Jin-Sung Lee

List of Publications by Year
in descending order

43 papers	506 citations	758635 12 h-index	713013 21 g-index
43 all docs	43 docs citations	43 times ranked	728 citing authors

#	Article	IF	Citations
1	Clinical application of a phenotypeâ€based <scp>NGS</scp> panel for differential diagnosis of inherited kidney disease and beyond. Clinical Genetics, 2021, 99, 236-249.	1.0	12
2	Potential role of stress-induced gluconeogenesis in disease aggravation and mortality in pyruvate dehydrogenase deficiency: A case-based hypothesis. Medical Hypotheses, 2021, 146, 110432.	0.8	0
3	Identification of a novel point mutation in DAX-1 gene in a patient with adrenal hypoplasia congenita. Annals of Pediatric Endocrinology and Metabolism, 2021, 26, 126-129.	0.8	2
4	Generation of induced pluripotent stem cells (KSCBi009-A) from a patient with Praderâ€“Willi syndrome (PWS) featuring deletion of the paternal chromosome region 15q11.2â€“q13. Stem Cell Research, 2020, 46, 101847.	0.3	0
5	Oral Pyridostigmineâ€responsive Visceral Myopathy With ACTG2 Mutations. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, e16-e17.	0.9	9
6	The First Case Series of Cryopyrin-Associated Periodic Syndrome in Korea. Allergy, Asthma and Immunology Research, 2019, 11, 583.	1.1	6
7	Genitopatellar Syndrome Secondary to De Novo KAT6B Mutation: The First Genetically Confirmed Case in South Korea. Yonsei Medical Journal, 2019, 60, 395.	0.9	2
8	Generation of patient-specific induced pluripotent stem cells (KSCBi007-A) derived from a patient with Praderâ€“Willi syndrome retain maternal uniparental disomy (UPD). Stem Cell Research, 2019, 41, 101647.	0.3	1
9	Systematic Approach for Drug Repositioning of Anti-Epileptic Drugs. Diagnostics, 2019, 9, 208.	1.3	6
10	Implementation of a Targeted Next-Generation Sequencing Panel for Constitutional Newborn Screening in High-Risk Neonates. Yonsei Medical Journal, 2019, 60, 1061.	0.9	4
11	A novel compound heterozygous mutation of the AIRE gene in a patient with autoimmune polyendocrine syndrome type 1. Annals of Pediatric Endocrinology and Metabolism, 2019, 24, 248-252.	0.8	3
12	Fructose-1,6-bisphosphatase deficiency presented with complex febrile convulsion. Neuroendocrinology Letters, 2019, 39, 533-536.	0.2	2
13	A Novel Heterozygous ANO3 Mutation with Basal Ganglia Dysfunction in a Patient with		

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19	Identification of disease comorbidity through hidden molecular mechanisms. Scientific Reports, 2016, 6, 39433.	1.6	42
20	Variations in plasma and urinary lipids in response to enzyme replacement therapy for Fabry disease patients by nanoflow UPLC-ESI-MS/MS. Analytical and Bioanalytical Chemistry, 2016, 408, 2265-2274.	1.9	16
21	Unravelling the mechanism of action of enzyme replacement therapy in Fabry disease. Journal of Human Genetics, 2016, 61, 143-149.	1.1	6
22	Complete form of pachydermoperiostosis with <scp>SLCO2A1</scp> gene mutation in a Korean family. Journal of Dermatology, 2015, 42, 655-657.	0.6	8
23	A Phase 2 Multi-center, Open-label, Switch-over Trial to Evaluate the Safety and Efficacy of AbcertinÂ® in Patients with Type 1 Gaucher Disease. Journal of Korean Medical Science, 2015, 30, 378.	1.1	9
24	Birt-Hogg-DubÃ© syndrome incidentally diagnosed during asthma management. Allergy Asthma & Respiratory Disease, 2015, 3, 232.	0.3	1
25	Lipidomic profiling of plasma and urine from patients with Gaucher disease during enzyme replacement therapy by nanoflow liquid chromatographyâ€“tandem mass spectrometry. Journal of Chromatography A, 2015, 1381, 132-139.	1.8	35
26	Germline mutation of Glu70Lys is highly frequent in Korean patients with von Hippelâ€“Lindau (VHL) disease. Journal of Human Genetics, 2014, 59, 488-493.	1.1	13
27	Novel mutations of KIT gene in two Korean patients: Variegated shades of phenotypes in tyrosine kinase 1 domain. Journal of Dermatological Science, 2014, 76, 74-76.	1.0	3
28	The First Case of Familial Mediterranean Fever Associated with Renal Amyloidosis in Korea. Yonsei Medical Journal, 2012, 53, 454.	0.9	12
29	A Korean Patient with Kniest Syndrome associated with Lipomeningomyelocele. Journal of Genetic Medicine, 2012, 9, 93-97.	0.1	0
30	Behavioral improvement after transplantation of neural precursors derived from embryonic stem cells into the globally ischemic brain of adolescent rats. Brain and Development, 2010, 32, 658-668.	0.6	7
31	MedRefSNP: A database of medically investigated SNPs. Human Mutation, 2009, 30, E460-E466.	1.1	11
32	PADB : Published Association Database. BMC Bioinformatics, 2007, 8, 348.	1.2	4
33	Recombinant adeno-associated virus mediated gene transfer in a mouse model for homocystinuria. Experimental and Molecular Medicine, 2006, 38, 652-661.	3.2	10
34	Treatment and management of patients with inherited metabolic diseases. Korean Journal of Pediatrics, 2006, 49, 1152.	1.9	2
35	Pyloric atresia-junctional epidermolysis bullosa syndrome showing novel 594insC/Q425P mutations in integrin beta4 gene (ITGB4). Experimental Dermatology, 2004, 13, 61-64.	1.4	13
36	In Vivo Differentiation of Mouse Embryonic Stem Cells into Hepatocytes. Cell Transplantation, 2002, 11, 359-368.	1.2	96

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37	Expression of genes involved in mammalian meiosis during the transition from egg to embryo. Molecular Reproduction and Development, 2001, 59, 144-158.	1.0	22
38	Characterization of a novel DNA polymorphism in the human CYP21 gene and application for DNA diagnosis of congenital adrenal hyperplasia. Clinical Endocrinology, 2000, 53, 419-422.	1.2	2
39	Prevalence of congenital malformations and genetic diseases in Korea. Journal of Human Genetics, 1999, 44, 30-34.	1.1	18
40	Effect of Topical Na-Hyaluronan on Hemidesmosome Formation in n-Heptanol-Induced Corneal Injury. Ophthalmic Research, 1998, 30, 96-100.	1.0	20
41	DNA-based prenatal diagnosis of a Korean family with tyrosinase-related oculocutaneous albinism (OCA1). Japanese Journal of Human Genetics, 1997, 42, 499-505.	0.8	9
42	Genetic investigation of the porphobilinogen deaminase gene in Swedish acute intermittent porphyria families. Human Genetics, 1997, 100, 63-66.	1.8	21
43	An unusual case of monozygotic epigastric heteropagus twinning. Journal of Pediatric Surgery, 1996, 31, 1457-1460.	0.8	26