Heon Yung Gee

List of Publications by Year
in descending order

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114 papers	7,768 citations	66343 42 h-index	54911 84 g-index
120 all docs	120 docs citations	120 times ranked	11773 citing authors

#	Article	IF	Citations
1	Autistic-like social behaviour in Shank2-mutant mice improved by restoring NMDA receptor function. Nature, 2012, 486, 261-265.	27.8	604
2	A Single-Gene Cause in 29.5% of Cases of Steroid-Resistant Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2015, 26, 1279-1289.	6.1	499
3	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	28.9	347
4	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. Journal of Clinical Investigation, 2013, 123, 5179-5189.	8.2	275
5	Rescue of Î”F508-CFTR Trafficking via a GRASP-Dependent Unconventional Secretion Pathway. Cell, 2011, 146, 746-760.	28.9	274
6	SGLT2 inhibition modulates NLRP3 inflammasome activity via ketones and insulin in diabetes with cardiovascular disease. Nature Communications, 2020, 11, 2127.	12.8	263
7	FAN1 mutations cause karyomegalic interstitial nephritis, linking chronic kidney failure to defective DNA damage repair. Nature Genetics, 2012, 44, 910-915.	21.4	205
8	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
9	ARHGDIA mutations cause nephrotic syndrome via defective RHO GTPase signaling. Journal of Clinical Investigation, 2013, 123, 3243-3253.	8.2	196
10	Mutations in RSPH1 Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. American Journal of Respiratory and Critical Care Medicine, 2014, 189, 707-717.	5.6	191
11	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 672-686.	6.2	184
12	ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. American Journal of Human Genetics, 2013, 93, 336-345.	6.2	183
13	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2018, 13, 53-62.	4.5	170
14	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	21.4	164
15	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. Journal of Clinical Investigation, 2017, 127, 912-928.	8.2	160
16	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. Journal of Clinical Investigation, 2015, 125, 2375-2384.	8.2	159
17	Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. Nature Genetics, 2016, 48, 457-465.	21.4	149
18	Mutations in DZIP1L, which encodes a ciliary-transition-zone protein, cause autosomal recessive polycystic kidney disease. Nature Genetics, 2017, 49, 1025-1034.	21.4	148

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19	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. American Journal of Human Genetics, 2013, 93, 711-720.	6.2	135
20	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213.	5.2	133
21	Cholesterol modulates cell signaling and protein networking by specifically interacting with PDZ domain-containing scaffold proteins. Nature Communications, 2012, 3, 1249.	12.8	129
22	Whole-exome resequencing reveals recessive mutations in TRAP1 in individuals with CAKUT and VACTERL association. Kidney International, 2014, 85, 1310-1317.	5.2	106
23	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 664-672.	4.5	105
24	Mutations in EMP2 Cause Childhood-Onset Nephrotic Syndrome. American Journal of Human Genetics, 2014, 94, 884-890.	6.2	101
25	FAT1 mutations cause a glomerulotubular nephropathy. Nature Communications, 2016, 7, 10822.	12.8	99
26	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. American Journal of Human Genetics, 2015, 96, 81-92.	6.2	98
27	A Small Molecule That Binds to an ATPase Domain of Hsc70 Promotes Membrane Trafficking of Mutant Cystic Fibrosis Transmembrane Conductance Regulator. Journal of the American Chemical Society, 2011, 133, 20267-20276.	13.7	93
28	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. American Journal of Human Genetics, 2014, 94, 905-914.	6.2	90
29	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. Nature Communications, 2018, 9, 1960.	12.8	90
30	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	8.2	89
31	Defects of CRB2 Cause Steroid-Resistant Nephrotic Syndrome. American Journal of Human Genetics, 2015, 96, 153-161.	6.2	88
32	A Multi-layered Quantitative InÂVivo Expression Atlas of the Podocyte Unravels Kidney Disease Candidate Genes. Cell Reports, 2018, 23, 2495-2508.	6.4	81
33	Unconventional protein secretion â€“ new insights into the pathogenesis and therapeutic targets of human diseases. Journal of Cell Science, 2018, 131, .	2.0	81
34	Rapid Detection of Monogenic Causes of Childhood-Onset Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 1109-1116.	4.5	74
35	Whole exome sequencing identifies causative mutations in the majority of consanguineous or familial cases with childhood-onset increased renal echogenicity. Kidney International, 2016, 89, 468-475.	5.2	74
36	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67

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37	WDR19 : An ancient, retrograde, intraflagellar ciliary protein is mutated in autosomal recessive retinitis pigmentosa and in Seniorâ€Loken syndrome. Clinical Genetics, 2013, 84, 150-159.	2.0	63
38	Mutation of the Mg2+ Transporter SLC41A1 Results in a Nephronophthisis-Like Phenotype. Journal of the American Society of Nephrology: JASN, 2013, 24, 967-977.	6.1	63
39	Mutations of the SLIT2â€“ROBO2 pathway genes SLIT2 and SRGAP1 confer risk for congenital anomalies of the kidney and urinary tract. Human Genetics, 2015, 134, 905-916.	3.8	62
40	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome: A systematic review. Autoimmunity Reviews, 2020, 19, 102526.	5.8	61
41	Misexpression screen delineates novel genes controlling Drosophila lifespan. Mechanisms of Ageing and Development, 2012, 133, 234-245.	4.6	53
42	Secreted metalloproteases ADAMTS9 and ADAMTS20 have a non-canonical role in ciliary vesicle growth during ciliogenesis. Nature Communications, 2019, 10, 953.	12.8	51
43	Unconventional secretion of transmembrane proteins. Seminars in Cell and Developmental Biology, 2018, 83, 59-66.	5.0	47
44	Specific autophagy and ESCRT components participate in the unconventional secretion of CFTR. Autophagy, 2018, 14, 1761-1778.	9.1	46
45	The HSP70 co-chaperone DNAJC14 targets misfolded pendrin for unconventional protein secretion. Nature Communications, 2016, 7, 11386.	12.8	43
46	Accuracy of Next-Generation Sequencing for Molecular Diagnosis in Patients With Infantile Nystagmus Syndrome. JAMA Ophthalmology, 2017, 135, 1376.	2.5	43
47	Rapid-Onset Obesity with Hypoventilation, Hypothalamic, Autonomic Dysregulation, and Neuroendocrine Tumors (ROHHADNET) Syndrome: A Systematic Review. BioMed Research International, 2018, 2018, 1-17.	1.9	42
48	Mutations in SLC26A1 Cause Nephrolithiasis. American Journal of Human Genetics, 2016, 98, 1228-1234.	6.2	41
49	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. Journal of Medical Genetics, 2016, 53, 208-214.	3.2	39
50	Advillin acts upstream of phospholipase C Ïµ1 in steroid-resistant nephrotic syndrome. Journal of Clinical Investigation, 2017, 127, 4257-4269.	8.2	39
51	ADCK4 Deficiency Destabilizes the Coenzyme Q Complex, Which Is Rescued by 2,4-Dihydroxybenzoic Acid Treatment. Journal of the American Society of Nephrology: JASN, 2020, 31, 1191-1211.	6.1	38
52	ZMYND10 stabilizes intermediate chain proteins in the cytoplasmic pre-assembly of dynein arms. PLoS Genetics, 2018, 14, e1007316.	3.5	37
53	ANO9/TMEM16J promotes tumourigenesis via EGFR and is a novel therapeutic target for pancreatic cancer. British Journal of Cancer, 2017, 117, 1798-1809.	6.4	35
54	Syntaxin 16 Binds to Cystic Fibrosis Transmembrane Conductance Regulator and Regulates Its Membrane Trafficking in Epithelial Cells. Journal of Biological Chemistry, 2010, 285, 35519-35527.	3.4	33

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55	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.	3.2	32
56	Loss of Epithelial Membrane Protein 2 Aggravates Podocyte Injury via Upregulation of Caveolin-1. Journal of the American Society of Nephrology: JASN, 2016, 27, 1066-1075.	6.1	32
57	The Cystic Fibrosis Transmembrane Conductance Regulator's Expanding SNARE Interactome. Traffic, 2011, 12, 364-371.	2.7	31
58	Synaptic Scaffolding Molecule Binds to and Regulates Vasoactive Intestinal Polypeptide Type-1 Receptor in Epithelial Cells. Gastroenterology, 2009, 137, 607-617.e4.	1.3	30
59	Analysis of 24 genes reveals a monogenic cause in 11.1% of cases with steroid-resistant nephrotic syndrome at a single center. Pediatric Nephrology, 2018, 33, 305-314.	1.7	30
60	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. American Journal of Human Genetics, 2017, 100, 323-333.	6.2	29
61	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. American Journal of Human Genetics, 2019, 104, 45-54.	6.2	29
62	Genetic Predisposition to Sporadic Congenital Hearing Loss in a Pediatric Population. Scientific Reports, 2017, 7, 45973.	3.3	28
63	Recent advances of animal model of focal segmental glomerulosclerosis. Clinical and Experimental Nephrology, 2018, 22, 752-763.	1.6	28
64	House dust mite extract activates apical Cl^{â”} channels through proteaseâ€activated receptor 2 in human airway epithelia. Journal of Cellular Biochemistry, 2010, 109, 1254-1263.	2.6	27
65	Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. Human Genetics, 2019, 138, 211-219.	3.8	26
66	In vivo outer hair cell gene editing ameliorates progressive hearing loss in dominant-negative Kcnq4 murine model. Theranostics, 2022, 12, 2465-2482.	10.0	26
67	Whole-exome sequencing identifies two novel mutations in KCNQ4 in individuals with nonsyndromic hearing loss. Scientific Reports, 2018, 8, 16659.	3.3	24
68	PLCE1 regulates the migration, proliferation, and differentiation of podocytes. Experimental and Molecular Medicine, 2020, 52, 594-603.	7.7	24
69	Mutations in KIRREL1, a slit diaphragm component, cause steroid-resistant nephrotic syndrome. Kidney International, 2019, 96, 883-889.	5.2	23
70	Functional characterization of ABCB4 mutations found in progressive familial intrahepatic cholestasis type 3. Scientific Reports, 2016, 6, 26872.	3.3	21
71	Genetic Inheritance of Late-Onset, Down-Sloping Hearing Loss and Its Implications for Auditory Rehabilitation. Ear and Hearing, 2020, 41, 114-124.	2.1	21
72	Adult-Onset Vitelliform Macular Dystrophy caused by BEST1 p.Ile38Ser Mutation is a Mild Form of Best Vitelliform Macular Dystrophy. Scientific Reports, 2017, 7, 9146.	3.3	20

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73	Rare KCNQ4 variants found in public databases underlie impaired channel activity that may contribute to hearing impairment. Experimental and Molecular Medicine, 2019, 51, 1-12.	7.7	16
74	Novel KCNQ4 variants in different functional domains confer genotype- and mechanism-based therapeutics in patients with nonsyndromic hearing loss. Experimental and Molecular Medicine, 2021, 53, 1192-1204.	7.7	16
75	The incidence rates and risk factors of Parkinson disease in patients with psoriasis: A nationwide population-based cohort study. Journal of the American Academy of Dermatology, 2020, 83, 1688-1695.	1.2	15
76	Activation of KCNQ4 as a Therapeutic Strategy to Treat Hearing Loss. International Journal of Molecular Sciences, 2021, 22, 2510.	4.1	15
77	Insulin-activated store-operated Ca2+ entry via Orai1 induces podocyte actin remodeling and causes proteinuria. Nature Communications, 2021, 12, 6537.	12.8	14
78	A synonymous variation in protease-activated receptor-2 isÂassociated with atopy in Korean children. Journal of Allergy and Clinical Immunology, 2011, 128, 1326-1334.e3.	2.9	13
79	Fecal Occult Blood Test Results of the National Colorectal Cancer Screening Program in South Korea (2006â€“2013). Scientific Reports, 2017, 7, 2804.	3.3	13
80	A recurrent mutation in KCNQ4 in Korean families with nonsyndromic hearing loss and rescue of the channel activity by KCNQ activators. Human Mutation, 2018, 40, 335-346.	2.5	13
81	Panel sequencing distinguishes monogenic forms of nephritis from nephrosis in children. Nephrology Dialysis Transplantation, 2019, 34, 474-485.	0.7	13
82	Contribution of SLC22A12 on hypouricemia and its clinical significance for screening purposes. Scientific Reports, 2019, 9, 14360.	3.3	13
83	Systematic evaluation of gene variants linked to hearing loss based on allele frequency threshold and filtering allele frequency. Scientific Reports, 2019, 9, 4583.	3.3	13
84	Grasp55â^'/â^' mice display impaired fat absorption and resistance to high-fat diet-induced obesity. Nature Communications, 2020, 11, 1418.	12.8	13
85	A novel HIF1AN substrate KANK3 plays a tumorâ€suppressive role in hepatocellular carcinoma. Cell Biology International, 2018, 42, 303-312.	3.0	12
86	Cystic kidneys in fetal Walkerâ€“Warburg syndrome with POMT2 mutation: Intrafamilial phenotypic variability in four siblings and review of literature. American Journal of Medical Genetics, Part A, 2017, 173, 2697-2702.	1.2	11
87	Genetics of vesicoureteral reflux and congenital anomalies of the kidney and urinary tract. Investigative and Clinical Urology, 2017, 58, S4.	2.0	11
88	Genomeâ€wide association study identifies TNFSF15 associated with childhood asthma. Allergy: European Journal of Allergy and Clinical Immunology, 2022, 77, 218-229.	5.7	11
89	A novel missense mutation in <scp>NR0B1</scp> causes delayedâ€onset primary adrenal insufficiency in adults. Clinical Genetics, 2017, 92, 344-346.	2.0	10
90	Differential genetic diagnoses of adult post-lingual hearing loss according to the audiogram pattern and novel candidate gene evaluation. Human Genetics, 2022, 141, 915-927.	3.8	9

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91	The L441P Mutation of Cystic Fibrosis Transmembrane conductance Regulator and its Molecular Pathogenic Mechanisms in a Korean Patient with Cystic Fibrosis. Journal of Korean Medical Science, 2010, 25, 166.	2.5	8
92	Whole-exome sequencing identified a missense mutation in WFS1 causing low-frequency hearing loss: a case report. BMC Medical Genetics, 2017, 18, 151.	2.1	8
93	Effects of Cold Agglutinin on the Accuracy of Complete Blood Count Results and Optimal Sample Pretreatment Protocols for Eliminating Such Effects. Annals of Laboratory Medicine, 2018, 38, 371-374.	2.5	8
94	A novel early truncation mutation in OTOG causes prelingual mild hearing loss without vestibular dysfunction. European Journal of Medical Genetics, 2019, 62, 81-84.	1.3	8
95	LCCL peptide cleavage after noise exposure exacerbates hearing loss and is associated with the monocyte infiltration in the cochlea. Hearing Research, 2021, 412, 108378.	2.0	8
96	OSBPL2 mutations impair autophagy and lead to hearing loss, potentially remedied by rapamycin. Autophagy, 2022, 18, 2593-2614.	9.1	8
97	COCH-related autosomal dominant nonsyndromic hearing loss: a phenotypeâ€“genotype study. Human Genetics, 2022, 141, 889-901.	3.8	7
98	The TECTA mutation R1890C is identified as one of the causes of genetic hearing loss: a case report. BMC Medical Genetics, 2019, 20, 57.	2.1	6
99	Microbiome analysis reveals that Ralstonia is responsible for decreased renal function in patients with ulcerative colitis. Clinical and Translational Medicine, 2021, 11, e322.	4.0	6
100	Shank2 mutant mice display a hypersecretory response to cholera toxin. Journal of Physiology, 2014, 592, 1809-1821.	2.9	5
101	Novel association between CDKAL1 and cholesterol efflux capacity: Replication after GWAS-based discovery. Atherosclerosis, 2018, 273, 21-27.	0.8	5
102	Clinical Heterogeneity Associated with MYO7A Variants Relies on Affected Domains. Biomedicines, 2022, 10, 798.	3.2	5
103	Expression of YAP and TAZ in molluscum contagiosum virus infected skin. British Journal of Dermatology, 2018, 179, 188-189.	1.5	4
104	Genomic Landscape and Mutational Spectrum of ADAMTS Family Genes in Mendelian Disorders Based on Gene Evidence Review for Variant Interpretation. Biomolecules, 2020, 10, 449.	4.0	4
105	Analysis of Conventional and Unconventional Trafficking of CFTR and Other Membrane Proteins. Methods in Molecular Biology, 2015, 1270, 137-154.	0.9	3
106	PDZ-based adaptor proteins in epithelial anion transport and VIP receptor regulation. Journal of Medical Investigation, 2009, 56, 302-305.	0.5	2
107	Dynamic Chronological Changes in Serum Triglycerides Are Associated With the Time Point for Non-alcoholic Fatty Liver Disease Development in the Nationwide Korean Population Cohort. Frontiers in Medicine, 2021, 8, 637241.	2.6	1
108	Heterogeneity of MYO15A variants significantly determine the feasibility of acoustic stimulation with hearing aid and cochlear implant. Hearing Research, 2021, 404, 108227.	2.0	1

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109	Autophagosome-mediated unconventional trafficking of CFTR. Pancreatology, 2013, 13, S18.	1.1	0
110	Mutations of IFT81, encoding an IFT-B core protein, as a rare cause of a ciliopathy. Cilia, 2015, 4, .	1.8	0
111	Combinatorial effect of ezetimibe and empagliflozin in non-alcoholic fatty liver disease in a mouse model and a liver organoid for disease modeling of hepatic steatosis. Journal of Hepatology, 2020, 73, S666-S667.	3.7	0
112	Biochemical and Functional Interaction between VPAC1 and Sâ€SCAM/MAGIâ€2. FASEB Journal, 2007, 21, A1322.	0.5	0
113	Uridine-5'-Triphosphate Stimulates Chloride Secretion via Cystic Fibrosis Transmembrane Conductance Regulator and Ca2+-Activated Chloride Channels in Cultured Human Middle Ear Epithelial Cells. Korean Journal of Otorhinolaryngology-Head and Neck Surgery, 2011, 54, 840.	0.2	0
114	RNA-Seq of Dysferlinopathy patients reveals differential gene for Limb-Girdle and Miyoshi subtypes. Proceedings for Annual Meeting of the Japanese Pharmacological Society, 2018, WCP2018, PO4-10-9.	0.0	0