Beom-Hee Lee

List of Publications by Year
in descending order

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128	1,512	394421	477307
papers	citations	19	29
		h-index	g-index

133	133	133	2864
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Spectrum of Mutations in Noonan Syndrome and Their Correlation with Phenotypes. Journal of Pediatrics, 2011, 159, 1029-1035.	1.8	78
2	Diagnostic yield and clinical utility of whole exome sequencing using an automated variant prioritization system, <scp>EVIDENCE</scp>. Clinical Genetics, 2020, 98, 562-570.	2.0	76
3	Mutations of the GLA gene in Korean patients with Fabry disease and frequency of the E66Q allele as a functional variant in Korean newborns. Journal of Human Genetics, 2010, 55, 512-517.	2.3	49
4	Diagnostic yield of targeted gene panel sequencing to identify the genetic etiology of disorders of sex development. Molecular and Cellular Endocrinology, 2017, 444, 19-25.	3.2	48
5	Genetic basis of Bartter syndrome in Korea. Nephrology Dialysis Transplantation, 2012, 27, 1516-1521.	0.7	44
6	Diverse genetic aetiologies and clinical outcomes of paediatric hypoparathyroidism. Clinical Endocrinology, 2015, 83, 790-796.	2.4	35
7	Cardiac Manifestations and Associations with Gene Mutations in Patients Diagnosed with RASopathies. Pediatric Cardiology, 2016, 37, 1539-1547.	1.3	34
8	Phenotype categorization of neurofibromatosis type I and correlation to NF1 mutation types. Journal of Human Genetics, 2020, 65, 79-89.	2.3	34
9	Clinical outcomes and the mutation spectrum of the OTC gene in patients with ornithine transcarbamylase deficiency. Journal of Human Genetics, 2015, 60, 501-507.	2.3	33
10	Endocrine dysfunctions in children with Williams-Beuren syndrome. Annals of Pediatric Endocrinology and Metabolism, 2016, 21, 15.	2.3	31
11	Long-term enzyme replacement therapy for Fabry disease: efficacy and unmet needs in cardiac and renal outcomes. Journal of Human Genetics, 2016, 61, 923-929.	2.3	30
12	Pharmacologic properties of high-dose ambroxol in four patients with Gaucher disease and myoclonic epilepsy. Journal of Medical Genetics, 2020, 57, 124-131.	3.2	29
13	Argininemia Presenting With Progressive Spastic Diplegia. Pediatric Neurology, 2011, 44, 218-220.	2.1	28
14	Enhanced thrombospondin-1 causes dysfunction of vascular endothelial cells derived from Fabry disease-induced pluripotent stem cells. EBioMedicine, 2020, 52, 102633.	6.1	28
15	Upgrading the evidence for the use of ambroxol in Gaucher disease and <scp>GBA</scp> related Parkinson: Investigator initiated registry based on real life data. American Journal of Hematology, 2021, 96, 545-551.	4.1	24
16	Psychological Impact of Quarantine on Caregivers at a Children's Hospital for Contact with Case of COVID-19. Journal of Korean Medical Science, 2020, 35, e255.	2.5	23
17	Clinical and genetic characteristics of patients with fatty acid oxidation disorders identified by newborn screening. BMC Pediatrics, 2018, 18, 103.	1.7	22
18	Moyamoya Syndrome in a Patient With Noonan-like Syndrome With Loose Anagen Hair. Pediatric Neurology, 2015, 52, 352-355.	2.1	21

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19	Biochemical and molecular characteristics of citrin deficiency in Korean children. Journal of Human Genetics, 2017, 62, 305-307.	2.3	21
20	Severe form of neuroblastoma amplified sequence deficiency in an infant with recurrent acute liver failure. Pediatrics International, 2018, 60, 302-304.	0.5	21
21	Phenotypes of atopic dermatitis identified by cluster analysis in early childhood. Journal of Dermatology, 2019, 46, 117-123.	1.2	21
22	Diagnostic Value of Ceruloplasmin in the Diagnosis of Pediatric Wilson's Disease. Pediatric Gastroenterology, Hepatology and Nutrition, 2015, 18, 187.	1.2	20
23	Fabry disease: characterisation of the plasma proteome pre- and post-enzyme replacement therapy. Journal of Medical Genetics, 2017, 54, 771-780.	3.2	20
24	Progressive mesenteric lymphadenopathy with protein-losing enteropathy; a devastating complication in Gaucher disease. Molecular Genetics and Metabolism, 2012, 105, 522-524.	1.1	19
25	Clinical, Biochemical, and Genetic Characterization of Glycogen Storage Type IX in a Child with Asymptomatic Hepatomegaly. Pediatric Gastroenterology, Hepatology and Nutrition, 2015, 18, 138.	1.2	19
26	Lowe syndrome: a single center's experience in Korea. Korean Journal of Pediatrics, 2014, 57, 140.	1.9	18
27	Quantitative analysis of methylation status at 11p15 and 7q21 for the genetic diagnosis of Beckwithâ€“Wiedemann syndrome and Silverâ€“Russell syndrome. Journal of Human Genetics, 2013, 58, 604-610.	2.3	17
28	Allele frequency of a 24â€‰bp duplication in exon 10 of the CHIT1 gene in the general Korean population and in Korean patients with Gaucher disease. Journal of Human Genetics, 2014, 59, 276-279.	2.3	17
29	MPV17 mutations in patients with hepatocerebral mitochondrial DNA depletion syndrome. Molecular Genetics and Metabolism Reports, 2016, 8, 74-76.	1.1	17
30	Long-term efficacy of recombinant human growth hormone therapy in short-statured patients with Noonan syndrome. Annals of Pediatric Endocrinology and Metabolism, 2016, 21, 26.	2.3	17
31	Nonalcoholic Fatty Liver Disease in 2 Siblings With Adultâ€onset Type II Citrullinemia. Journal of Pediatric Gastroenterology and Nutrition, 2010, 50, 682-685.	1.8	16
32	Mutation Spectrum of STAR and the Founder Effect of p.Q258* in Korean Patients with Congenital Lipoid Adrenal Hyperplasia. Molecular Medicine, 2017, 23, 149-154.	4.4	16
33	The Rho-associated kinase inhibitor fasudil can replace Y-27632 for use in human pluripotent stem cell research. PLoS ONE, 2020, 15, e0233057.	2.5	16
34	High prevalence of neonatal presentation in Korean patients with citrullinemia type 1, and their shared mutations. Molecular Genetics and Metabolism, 2013, 108, 18-24.	1.1	15
35	Chromosome 1p36.22p36.21 duplications/triplication causes Setleis syndrome (focal facial dermal) Tj ETQq1 1 0.784314 rgBT /Overl	1.2	15
36	Characteristic dysmorphic features in congenital disorders of glycosylation type IIb. Journal of Human Genetics, 2018, 63, 383-386.	2.3	15

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37	Clinical and molecular spectra of BRAF-associated RASopathy. Journal of Human Genetics, 2021, 66, 389-399.	2.3	15
38	DEND Syndrome with Heterozygous KCNJ11 Mutation Successfully Treated with Sulfonylurea. Journal of Korean Medical Science, 2017, 32, 1042.	2.5	14
39	Genotype and Phenotype Analysis in Pediatric Patients with Cystinuria. Journal of Korean Medical Science, 2017, 32, 310.	2.5	14
40	Diagnostic performance of automated, streamlined, daily updated exome analysis in patients with neurodevelopmental delay. Molecular Medicine, 2022, 28, 38.	4.4	14
41	The early molecular processes underlying the neurological manifestations of an animal model of Wilson's disease. Metallomics, 2013, 5, 532.	2.4	13
42	Variable phenotypes of multiple synostosis syndrome in patients with novel NOG mutations. Joint Bone Spine, 2014, 81, 533-536.	1.6	12
43	Impaired osteogenesis in Menkes disease-derived induced pluripotent stem cells. Stem Cell Research and Therapy, 2015, 6, 160.	5.5	12
44	Setleis syndrome due to inheritance of the 1p36.22p36.21 duplication: evidence for lack of penetrance. Journal of Human Genetics, 2015, 60, 717-722.	2.3	12
45	Setleis syndrome: clinical, molecular and structural studies of the first <scp>TWIST2</scp> missense mutation. Clinical Genetics, 2015, 88, 489-493.	2.0	12
46	Clinical and molecular characterisation of Holtâ€“Oram syndrome focusing on cardiac manifestations. Cardiology in the Young, 2015, 25, 1093-1098.	0.8	11
47	Clinical features, outcomes, and genetic analysis in <scp>K</scp>orean children with <scp>A</scp>lagille syndrome. Pediatrics International, 2015, 57, 552-557.	0.5	11
48	Malfunction in Mitochondrial Î²-Oxidation Contributes to Lipid Accumulation in Hepatocyte-Like Cells Derived from Citrin Deficiency-Induced Pluripotent Stem Cells. Stem Cells and Development, 2016, 25, 636-647.	2.1	11
49	Genotype-Phenotype Correlation ofSMN1andNAIPDeletions in Korean Patients with Spinal Muscular		

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55	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.	2.5	9
56	Further delineation of COG8-CDG: A case with novel compound heterozygous mutations diagnosed by targeted exome sequencing. Clinica Chimica Acta, 2017, 471, 191-195.	1.1	9
57	The GBA p.G85E mutation in Korean patients with non-neuronopathic Gaucher disease: founder and neuroprotective effects. Orphanet Journal of Rare Diseases, 2020, 15, 318.	2.7	9
58	SHP2 mutations induce precocious gliogenesis of Noonan syndrome-derived iPSCs during neural development in vitro. Stem Cell Research and Therapy, 2020, 11, 209.	5.5	9
59	De Novo Development of mtDNA Deletion Due to Decreased POLG and SSBP1 Expression in Humans. Genes, 2021, 12, 284.	2.4	9
60	Dysregulated ECM remodeling proteins lead to aberrant osteogenesis of Costello syndrome iPSCs. Stem Cell Reports, 2021, 16, 1985-1998.	4.8	9
61	Identification of 1p36 deletion syndrome in patients with facial dysmorphism and developmental delay. Korean Journal of Pediatrics, 2016, 59, 16.	1.9	9
62	Atypical Manifestation of Carnitine Palmitoyltransferase 1A Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2015, 60, e19-22.	1.8	8
63	Short-term efficacy of N-carbamylglutamate in a patient with N-acetylglutamate synthase deficiency. Journal of Human Genetics, 2015, 60, 395-397.	2.3	8
64	Neurofibromatosis type I: points to be considered by general pediatricians. Clinical and Experimental Pediatrics, 2021, 64, 149-156.	2.2	8
65	Chylous Manifestations and Management of Gorham-Stout Syndrome. Korean Journal of Thoracic and Cardiovascular Surgery, 2019, 52, 44-46.	0.6	8
66	Turner syndrome presented with tall stature due to overdosage of theSHOXgene. Annals of Pediatric Endocrinology and Metabolism, 2015, 20, 110.	2.3	8
67	Histological, biochemical, and genetic characterization of early-onset fulminating sialidosis type 2 in a Korean neonate with hydrops fetalis. Brain and Development, 2014, 36, 171-175.	1.1	7
68	Modeling of Menkes disease via human induced pluripotent stem cells. Biochemical and Biophysical Research Communications, 2014, 444, 311-318.	2.1	7
69	Long-term Consequences of Congenital Adrenal Hyperplasia due to Classic 21-hydroxylase Deficiency in Adolescents and Adults. Experimental and Clinical Endocrinology and Diabetes, 2017, 125, 196-201.	1.2	7
70	A multicenter, open-label, phase III study of Abcertin in Gaucher disease. Medicine (United States), 2017, 96, e8492.	1.0	7
71	Rare Frequency of Mutations in Pituitary Transcription Factor Genes in Combined Pituitary Hormone or Isolated Growth Hormone Deficiencies in Korea. Yonsei Medical Journal, 2017, 58, 527.	2.2	7
72	Biochemical and molecular characterisation of neurological Wilson disease. Journal of Medical Genetics, 2018, 55, 587-593.	3.2	7

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73	Characterization of the Subventricular-Thalamo-Cortical Circuit in the NP-C Mouse Brain, and New Insights Regarding Treatment. Molecular Therapy, 2019, 27, 1507-1526.	8.2	7
74	Clinical characteristics and disease progression of retinitis pigmentosa associated with PDE6B mutations in Korean patients. Scientific Reports, 2020, 10, 19540.	3.3	7
75	Diagnosis of metachromatic leukodystrophy in a patient with regression and Phelan-McDermid syndrome. Brain and Development, 2020, 42, 414-417.	1.1	7
76	Noonan syndrome and RASopathies: Clinical features, diagnosis and management. Journal of Genetic Medicine, 2019, 16, 1-9.	0.2	7
77	Impaired Osteogenesis of Disease-Specific Induced Pluripotent Stem Cells Derived from a CFC Syndrome Patient. International Journal of Molecular Sciences, 2017, 18, 2591.	4.1	6
78	Low prevalence of argininosuccinate lyase deficiency among inherited urea cycle disorders in Korea. Journal of Human Genetics, 2018, 63, 911-917.	2.3	6
79	Clinical Application of Whole Exome Sequencing to Identify Rare but Remediable Neurologic Disorders. Journal of Clinical Medicine, 2020, 9, 3724.	2.4	6
80	Ultra-rare renal diseases diagnosed with whole-exome sequencing: Utility in diagnosis and management. BMC Medical Genomics, 2021, 14, 177.	1.5	6
81	Efficacy and Safety of Selumetinib in Pediatric Patients With Neurofibromatosis Type 1. Neurology, 2022, 98, .	1.1	6
82	Clinical and genetic features of four patients with Pearson syndrome. Medicine (United States), 2022, 101, e28793.	1.0	6
83	Lysinuric protein intolerance with homozygous SLC7A7 mutation caused by maternal uniparental isodisomy of chromosome 14. Journal of Human Genetics, 2019, 64, 1137-1140.	2.3	5
84	Phenotypic and molecular spectrum of Korean patients with Lesch-Nyhan syndrome and attenuated clinical variants. Metabolic Brain Disease, 2019, 34, 1335-1340.	2.9	5
85	Phenotypic and molecular spectra of patients with switch/sucrose nonfermenting complex-related intellectual disability disorders in Korea. BMC Medical Genomics, 2021, 14, 254.	1.5	5
86	Diagnosis of Schaaf-Yang syndrome in Korean children with developmental delay and hypotonia. Medicine (United States), 2020, 99, e23864.	1.0	5
87	Identification of a novel therapeutic target underlying atypical manifestation of Gaucher disease. Clinical and Translational Medicine, 2022, 12, e862.	4.0	5
88	Low prevalence of classical galactosemia in Korean population. Journal of Human Genetics, 2011, 56, 94-96.	2.3	4
89	Three Novel Pathogenic Mutations in K_{ATP} Channel Genes and Somatic Imprinting Alterations of the 11p15 Region in Pancreatic Tissue in Patients with Congenital Hyperinsulinism. Hormone Research in Paediatrics, 2015, 83, 204-210.	1.8	4
90	Life-threatening bleeding from gastric mucosal angiokeratomas during anticoagulation. Medicine (United States), 2017, 96, e6063.	1.0	4

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91	Association between ARID2 and RAS-MAPK pathway in intellectual disability and short stature. Journal of Medical Genetics, 2021, 58, 767-777.	3.2	4
92	Phelan-McDermid syndrome presenting with developmental delays and facial dysmorphisms. Korean Journal of Pediatrics, 2016, 59, S25.	1.9	4
93	A Case of Stickler Syndrome Type I Caused by a Novel Variant of COL2A1 Gene. Journal of Genetic Medicine, 2011, 8, 125-129.	0.2	4
94	Long-term clinical outcome and the identification of homozygousCYP27B1gene mutations in a patient with vitamin D hydroxylation-deficient rickets type 1A. Annals of Pediatric Endocrinology and Metabolism, 2016, 21, 169.	2.3	4
95	Turner syndrome with primary hyperparathyroidism. Annals of Pediatric Endocrinology and Metabolism, 2013, 18, 85.	2.3	4
96	Unfavorable clinical outcomes in patients with carbamoyl phosphate synthetase 1 deficiency. Clinica Chimica Acta, 2022, 526, 55-61.	1.1	4
97	Whole-body MRI evaluation in neurofibromatosis type 1 patients younger than 3 years old and the genetic contribution to disease progression. Orphanet Journal of Rare Diseases, 2022, 17, 24.	2.7	4
98	Focal facial dermal dysplasia type 4: identification of novel CYP26C1 mutations in unrelated patients. Journal of Human Genetics, 2018, 63, 257-261.	2.3	3
99	Novel ATP8B1 Gene Mutations in a Child with Progressive Familial Intrahepatic Cholestasis Type 1. Pediatric Gastroenterology, Hepatology and Nutrition, 2019, 22, 479.	1.2	3
100	Hematopoietic stem cell transplantation in an infant with dedicator of cytokinesis 8 (DOCK8) deficiency associated with systemic lupus erythematosus. Medicine (United States), 2021, 100, e20866.	1.0	3
101	Neurovascular Manifestation of Loeys-Dietz Syndrome: A Case Report. Journal of Genetic Medicine, 2013, 10, 47-51.	0.2	3
102	Case Report: Mevalonic Aciduria Complicated by Acute Myeloid Leukemia After Hematopoietic Stem Cell Transplantation. Frontiers in Immunology, 2021, 12, 782780.	4.8	3
103	Efficacy of Living Donor Liver Transplantation in Patients with Methylmalonic Acidemia. Pediatric Gastroenterology, Hepatology and Nutrition, 2021, 24, 288.	1.2	2
104	Two cases of TSC2/PKD1 contiguous gene deletion syndrome. Journal of Genetic Medicine, 2016, 13, 36-40.	0.2	2
105	Ehlers-Danlos syndrome VIII with novel C1R variant accompanying white matter changes. Journal of Genetic Medicine, 2019, 16, 43-47.	0.2	2
106	Surgical Interventions and Clinical Outcomes of Patients with Trisomy 18: a Single Center Experience. Perinatology, 2020, 31, 129.	0.1	2
107	Genotype-phenotype correlations and long-term efficacy of pamidronate therapy in patients with osteogenesis imperfecta. Annals of Pediatric Endocrinology and Metabolism, 2022, 27, 22-29.	2.3	2
108	Long-Term Follow-Up of Peripheral Pigmentary Retinopathy in Asian Patients with Danon Disease. Genes, 2020, 11, 1356.	2.4	1

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109	Fatal outcome of autosomal recessive polycystic kidney disease in neonates with recessive PKHD1 mutations. Medicine (United States), 2020, 99, e20113.	1.0	1
110	Rapidly Progressive Parkinsonism and Dementia with No Insomnia due to the PRNP D178N Mutation.		

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127	Beckwith-Wiedemann Syndrome and Jacobsen Syndrome Caused by 11pter Duplication and 11qter Deletion Inherited from Paternal Pericentric Inversion. Laboratory Medicine Online, 2020, 10, 255-261.	0.2	0
128	Psychological characteristics of adult neurofibromatosis type 1 patients seeking elective surgery. Archives of Aesthetic Plastic Surgery, 2020, 26, 150-156.	0.2	0