports, 2017, 7, 9609.	1.6	31
139	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.	0.8	3
140	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. EMBO Molecular Medicine, 2017, 9, 1711-1731.	3.3	66
141	Screening of deafness-causing DNA variants that are common in patients of European ancestry using a microarray-based approach. PLoS ONE, 2017, 12, e0169219.	1.1	26
142	C3 Glomerulopathy., 2017,, 1-14.		1
143	Reducing the Cost of the Diagnostic Odyssey in Early Onset Epileptic Encephalopathies. BioMed Research International, 2016, 2016, 1-8.	0.9	45
144	International Pediatric ORL Group (IPOG) laryngomalacia consensus recommendations. International Journal of Pediatric Otorhinolaryngology, 2016, 86, 256-261.	0.4	70

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145	Detection and Confirmation of Deafness-Causing Copy Number Variations in the <i>STRC</i> Gene by Massively Parallel Sequencing and Comparative Genomic Hybridization. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2016, 125, 918-923.	0.6	28
146	RNA Interference Prevents Autosomal-Dominant Hearing Loss. <i>American Journal of Human Genetics</i> , 2016, 98, 1101-1113.	2.6	95
147	International Pediatric Otolaryngology Group (IPOG) consensus recommendations: Hearing loss in the pediatric patient. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2016, 90, 251-258.	0.4	88
148	Familial C3 glomerulonephritis caused by a novel CFHR5-CFHR2 fusion gene. <i>Molecular Immunology</i> , 2016, 77, 89-96.	1.0	41
149	Complement inhibition in C3 glomerulopathy. <i>Seminars in Immunology</i> , 2016, 28, 241-249.	2.7	40
150	Navigating genetic diagnostics in patients with hearing loss. <i>Current Opinion in Pediatrics</i> , 2016, 28, 705-712.	1.0	33
151	Ketogenic diet – A novel treatment for early epileptic encephalopathy due to PIGA deficiency. <i>Brain and Development</i> , 2016, 38, 848-851.	0.6	34
152	Audioprofile Surfaces. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2016, 125, 361-368.	0.6	8
153	Diagnosis of complement alternative pathway disorders. <i>Kidney International</i> , 2016, 89, 278-288.	2.6	74
154	Discontinuation of dialysis with eculizumab therapy in a pediatric patient with dense deposit disease. <i>Pediatric Nephrology</i> , 2016, 31, 683-687.	0.9	12
155	Comprehensive genetic testing in the clinical evaluation of 1119 patients with hearing loss. <i>Human Genetics</i> , 2016, 135, 441-450.	1.8	373
156	C4 Glomerulopathy: A Disease Entity Associated With C4d Deposition. <i>American Journal of Kidney Diseases</i> , 2016, 67, 949-953.	2.1	23
157	Deafness. , 2016, , 197-201.		0
158	Mutations in Complement Factor H Impair Alternative Pathway Regulation on Mouse Glomerular Endothelial Cells in Vitro. <i>Journal of Biological Chemistry</i> , 2016, 291, 4974-4981.	1.6	18
159	Mayo Clinic/Renal Pathology Society Consensus Report on Pathologic Classification, Diagnosis, and Reporting of GN. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 1278-1287.	3.0	210
160	High-Throughput Genetic Testing for Thrombotic Microangiopathies and C3 Glomerulopathies. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 1245-1253.	3.0	89
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255	AudioGene: Computer-based prediction of genetic factors involved in non-syndromic hearing impairment. , 2011, , .		0
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281	Mutations in <i>Grxcr1</i> Are The Basis for Inner Ear Dysfunction in the Pirouette Mouse. <i>American Journal of Human Genetics</i> , 2010, 86, 148-160.	2.6	49
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299	Membranoproliferative Glomerulonephritis Secondary to Monoclonal Gammopathy. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2010, 5, 770-782.	2.2	156
300	Genetic Sensorineural Hearing Loss. , 2010, , 2086-2099.		1
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303	Therapeutic Regulation of Gene Expression in the Inner Ear using RNA Interference. <i>Advances in Oto-Rhino-Laryngology</i> , 2009, 66, 13-36.	1.6	33
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306	Recurrent Atypical Hemolytic Uremic Syndrome Associated With Factor I Mutation in a Living Related Renal Transplant Recipient. <i>American Journal of Kidney Diseases</i> , 2009, 53, 321-326.	2.1	29

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