Beom-Hee Lee

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2255126/publications.pdf

Version: 2024-02-01

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	Unfavorable clinical outcomes in patients with carbamoyl phosphate synthetase 1 deficiency. Clinica Chimica Acta, 2022, 526, 55-61.	1.1	4
2	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
3	Efficacy and Safety of Selumetinib in Pediatric Patients With Neurofibromatosis Type 1. Neurology, 2022, 98, .	1.1	6
4	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
5	Clinical and genetic features of four patients with Pearson syndrome. Medicine (United States), 2022, 101, e28793.	1.0	6
6	Diagnostic performance of automated, streamlined, daily updated exome analysis in patients with neurodevelopmental delay. Molecular Medicine, 2022, 28, 38.	4.4	14
7	Identification of a novel therapeutic target underlying atypical manifestation of Gaucher disease. Clinical and Translational Medicine, 2022, 12, e862.	4.0	5
8	Phenotypic and Genetic Complexity in Pediatric Movement Disorders. Frontiers in Genetics, 2022, 13, .	2.3	1
9	Association between ARID2 and RAS-MAPK pathway in intellectual disability and short stature. Journal of Medical Genetics, 2021, 58, 767-777.	3.2	4
10	Clinical and molecular spectra of BRAF-associated RASopathy. Journal of Human Genetics, 2021, 66, 389-399.	2.3	15
11	Efficacy of Living Donor Liver Transplantation in Patients with Methylmalonic Acidemia. Pediatric Gastroenterology, Hepatology and Nutrition, 2021, 24, 288.	1.2	2
12	Rapidly Progressive Parkinsonism and Dementia with No Insomnia due to the PRNP D178N Mutation.		

#	Article	IF	Citations
19	Clinical Genetic Testing in Children with Kidney Disease. Childhood Kidney Diseases, 2021, 25, 14-21.	0.4	1
20	Ultra-rare renal diseases diagnosed with whole-exome sequencing: Utility in diagnosis and management. BMC Medical Genomics, 2021, 14, 177.	1.5	6
21	Dysregulated ECM remodeling proteins lead to aberrant osteogenesis of Costello syndrome iPSCs. Stem Cell Reports, 2021, 16, 1985-1998.	4.8	9
22	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
23	Identification of FOXG1 mutations in infantile hypotonia and postnatal microcephaly. Medicine (United) Tj ETQq1 1 0.784314 rgBT /Ove	1.0	1
24	Case Report: Mevalonic Aciduria Complicated by Acute Myeloid Leukemia After Hematopoietic Stem Cell Transplantation. Frontiers in Immunology, 2021, 12, 782780.	4.8	3
25	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
26	Phenotype categorization of neurofibromatosis type I and correlation to NF1 mutation types. Journal of Human Genetics, 2020, 65, 79-89.	2.3	34
27	Long-Term Follow-Up of Peripheral Pigmentary Retinopathy in Asian Patients with Danon Disease. Genes, 2020, 11, 1356.	2.4	1
28	Clinical Application of Whole Exome Sequencing to Identify Rare but Remediable Neurologic Disorders. Journal of Clinical Medicine, 2020, 9, 3724.	2.4	6
29	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
30	Clinical characteristics and disease progression of retinitis pigmentosa associated with PDE6B mutations in Korean patients. Scientific Reports, 2020, 10, 19540.	3.3	7
31	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
32	Fatal outcome of autosomal recessive polycystic kidney disease in neonates with recessive PKHD1 mutations. Medicine (United States), 2020, 99, e20113.	1.0	1
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	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
35	Diagnosis of metachromatic leukodystrophy in a patient with regression and Phelan-McDermid syndrome. Brain and Development, 2020, 42, 414-417.	1.1	7
36	Enhanced thrombospondin-1 causes dysfunction of vascular endothelial cells derived from Fabry disease-induced pluripotent stem cells. EBioMedicine, 2020, 52, 102633.	6.1	28

#	Article	IF	Citations
37	Human Embryonic Stem Cell-Derived Wilsonâ€™s Disease Model for Screening Drug Efficacy. Cells, 2020, 9, 872.	4.1	10
38	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
39	Hypotonia, Ataxia, and Delayed Development Syndrome caused by the EBF3 mutation in a Korean boy with muscle hypotonia. Journal of Genetic Medicine, 2020, 17, 92-96.	0.2	0
40	Diagnosis of Schaaf-Yang syndrome in Korean children with developmental delay and hypotonia. Medicine (United States), 2020, 99, e23864.	1.0	5
41	Surgical Interventions and Clinical Outcomes of Patients with Trisomy 18: a Single Center Experience. Perinatology, 2020, 31, 129.	0.1	2
42	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
43	Psychological characteristics of adult neurofibromatosis type 1 patients seeking elective surgery. Archives of Aesthetic Plastic Surgery, 2020, 26, 150-156.	0.2	0
44	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
45	Identification of extremely rare mitochondrial disorders by whole exome sequencing. Journal of Human Genetics, 2019, 64, 1117-1125.	2.3	10
46	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
47	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
48	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
49	Phenotypes of atopic dermatitis identified by cluster analysis in early childhood. Journal of Dermatology, 2019, 46, 117-123.	1.2	21
50	Chylous Manifestations and Management of Gorham-Stout Syndrome. Korean Journal of Thoracic and Cardiovascular Surgery, 2019, 52, 44-46.	0.6	8
51	Noonan syndrome and RASopathies: Clinical features, diagnosis and management. Journal of Genetic Medicine, 2019, 16, 1-9.	0.2	7
52	Ehlers-Danlos syndrome VIII with novel C1R variant accompanying white matter changes. Journal of Genetic Medicine, 2019, 16, 43-47.	0.2	2
53	An ANKRD11 exonic deletion accompanied by a congenital megacolon in an infant with KBG syndrome. Journal of Genetic Medicine, 2019, 16, 39-42.	0.2	1
54	Identification of Potockiâ€“Lupski syndrome in patients with developmental delay and growth failure. Journal of Genetic Medicine, 2019, 16, 49-54.	0.2	0

#	Article	IF	Citations
55	Biochemical and molecular analyses of infantile sialic acid storage disease in a patient with nonimmune hydrops fetalis. Clinica Chimica Acta, 2018, 482, 199-202.	1.1	0
56	Biochemical and molecular characterisation of neurological Wilson disease. Journal of Medical Genetics, 2018, 55, 587-593.	3.2	7
57	Characteristic dysmorphic features in congenital disorders of glycosylation type IIb. Journal of Human Genetics, 2018, 63, 383-386.	2.3	15
58	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
59	Clinical and genetic characteristics of patients with fatty acid oxidation disorders identified by newborn screening. BMC Pediatrics, 2018, 18, 103.	1.7	22
60	Severe form of neuroblastoma amplified sequence deficiency in an infant with recurrent acute liver failure. Pediatrics International, 2018, 60, 302-304.	0.5	21
61	Langer-Giedion Syndrome with 8q23.1â€“q24.13 Deletion by Complex Three-way Translocation. Laboratory Medicine Online, 2018, 8, 29.	0.2	0
62	Low prevalence of argininosuccinate lyase deficiency among inherited urea cycle disorders in Korea. Journal of Human Genetics, 2018, 63, 911-917.	2.3	6
63	A novel mutation in XLRS1 gene in X-linked juvenile retinoschisis. Journal of Genetic Medicine, 2018, 15, 13-16.	0.2	1
64	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
65	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
66	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
67	Fabry disease: characterisation of the plasma proteome pre- and post-enzyme replacement therapy. Journal of Medical Genetics, 2017, 54, 771-780.	3.2	20
68	Life-threatening bleeding from gastric mucosal angiokeratomas during anticoagulation. Medicine (United States), 2017, 96, e6063.	1.0	4
69	A multicenter, open-label, phase III study of Abcertin in Gaucher disease. Medicine (United States), 2017, 96, e8492.	1.0	7
70	The focal facial dermal dysplasias: phenotypic spectrum and molecular genetic heterogeneity. Journal of Medical Genetics, 2017, 54, 585-590.	3.2	10
71	Biochemical and molecular characteristics of citrin deficiency in Korean children. Journal of Human Genetics, 2017, 62, 305-307.	2.3	21
72	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

#	Article	IF	Citations
73	DEND Syndrome with Heterozygous KCNJ11 Mutation Successfully Treated with Sulfonylurea. Journal of Korean Medical Science, 2017, 32, 1042.	2.5	14
74	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
75	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
76	Genotype-Phenotype Correlation ofSMN1andNAIPDeletions in Korean Patients with Spinal Muscular		

#	Article	IF	Citations
91	Clinical and molecular characterisation of Holtâ€“Oram syndrome focusing on cardiac manifestations. Cardiology in the Young, 2015, 25, 1093-1098.	0.8	11
92	Diverse genetic aetiologies and clinical outcomes of paediatric hypoparathyroidism. Clinical Endocrinology, 2015, 83, 790-796.	2.4	35
93	Atypical Manifestation of Carnitine Palmitoyltransferase 1A Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2015, 60, e19-22.	1.8	8
94	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
95	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
96	Diagnostic Value of Ceruloplasmin in the Diagnosis of Pediatric Wilson's Disease. Pediatric Gastroenterology, Hepatology and Nutrition, 2015, 18, 187.	1.2	20
97	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
98	Impaired osteogenesis in Menkes disease-derived induced pluripotent stem cells. Stem Cell Research and Therapy, 2015, 6, 160.	5.5	12
99	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
100	Moyamoya Syndrome in a Patient With Noonan-like Syndrome With Loose Anagen Hair. Pediatric Neurology, 2015, 52, 352-355.	2.1	21
101	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
102	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
103	Chromosome 1p36.22p36.21 duplications/triplication causes Setleis syndrome (focal facial dermal) Tj ETQq1 1 0.784314 rgBT /Overl	1.2	15
104	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
105	Setleis syndrome: clinical, molecular and structural studies of the first <scp>TWIST2</scp> missense mutation. Clinical Genetics, 2015, 88, 489-493.	2.0	12
106	Turner syndrome presented with tall stature due to overdosage of theSHOXgene. Annals of Pediatric Endocrinology and Metabolism, 2015, 20, 110.	2.3	8
107	Variable phenotypes of multiple synostosis syndrome in patients with novel NOG mutations. Joint Bone Spine, 2014, 81, 533-536.	1.6	12
108	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

#	Article	IF	Citations
109	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
110	Modeling of Menkes disease via human induced pluripotent stem cells. Biochemical and Biophysical Research Communications, 2014, 444, 311-318.	2.1	7
111	Lowe syndrome: a single center's experience in Korea. Korean Journal of Pediatrics, 2014, 57, 140.	1.9	18
112	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
113	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
114	The early molecular processes underlying the neurological manifestations of an animal model of Wilson's disease. Metallomics, 2013, 5, 532.	2.4	13
115	Neurovascular Manifestation of Loeys-Dietz Syndrome: A Case Report. Journal of Genetic Medicine, 2013, 10, 47-51.	0.2	3
116	Turner syndrome with primary hyperparathyroidism. Annals of Pediatric Endocrinology and Metabolism, 2013, 18, 85.	2.3	4
117	An Incidentally Identified Sporadic Case with Adrenoleukodystrophy with the ABCD1 Mutation. Journal of Genetic Medicine, 2013, 10, 43-46.	0.2	1
118	Genetic basis of Bartter syndrome in Korea. Nephrology Dialysis Transplantation, 2012, 27, 1516-1521.	0.7	44
119	Progressive mesenteric lymphadenopathy with protein-losing enteropathy; a devastating complication in Gaucher disease. Molecular Genetics and Metabolism, 2012, 105, 522-524.	1.1	19
120	Manifestation of Giant Bilateral Symptomatic Adrenal Myelolipomas in an Adult Patient with Congenital Adrenal Hyperplasia. Annals of Pediatric Endocrinology and Metabolism, 2012, 17, 244.	2.3	1
121	A Case with Emanuel Syndrome Resulting from a Maternal Balanced Translocation. Journal of Genetic Medicine, 2012, 9, 35-37.	0.2	0
122	Argininemia Presenting With Progressive Spastic Diplegia. Pediatric Neurology, 2011, 44, 218-220.	2.1	28
123	Spectrum of Mutations in Noonan Syndrome and Their Correlation with Phenotypes. Journal of Pediatrics, 2011, 159, 1029-1035.	1.8	78
124	Low prevalence of classical galactosemia in Korean population. Journal of Human Genetics, 2011, 56, 94-96.	2.3	4
125	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
126	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

#	Article	IF	Citations
127	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
128	A Case with Transient Hyperammonemia of Newborn. Journal of Genetic Medicine, 2010, 7, 87-110.	0.2	0