

Heon Yung Gee

List of Publications by Citations

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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	Autistic-like social behaviour in Shank2-mutant mice improved by restoring NMDA receptor function. <i>Nature</i> , 2012 , 486, 261-5	50.4	467
108	A single-gene cause in 29.5% of cases of steroid-resistant nephrotic syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2015 , 26, 1279-89	12.7	378
107	Exome capture reveals ZNF423 and CEP164 mutations, linking renal ciliopathies to DNA damage response signaling. <i>Cell</i> , 2012 , 150, 533-48	56.2	266
106	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. <i>Journal of Clinical Investigation</i> , 2013 , 123, 5179-89	15.9	231
105	Rescue of B508-CFTR trafficking via a GRASP-dependent unconventional secretion pathway. <i>Cell</i> , 2011 , 146, 746-60	56.2	228
104	ARHGDI1 mutations cause nephrotic syndrome via defective RHO GTPase signaling. <i>Journal of Clinical Investigation</i> , 2013 , 123, 3243-53	15.9	168
103	FAN1 mutations cause karyomegalic interstitial nephritis, linking chronic kidney failure to defective DNA damage repair. <i>Nature Genetics</i> , 2012 , 44, 910-5	36.3	167
102	Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. <i>American Journal of Human Genetics</i> , 2013 , 93, 915-25	11	155
101	ZMYND10 is mutated in primary ciliary dyskinesia and interacts with LRRC6. <i>American Journal of Human Genetics</i> , 2013 , 93, 336-45	11	144
100	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013 , 93, 672-86	11	140
99	Mutations in RSPH1 cause primary ciliary dyskinesia with a unique clinical and ciliary phenotype. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2014 , 189, 707-17	10.2	139
98	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2015 , 125, 2375-84	15.9	133
97	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. <i>Journal of Clinical Investigation</i> , 2017 , 127, 912-928	15.9	112
96	Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. <i>Nature Genetics</i> , 2016 , 48, 457-65	36.3	109
95	Mutations in SPAG1 cause primary ciliary dyskinesia associated with defective outer and inner dynein arms. <i>American Journal of Human Genetics</i> , 2013 , 93, 711-20	11	109
94	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. <i>Nature Genetics</i> , 2017 , 49, 1529-1538	36.3	105
93	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2018 , 13, 53-62	6.9	103

92	Mutations in DZIP1L, which encodes a ciliary-transition-zone protein, cause autosomal recessive polycystic kidney disease. <i>Nature Genetics</i> , 2017 , 49, 1025-1034	36.3	99
91	Cholesterol modulates cell signaling and protein networking by specifically interacting with PDZ domain-containing scaffold proteins. <i>Nature Communications</i> , 2012 , 3, 1249	17.4	99
90	SGLT2 inhibition modulates NLRP3 inflammasome activity via ketones and insulin in diabetes with cardiovascular disease. <i>Nature Communications</i> , 2020 , 11, 2127	17.4	96
89	Whole-exome resequencing reveals recessive mutations in TRAP1 in individuals with CAKUT and VACTERL association. <i>Kidney International</i> , 2014 , 85, 1310-7	9.9	94
88	Mutations in EMP2 cause childhood-onset nephrotic syndrome. <i>American Journal of Human Genetics</i> , 2014 , 94, 884-90	11	86
87	A small molecule that binds to an ATPase domain of Hsc70 promotes membrane trafficking of mutant cystic fibrosis transmembrane conductance regulator. <i>Journal of the American Chemical Society</i> , 2011 , 133, 20267-76	16.4	81
86	Mutations of IFT81, encoding an IFT-B core protein, as a rare cause of a ciliopathy. <i>Cilia</i> , 2015 , 4,	5.5	78
85	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis and nephrocalcinosis. <i>Kidney International</i> , 2018 , 93, 204-213	9.9	77
84	Defects of CRB2 cause steroid-resistant nephrotic syndrome. <i>American Journal of Human Genetics</i> , 2015 , 96, 153-61	11	76
83	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2016 , 11, 664-72	6.9	74
82	FAT1 mutations cause a glomerulotubular nephropathy. <i>Nature Communications</i> , 2016 , 7, 10822	17.4	69
81	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. <i>Nature Communications</i> , 2018 , 9, 1960	17.4	68
80	DCDC2 mutations cause a renal-hepatic ciliopathy by disrupting Wnt signaling. <i>American Journal of Human Genetics</i> , 2015 , 96, 81-92	11	66
79	Rapid detection of monogenic causes of childhood-onset steroid-resistant nephrotic syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2014 , 9, 1109-16	6.9	64
78	Whole exome sequencing identifies causative mutations in the majority of consanguineous or familial cases with childhood-onset increased renal echogenicity. <i>Kidney International</i> , 2016 , 89, 468-475	9.9	60
77	Mutations of CEP83 cause infantile nephronophthisis and intellectual disability. <i>American Journal of Human Genetics</i> , 2014 , 94, 905-14	11	59
76	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. <i>Kidney International</i> , 2014 , 85, 880-7	9.9	55
75	Mutation of the Mg ²⁺ transporter SLC41A1 results in a nephronophthisis-like phenotype. <i>Journal of the American Society of Nephrology: JASN</i> , 2013 , 24, 967-77	12.7	49

74	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2018 , 128, 4313-4328	15.9	49
73	Mutations of the SLIT2-ROBO2 pathway genes SLIT2 and SRGAP1 confer risk for congenital anomalies of the kidney and urinary tract. <i>Human Genetics</i> , 2015 , 134, 905-16	6.3	48
72	A Multi-layered Quantitative In Vivo Expression Atlas of the Podocyte Unravels Kidney Disease Candidate Genes. <i>Cell Reports</i> , 2018 , 23, 2495-2508	10.6	48
71	WDR19: an ancient, retrograde, intraflagellar ciliary protein is mutated in autosomal recessive retinitis pigmentosa and in Senior-Loken syndrome. <i>Clinical Genetics</i> , 2013 , 84, 150-9	4	47
70	Unconventional protein secretion - new insights into the pathogenesis and therapeutic targets of human diseases. <i>Journal of Cell Science</i> , 2018 , 131,	5.3	41
69	Misexpression screen delineates novel genes controlling Drosophila lifespan. <i>Mechanisms of Ageing and Development</i> , 2012 , 133, 234-45	5.6	40
68	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. <i>Journal of Medical Genetics</i> , 2016 , 53, 208-14	5.8	33
67	The HSP70 co-chaperone DNAJC14 targets misfolded pendrin for unconventional protein secretion. <i>Nature Communications</i> , 2016 , 7, 11386	17.4	32
66	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome: A systematic review. <i>Autoimmunity Reviews</i> , 2020 , 19, 102526	13.6	31
65	Unconventional secretion of transmembrane proteins. <i>Seminars in Cell and Developmental Biology</i> , 2018 , 83, 59-66	7.5	31
64	Secreted metalloproteases ADAMTS9 and ADAMTS20 have a non-canonical role in ciliary vesicle growth during ciliogenesis. <i>Nature Communications</i> , 2019 , 10, 953	17.4	31
63	Syntaxin 16 binds to cystic fibrosis transmembrane conductance regulator and regulates its membrane trafficking in epithelial cells. <i>Journal of Biological Chemistry</i> , 2010 , 285, 35519-27	5.4	28
62	Mutations in SLC26A1 Cause Nephrolithiasis. <i>American Journal of Human Genetics</i> , 2016 , 98, 1228-1234	11	28
61	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. <i>Journal of Medical Genetics</i> , 2015 , 52, 657-65	5.8	27
60	Accuracy of Next-Generation Sequencing for Molecular Diagnosis in Patients With Infantile Nystagmus Syndrome. <i>JAMA Ophthalmology</i> , 2017 , 135, 1376-1385	3.9	27
59	Loss of Epithelial Membrane Protein 2 Aggravates Podocyte Injury via Upregulation of Caveolin-1. <i>Journal of the American Society of Nephrology: JASN</i> , 2016 , 27, 1066-75	12.7	26
58	Synaptic scaffolding molecule binds to and regulates vasoactive intestinal polypeptide type-1 receptor in epithelial cells. <i>Gastroenterology</i> , 2009 , 137, 607-17, 617.e1-4	13.3	26
57	Genetic Predisposition to Sporadic Congenital Hearing Loss in a Pediatric Population. <i>Scientific Reports</i> , 2017 , 7, 45973	4.9	25

56	The cystic fibrosis transmembrane conductance regulator α expanding SNARE interactome. <i>Traffic</i> , 2011 , 12, 364-71	5.7	25
55	Advillin acts upstream of phospholipase C β 1 in steroid-resistant nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2017 , 127, 4257-4269	15.9	25
54	Specific autophagy and ESCRT components participate in the unconventional secretion of CFTR. <i>Autophagy</i> , 2018 , 14, 1761-1778	10.2	25
53	ANO9/TMEM16J promotes tumorigenesis via EGFR and is a novel therapeutic target for pancreatic cancer. <i>British Journal of Cancer</i> , 2017 , 117, 1798-1809	8.7	24
52	ADCK4 Deficiency Destabilizes the Coenzyme Q Complex, Which Is Rescued by 2,4-Dihydroxybenzoic Acid Treatment. <i>Journal of the American Society of Nephrology: JASN</i> , 2020 , 31, 1191-1211	12.7	23
51	ZMYND10 stabilizes intermediate chain proteins in the cytoplasmic pre-assembly of dynein arms. <i>PLoS Genetics</i> , 2018 , 14, e1007316	6	23
50	House dust mite extract activates apical Cl ⁻ channels through protease-activated receptor 2 in human airway epithelia. <i>Journal of Cellular Biochemistry</i> , 2010 , 109, 1254-63	4.7	23
49	Analysis of 24 genes reveals a monogenic cause in 11.1% of cases with steroid-resistant nephrotic syndrome at a single center. <i>Pediatric Nephrology</i> , 2018 , 33, 305-314	3.2	21
48	Rapid-Onset Obesity with Hypoventilation, Hypothalamic, Autonomic Dysregulation, and Neuroendocrine Tumors (ROHHADNET) Syndrome: A Systematic Review. <i>BioMed Research International</i> , 2018 , 2018, 1250721	3	21
47	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. <i>American Journal of Human Genetics</i> , 2017 , 100, 323-333	11	19
46	Recent advances of animal model of focal segmental glomerulosclerosis. <i>Clinical and Experimental Nephrology</i> , 2018 , 22, 752-763	2.5	16
45	Mutations in KIRREL1, a slit diaphragm component, cause steroid-resistant nephrotic syndrome. <i>Kidney International</i> , 2019 , 96, 883-889	9.9	16
44	Whole-exome sequencing identifies two novel mutations in KCNQ4 in individuals with nonsyndromic hearing loss. <i>Scientific Reports</i> , 2018 , 8, 16659	4.9	16
43	Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. <i>Human Genetics</i> , 2019 , 138, 211-219	6.3	15
42	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. <i>American Journal of Human Genetics</i> , 2019 , 104, 45-54	11	15
41	Functional characterization of ABCB4 mutations found in progressive familial intrahepatic cholestasis type 3. <i>Scientific Reports</i> , 2016 , 6, 26872	4.9	13
40	Adult-Onset Vitelliform Macular Dystrophy caused by BEST1 p.Ile38Ser Mutation is a Mild Form of Best Vitelliform Macular Dystrophy. <i>Scientific Reports</i> , 2017 , 7, 9146	4.9	12
39	Genetic Inheritance of Late-Onset, Down-Sloping Hearing Loss and Its Implications for Auditory Rehabilitation. <i>Ear and Hearing</i> , 2020 , 41, 114-124	3.4	12

38	A recurrent mutation in KCNQ4 in Korean families with nonsyndromic hearing loss and rescue of the channel activity by KCNQ activators. <i>Human Mutation</i> , 2019 , 40, 335-346	4.7	11
37	A synonymous variation in protease-activated receptor-2 is associated with atopy in Korean children. <i>Journal of Allergy and Clinical Immunology</i> , 2011 , 128, 1326-1334.e3	11.5	10
36	Systematic evaluation of gene variants linked to hearing loss based on allele frequency threshold and filtering allele frequency. <i>Scientific Reports</i> , 2019 , 9, 4583	4.9	9
35	A novel missense mutation in NROB1 causes delayed-onset primary adrenal insufficiency in adults. <i>Clinical Genetics</i> , 2017 , 92, 344-346	4	8
34	Panel sequencing distinguishes monogenic forms of nephritis from nephrosis in children. <i>Nephrology Dialysis Transplantation</i> , 2019 , 34, 474-485	4.3	8
33	Rare KCNQ4 variants found in public databases underlie impaired channel activity that may contribute to hearing impairment. <i>Experimental and Molecular Medicine</i> , 2019 , 51, 1-12	12.8	8
32	Cystic kidneys in fetal Walker-Warburg syndrome with POMT2 mutation: Intrafamilial phenotypic variability in four siblings and review of literature. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 2697-2702	2.5	8
31	Fecal Occult Blood Test Results of the National Colorectal Cancer Screening Program in South Korea (2006-2013). <i>Scientific Reports</i> , 2017 , 7, 2804	4.9	8
30	PLCE1 regulates the migration, proliferation, and differentiation of podocytes. <i>Experimental and Molecular Medicine</i> , 2020 , 52, 594-603	12.8	7
29	Genetics of vesicoureteral reflux and congenital anomalies of the kidney and urinary tract. <i>Investigative and Clinical Urology</i> , 2017 , 58, S4-S13	1.9	7
28	A novel early truncation mutation in OTOG causes prelingual mild hearing loss without vestibular dysfunction. <i>European Journal of Medical Genetics</i> , 2019 , 62, 81-84	2.6	7
27	The L441P mutation of cystic fibrosis transmembrane conductance regulator and its molecular pathogenic mechanisms in a Korean patient with cystic fibrosis. <i>Journal of Korean Medical Science</i> , 2010 , 25, 166-71	4.7	7
26	Grasp55 mice display impaired fat absorption and resistance to high-fat diet-induced obesity. <i>Nature Communications</i> , 2020 , 11, 1418	17.4	6
25	Effects of Cold Agglutinin on the Accuracy of Complete Blood Count Results and Optimal Sample Pretreatment Protocols for Eliminating Such Effects. <i>Annals of Laboratory Medicine</i> , 2018 , 38, 371-374	3.1	6
24	Activation of KCNQ4 as a Therapeutic Strategy to Treat Hearing Loss. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	6
23	A novel HIF1AN substrate KANK3 plays a tumor-suppressive role in hepatocellular carcinoma. <i>Cell Biology International</i> , 2018 , 42, 303-312	4.5	5
22	Contribution of SLC22A12 on hypouricemia and its clinical significance for screening purposes. <i>Scientific Reports</i> , 2019 , 9, 14360	4.9	5
21	Shank2 mutant mice display a hypersecretory response to cholera toxin. <i>Journal of Physiology</i> , 2014 , 592, 1809-21	3.9	4

20	The incidence rates and risk factors of Parkinson disease in patients with psoriasis: A nationwide population-based cohort study. <i>Journal of the American Academy of Dermatology</i> , 2020 , 83, 1688-1695	4.5	4
19	Whole-exome sequencing identified a missense mutation in WFS1 causing low-frequency hearing loss: a case report. <i>BMC Medical Genetics</i> , 2017 , 18, 151	2.1	3
18	Expression of YAP and TAZ in molluscum contagiosum virus infected skin. <i>British Journal of Dermatology</i> , 2018 , 179, 188-189	4	3
17	Insulin-activated store-operated Ca entry via Orai1 induces podocyte actin remodeling and causes proteinuria. <i>Nature Communications</i> , 2021 , 12, 6537	17.4	3
16	outer hair cell gene editing ameliorates progressive hearing loss in dominant-negative murine model. <i>Theranostics</i> , 2022 , 12, 2465-2482	12.1	3
15	TheTECTA mutation R1890C is identified as one of the causes of genetic hearing loss: a case report. <i>BMC Medical Genetics</i> , 2019 , 20, 57	2.1	2
14	Novel association between CDKAL1 and cholesterol efflux capacity: Replication after GWAS-based discovery. <i>Atherosclerosis</i> , 2018 , 273, 21-27	3.1	2
13	PDZ-based adaptor proteins in epithelial anion transport and VIP receptor regulation. <i>Journal of Medical Investigation</i> , 2009 , 56 Suppl, 302-5	1.2	2
12	Analysis of conventional and unconventional trafficking of CFTR and other membrane proteins. <i>Methods in Molecular Biology</i> , 2015 , 1270, 137-54	1.4	2
11	Genome-wide association study identifies TNFSF15 associated with childhood asthma. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 2021 ,	9.3	2
10	COCH-related autosomal dominant nonsyndromic hearing loss: a phenotype-genotype study. <i>Human Genetics</i> , 2021 , 1	6.3	2
9	Differential genetic diagnoses of adult post-lingual hearing loss according to the audiogram pattern and novel candidate gene evaluation. <i>Human Genetics</i> , 2021 , 1	6.3	2
8	LCCL peptide cleavage after noise exposure exacerbates hearing loss and is associated with the monocyte infiltration in the cochlea. <i>Hearing Research</i> , 2021 , 412, 108378	3.9	1
7	Novel KCNQ4 variants in different functional domains confer genotype- and mechanism-based therapeutics in patients with nonsyndromic hearing loss. <i>Experimental and Molecular Medicine</i> , 2021 , 53, 1192-1204	12.8	1
6	mutations impair autophagy and lead to hearing loss, potentially remedied by rapamycin. <i>Autophagy</i> , 2022 , 1-22	10.2	1
5	Microbiome analysis reveals that Ralstonia is responsible for decreased renal function in patients with ulcerative colitis. <i>Clinical and Translational Medicine</i> , 2021 , 11, e322	5.7	0
4	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. <i>Frontiers in Medicine</i> , 2021 , 8, 637241	4.9	0
3	Heterogeneity of MYO15A variants significantly determine the feasibility of acoustic stimulation with hearing aid and cochlear implant. <i>Hearing Research</i> , 2021 , 404, 108227	3.9	0

- 2 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
- 1 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