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	In Vivo Differentiation of Mouse Embryonic Stem Cells into Hepatocytes. Cell Transplantation, 2002, 11, 359-368.	1.2	96
2	Identification of disease comorbidity through hidden molecular mechanisms. Scientific Reports, 2016, 6, 39433.	1.6	42
3	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
4	An unusual case of monozygotic epigastric heteropagus twinning. Journal of Pediatric Surgery, 1996, 31, 1457-1460.	0.8	26
5	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
6	Genetic investigation of the porphobilinogen deaminase gene in Swedish acute intermittent porphyria families. Human Genetics, 1997, 100, 63-66.	1.8	21
7	Effect of Topical Na-Hyaluronan on Hemidesmosome Formation in n-Heptanol-Induced Corneal Injury. Ophthalmic Research, 1998, 30, 96-100.	1.0	20
8	Prevalence of congenital malformations and genetic diseases in Korea. Journal of Human Genetics, 1999, 44, 30-34.	1.1	18
9	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
10	Prevalence of Rare Genetic Variations and Their Implications in NGS-data Interpretation. Scientific Reports, 2017, 7, 9810.	1.6	16
11	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
12	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
13	The First Case of Familial Mediterranean Fever Associated with Renal Amyloidosis in Korea. Yonsei Medical Journal, 2012, 53, 454.	0.9	12
14	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
15	MedRefSNP: A database of medically investigated SNPs. Human Mutation, 2009, 30, E460-E466.	1.1	11
16	Rare presentation of Rothmund-Thomson syndrome with predominantly cutaneous findings. JAAD Case Reports, 2017, 3, 172-174.	0.4	11
17	Recombinant adeno-associated virus mediated gene transfer in a mouse model for homocystinuria. Experimental and Molecular Medicine, 2006, 38, 652-661.	3.2	10
18	A Novel Heterozygous ANO3 Mutation with Basal Ganglia Dysfunction in a Patient with		

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19	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
20	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
21	Oral Pyridostigmineâ€responsive Visceral Myopathy With ACTG2 Mutations. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, e16-e17.	0.9	9
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	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
24	Alpers-Huttenlocher Syndrome First Presented with Hepatic Failure: Can Liver Transplantation Be Considered as Treatment Option?. Pediatric Gastroenterology, Hepatology and Nutrition, 2017, 20, 259.	0.4	7
25	Unravelling the mechanism of action of enzyme replacement therapy in Fabry disease. Journal of Human Genetics, 2016, 61, 143-149.	1.1	6
26	Congenital Orbital Fibrosis: Molecular Genetic Analysis by Whole-Exome and Mitochondrial Genome Sequencing. Yonsei Medical Journal, 2017, 58, 1078.	0.9	6
27	The First Case Series of Cryopyrin-Associated Periodic Syndrome in Korea. Allergy, Asthma and Immunology Research, 2019, 11, 583.	1.1	6
28	Systematic Approach for Drug Repositioning of Anti-Epileptic Drugs. Diagnostics, 2019, 9, 208.	1.3	6
29	PADB : Published Association Database. BMC Bioinformatics, 2007, 8, 348.	1.2	4
30	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
31	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
32	Clinical application of next-generation sequencing for the diagnosis of segmental neurofibromatosis. Journal of Dermatological Science, 2017, 88, 370-372.	1.0	3
33	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
34	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
35	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
36	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

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37	Treatment and management of patients with inherited metabolic diseases. Korean Journal of Pediatrics, 2006, 49, 1152.	1.9	2
38	Fructose-1,6-bisphosphatase deficiency presented with complex febrile convulsion. Neuroendocrinology Letters, 2019, 39, 533-536.	0.2	2
39	Birt-Hogg-DubÃ© syndrome incidentally diagnosed during asthma management. Allergy Asthma & Respiratory Disease, 2015, 3, 232.	0.3	1
40	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
41	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
42	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
43	A Korean Patient with Kniest Syndrome associated with Lipomeningomyelocele. Journal of Genetic Medicine, 2012, 9, 93-97.	0.1	0