Heon Yung Gee

List of Publications by Year in Descending Order

109 papers	5,638 citations	40 h-index	74 g-index
119 ext. papers	6,945 ext. citations	10 avg, IF	4.94 L-index

#	Paper	IF	Citations
109	outer hair cell gene editing ameliorates progressive hearing loss in dominant-negative murine model.. *Theranostics*, **2022**, 12, 2465-2482	12.1	3
108	mutations impair autophagy and lead to hearing loss, potentially remedied by rapamycin.. *Autophagy*, **2022**, 1-22	10.2	1
107	Insulin-activated store-operated Ca entry via Orai1 induces podocyte actin remodeling and causes proteinuria. *Nature Communications*, **2021**, 12, 6537	17.4	3
106	LCCL peptide cleavage after noise exposure exacerbates hearing loss and is associated with the monocyte infiltration in the cochlea. *Hearing Research*, **2021**, 412, 108378	3.9	1
105	Microbiome analysis reveals that Ralstonia is responsible for decreased renal function in patients with ulcerative colitis. *Clinical and Translational Medicine*, **2021**, 11, e322	5.7	0
104	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	4.9	0
103	Activation of KCNQ4 as a Therapeutic Strategy to Treat Hearing Loss. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	6
102	Heterogeneity of MYO15A variants significantly determine the feasibility of acoustic stimulation with hearing aid and cochlear implant. *Hearing Research*, **2021**, 404, 108227	3.9	0
101	Genome-wide association study identifies TNFSF15 associated with childhood asthma. *Allergy: European Journal of Allergy and Clinical Immunology*, **2021**,	9.3	2
100	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	12.8	1
99	COCH-related autosomal dominant nonsyndromic hearing loss: a phenotype-genotype study. *Human Genetics*, **2021**, 1	6.3	2
98	Differential genetic diagnoses of adult post-lingual hearing loss according to the audiogram pattern and novel candidate gene evaluation. *Human Genetics*, **2021**, 1	6.3	2
97	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	12.7	23
96	Grasp55 mice display impaired fat absorption and resistance to high-fat diet-induced obesity. *Nature Communications*, **2020**, 11, 1418	17.4	6
95	SGLT2 inhibition modulates NLRP3 inflammasome activity via ketones and insulin in diabetes with cardiovascular disease. *Nature Communications*, **2020**, 11, 2127	17.4	96
94	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome: A systematic review. *Autoimmunity Reviews*, **2020**, 19, 102526	13.6	31
93	PLCE1 regulates the migration, proliferation, and differentiation of podocytes. *Experimental and Molecular Medicine*, **2020**, 52, 594-603	12.8	7

92	Genetic Inheritance of Late-Onset, Down-Sloping Hearing Loss and Its Implications for Auditory Rehabilitation. *Ear and Hearing*, **2020**, 41, 114-124	3.4	12
91	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	4.5	4
90	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	2
89	Systematic evaluation of gene variants linked to hearing loss based on allele frequency threshold and filtering allele frequency. *Scientific Reports*, **2019**, 9, 4583	4.9	9
88	A novel early truncation mutation in OTOG causes prelingual mild hearing loss without vestibular dysfunction. *European Journal of Medical Genetics*, **2019**, 62, 81-84	2.6	7
87	Panel sequencing distinguishes monogenic forms of nephritis from nephrosis in children. *Nephrology Dialysis Transplantation*, **2019**, 34, 474-485	4.3	8
86	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	12.8	8
85	Mutations in KIRREL1, a slit diaphragm component, cause steroid-resistant nephrotic syndrome. *Kidney International*, **2019**, 96, 883-889	9.9	16
84	Contribution of SLC22A12 on hypouricemia and its clinical significance for screening purposes. *Scientific Reports*, **2019**, 9, 14360	4.9	5
83	Secreted metalloproteases ADAMTS9 and ADAMTS20 have a non-canonical role in ciliary vesicle growth during ciliogenesis. *Nature Communications*, **2019**, 10, 953	17.4	31
82	Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. *Human Genetics*, **2019**, 138, 211-219	6.3	15
81	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. *American Journal of Human Genetics*, **2019**, 104, 45-54	11	15
80	A recurrent mutation in KCNQ4 in Korean families with nonsyndromic hearing loss and rescue of the channel activity by KCNQ activators. *Human Mutation*, **2019**, 40, 335-346	4.7	11
79	Expression of YAP and TAZ in molluscum contagiosum virus infected skin. *British Journal of Dermatology*, **2018**, 179, 188-189	4	3
78	Novel association between CDKAL1 and cholesterol efflux capacity: Replication after GWAS-based discovery. *Atherosclerosis*, **2018**, 273, 21-27	3.1	2
77	Unconventional secretion of transmembrane proteins. *Seminars in Cell and Developmental Biology*, **2018**, 83, 59-66	7.5	31
76	Recent advances of animal model of focal segmental glomerulosclerosis. *Clinical and Experimental Nephrology*, **2018**, 22, 752-763	2.5	16
75	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis and nephrocalcinosis. *Kidney International*, **2018**, 93, 204-213	9.9	77

74	A novel HIF1AN substrate KANK3 plays a tumor-suppressive role in hepatocellular carcinoma. *Cell Biology International*, **2018**, 42, 303-312	4.5	5
73	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	3.2	21
72	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	3.1	6
71	ZMYND10 stabilizes intermediate chain proteins in the cytoplasmic pre-assembly of dynein arms. *PLoS Genetics*, **2018**, 14, e1007316	6	23
70	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		
69	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. *Clinical Journal of the American Society of Nephrology: CJASN*, **2018**, 13, 53-62	6.9	103
68	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. *Journal of Clinical Investigation*, **2018**, 128, 4313-4328	15.9	49
67	Whole-exome sequencing identifies two novel mutations in KCNQ4 in individuals with nonsyndromic hearing loss. *Scientific Reports*, **2018**, 8, 16659	4.9	16
66	Rapid-Onset Obesity with Hypoventilation, Hypothalamic, Autonomic Dysregulation, and Neuroendocrine Tumors (ROHHADNET) Syndrome: A Systematic Review. *BioMed Research International*, **2018**, 2018, 1250721	3	21
65	A Multi-layered Quantitative In Vivo Expression Atlas of the Podocyte Unravels Kidney Disease Candidate Genes. *Cell Reports*, **2018**, 23, 2495-2508	10.6	48
64	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. *Nature Communications*, **2018**, 9, 1960	17.4	68
63	Unconventional protein secretion - new insights into the pathogenesis and therapeutic targets of human diseases. *Journal of Cell Science*, **2018**, 131,	5.3	41
62	Specific autophagy and ESCRT components participate in the unconventional secretion of CFTR. *Autophagy*, **2018**, 14, 1761-1778	10.2	25
61	A novel missense mutation in NR0B1 causes delayed-onset primary adrenal insufficiency in adults. *Clinical Genetics*, **2017**, 92, 344-346	4	8
60	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. *American Journal of Human Genetics*, **2017**, 100, 323-333	11	19
59	Mutations in DZIP1L, which encodes a ciliary-transition-zone protein, cause autosomal recessive polycystic kidney disease. *Nature Genetics*, **2017**, 49, 1025-1034	36.3	99
58	Genetic Predisposition to Sporadic Congenital Hearing Loss in a Pediatric Population. *Scientific Reports*, **2017**, 7, 45973	4.9	25
57	ANO9/TMEM16J promotes tumourigenesis via EGFR and is a novel therapeutic target for pancreatic cancer. *British Journal of Cancer*, **2017**, 117, 1798-1809	8.7	24

56	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	3
55	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. *Journal of Clinical Investigation*, **2017**, 127, 912-928	15.9	112
54	Genetics of vesicoureteral reflux and congenital anomalies of the kidney and urinary tract. *Investigative and Clinical Urology*, **2017**, 58, S4-S13	1.9	7
53	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	4.9	12
52	Cystic kidneys in fetal Walker-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	2.5	8
51	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. *Nature Genetics*, **2017**, 49, 1529-1538	36.3	105
50	Accuracy of Next-Generation Sequencing for Molecular Diagnosis in Patients With Infantile Nystagmus Syndrome. *JAMA Ophthalmology*, **2017**, 135, 1376-1385	3.9	27
49	Fecal Occult Blood Test Results of the National Colorectal Cancer Screening Program in South Korea (2006-2013). *Scientific Reports*, **2017**, 7, 2804	4.9	8
48	Advillin acts upstream of phospholipase C ?1 in steroid-resistant nephrotic syndrome. *Journal of Clinical Investigation*, **2017**, 127, 4257-4269	15.9	25
47	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	9.9	60
46	FAT1 mutations cause a glomerulotubular nephropathy. *Nature Communications*, **2016**, 7, 10822	17.4	69
45	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. *Clinical Journal of the American Society of Nephrology: CJASN*, **2016**, 11, 664-72	6.9	74
44	Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. *Nature Genetics*, **2016**, 48, 457-65	36.3	109
43	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. *Journal of Medical Genetics*, **2016**, 53, 208-14	5.8	33
42	Loss of Epithelial Membrane Protein 2 Aggravates Podocyte Injury via Upregulation of Caveolin-1. *Journal of the American Society of Nephrology: JASN*, **2016**, 27, 1066-75	12.7	26
41	The HSP70 co-chaperone DNAJC14 targets misfolded pendrin for unconventional protein secretion. *Nature Communications*, **2016**, 7, 11386	17.4	32
40	Functional characterization of ABCB4 mutations found in progressive familial intrahepatic cholestasis type 3. *Scientific Reports*, **2016**, 6, 26872	4.9	13
39	Mutations in SLC26A1 Cause Nephrolithiasis. *American Journal of Human Genetics*, **2016**, 98, 1228-1234	11	28

38	DCDC2 mutations cause a renal-hepatic ciliopathy by disrupting Wnt signaling. *American Journal of Human Genetics*, **2015**, 96, 81-92	11	66
37	Mutations of the SLIT2-ROBO2 pathway genes SLIT2 and SRGAP1 confer risk for congenital anomalies of the kidney and urinary tract. *Human Genetics*, **2015**, 134, 905-16	6.3	48
36	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. *Journal of Medical Genetics*, **2015**, 52, 657-65	5.8	27
35	A single-gene cause in 29.5% of cases of steroid-resistant nephrotic syndrome. *Journal of the American Society of Nephrology: JASN*, **2015**, 26, 1279-89	12.7	378
34	Mutations of IFT81, encoding an IFT-B core protein, as a rare cause of a ciliopathy. *Cilia*, **2015**, 4,	5.5	78
33	Defects of CRB2 cause steroid-resistant nephrotic syndrome. *American Journal of Human Genetics*, **2015**, 96, 153-61	11	76
32	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. *Journal of Clinical Investigation*, **2015**, 125, 2375-84	15.9	133
31	Analysis of conventional and unconventional trafficking of CFTR and other membrane proteins. *Methods in Molecular Biology*, **2015**, 1270, 137-54	1.4	2
30	Whole-exome resequencing reveals recessive mutations in TRAP1 in individuals with CAKUT and VACTERL association. *Kidney International*, **2014**, 85, 1310-7	9.9	94
29	Mutations of CEP83 cause infantile nephronophthisis and intellectual disability. *American Journal of Human Genetics*, **2014**, 94, 905-14	11	59
28	Rapid detection of monogenic causes of childhood-onset steroid-resistant nephrotic syndrome. *Clinical Journal of the American Society of Nephrology: CJASN*, **2014**, 9, 1109-16	6.9	64
27	Mutations in RSPH1 cause primary ciliary dyskinesia with a unique clinical and ciliary phenotype. *American Journal of Respiratory and Critical Care Medicine*, **2014**, 189, 707-17	10.2	139
26	Shank2 mutant mice display a hypersecretory response to cholera toxin. *Journal of Physiology*, **2014** , 592, 1809-21	3.9	4
25	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. *Kidney International*, **2014**, 85, 880-7	9.9	55
24	Mutations in EMP2 cause childhood-onset nephrotic syndrome. *American Journal of Human Genetics* , **2014**, 94, 884-90	11	86
23	ZMYND10 is mutated in primary ciliary dyskinesia and interacts with LRRC6. *American Journal of Human Genetics*, **2013**, 93, 336-45	11	144
22	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. *American Journal of Human Genetics*, **2013**, 93, 672-86	11	140
21	Mutations in SPAG1 cause primary ciliary dyskinesia associated with defective outer and inner dynein arms. *American Journal of Human Genetics*, **2013**, 93, 711-20	11	109

20	WDR19: an ancient, retrograde, intraflagellar ciliary protein is mutated in autosomal recessive retinitis pigmentosa and in Senior-Loken syndrome. *Clinical Genetics*, **2013**, 84, 150-9	4	47
19	Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. *American Journal of Human Genetics*, **2013**, 93, 915-25	11	155
18	Mutation of the Mg2+ transporter SLC41A1 results in a nephronophthisis-like phenotype. *Journal of the American Society of Nephrology: JASN*, **2013**, 24, 967-77	12.7	49
17	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. *Journal of Clinical Investigation*, **2013**, 123, 5179-89	15.9	231
16	ARHGDIA mutations cause nephrotic syndrome via defective RHO GTPase signaling. *Journal of Clinical Investigation*, **2013**, 123, 3243-53	15.9	168
15	Misexpression screen delineates novel genes controlling Drosophila lifespan. *Mechanisms of Ageing and Development*, **2012**, 133, 234-45	5.6	40
14	Cholesterol modulates cell signaling and protein networking by specifically interacting with PDZ domain-containing scaffold proteins. *Nature Communications*, **2012**, 3, 1249	17.4	99
13	Exome capture reveals ZNF423 and CEP164 mutations, linking renal ciliopathies to DNA damage response signaling. *Cell*, **2012**, 150, 533-48	56.2	266
12	Autistic-like social behaviour in Shank2-mutant mice improved by restoring NMDA receptor function. *Nature*, **2012**, 486, 261-5	50.4	467
11	FAN1 mutations cause karyomegalic interstitial nephritis, linking chronic kidney failure to defective DNA damage repair. *Nature Genetics*, **2012**, 44, 910-5	36.3	167
10	A synonymous variation in protease-activated receptor-2 is associated with atopy in Korean children. *Journal of Allergy and Clinical Immunology*, **2011**, 128, 1326-1334.e3	11.5	10
9	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, 20267-76	16.4	81
8	Rescue of F508-CFTR trafficking via a GRASP-dependent unconventional secretion pathway. *Cell*, **2011**, 146, 746-60	56.2	228
7	The cystic fibrosis transmembrane conductance regulator expanding SNARE interactome. *Traffic*, **2011**, 12, 364-71	5.7	25
6	Uridine-5Triphosphate 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	
5	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, 166-71	4.7	7
4	Syntaxin 16 binds to cystic fibrosis transmembrane conductance regulator and regulates its membrane trafficking in epithelial cells. *Journal of Biological Chemistry*, **2010**, 285, 35519-27	5.4	28
3	House dust mite extract activates apical Cl(-) channels through protease-activated receptor 2 in human airway epithelia. *Journal of Cellular Biochemistry*, **2010**, 109, 1254-63	4.7	23

2	PDZ-based adaptor proteins in epithelial anion transport and VIP receptor regulation. *Journal of Medical Investigation*, **2009**, 56 Suppl, 302-5	1.2	2
1	Synaptic scaffolding molecule binds to and regulates vasoactive intestinal polypeptide type-1 receptor in epithelial cells. *Gastroenterology*, **2009**, 137, 607-17, 617.e1-4	13.3	26