Peter Kannu

List of Publications by Year in descending order

50 papers	1,436 citations	516710 16 h-index	345221 36 g-index
53 all docs	53 docs citations	53 times ranked	3458 citing authors

#	Article	IF	Citations
1	<scp>ELOVL4</scp> with erythrokeratoderma: A pediatric case and emerging genodermatosis. American Journal of Medical Genetics, Part A, 2021, 185, 1619-1623.	1.2	4
2	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
3	Evolving Management of Fibrodysplasia Ossificans Progressiva. Journal of Pediatrics, 2021, 232, S9-S15.	1.8	2
4	Improving the Diagnosis of Fibrodysplasia Ossificans Progressiva. Journal of Pediatrics, 2021, 232, S3-S8.	1.8	1
5	Hardened Hope: Care Advances for Fibrodysplasia Ossificans Progressiva. Journal of Pediatrics, 2021, 232, S1-S2.	1.8	0
6	Infigratinib in Children With Achondroplasia (ACH): Design of PROPEL2 - A Phase 2, Open-Label, Dose-Escalation and Dose-Expansion Study. Journal of the Endocrine Society, 2021, 5, A667-A668.	0.2	0
7	Diagnostic utility of next-generation sequence genetic panel testing in children presenting with a clinically significant fracture history. Archives of Osteoporosis, 2021, 16, 88.	2.4	3
8	Lost bones: differential diagnosis of acro-osteolysis seen by the pediatric rheumatologist. Pediatric Rheumatology, 2021, 19, 113.	2.1	8
9	Unique retinal signaling defect in GNB5-related disease. Documenta Ophthalmologica, 2020, 140, 273-277.	2.2	7
10	Aplasia cutis congenita associated with a heterozygous lossâ€ofâ€function UBA2 variant. British Journal of Dermatology, 2020, 182, 792-794.	1.5	4
11	Haploinsufficiency of RREB1 causes a Noonan-like RASopathy via epigenetic reprogramming of RAS-MAPK pathway genes. Nature Communications, 2020, 11, 4673.	12.8	19
12	SUN-093 Prospective Clinical Assessment Study in Children with Achondroplasia: The PROPEL Trial. Journal of the Endocrine Society, 2020, 4, .	0.2	0
13	A novel ENPP1 mutation identified in a multigenerational family affected by Cole disease. Pediatric Dermatology, 2020, 37, 868-871.	0.9	5
14	Epidermal growth factor receptor deficiency: Expanding the phenotype beyond infancy. Journal of Dermatology, 2020, 47, 898-902.	1.2	3
15	Thiemann disease and familial digital arthropathy â€“ brachydactyly: two sides of the same coin?. Orphanet Journal of Rare Diseases, 2019, 14, 156.	2.7	3
16	The epileptology of GNB5 encephalopathy. Epilepsia, 2019, 60, e121-e127.	5.1	13
17	Unraveling incontinentia pigmenti: A comparison of phenotype and genotype variants. Journal of the American Academy of Dermatology, 2019, 81, 1142-1149.	1.2	15
18	Atypical COL3A1 variants (glutamic acid to lysine) cause vascular Ehlersâ€“Danlos syndrome with a consistent phenotype of tissue fragility and skin hyperextensibility. Genetics in Medicine, 2019, 21, 2081-2091.	2.4	16

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19	Hypophosphatasia: Canadian update on diagnosis and management. Osteoporosis International, 2019, 30, 1713-1722.	3.1	28
20	Disruption of the PTHLH regulatory landscape results in features consistent with hyperparathyroid disease. American Journal of Medical Genetics, Part A, 2019, 179, 663-667.	1.2	2
21	Whole-exome sequencing identifies a homozygous pathogenic variant in TAT in a girl with palmoplantar keratoderma. Molecular Genetics and Metabolism Reports, 2019, 21, 100534.	1.1	1
22	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
23	Characteristic MR Imaging Findings of the Neonatal Brain in RASopathies. American Journal of Neuroradiology, 2018, 39, 1146-1152.	2.4	12
24	Enzyme replacement therapy in perinatal hypophosphatasia: Case report of a negative outcome and lessons for clinical practice. Molecular Genetics and Metabolism Reports, 2018, 14, 22-26.	1.1	14
25	Riboflavin transporter deficiency mimicking mitochondrial myopathy caused by complex II deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 399-403.	1.2	18
26	Genome-wide sequencing expands the phenotypic spectrum of EP300 variants. European Journal of Medical Genetics, 2018, 61, 125-129.	1.3	8
27	UNRAVELING INCONTINENTIA PIGMENTI: A COMPARISON OF PHENOTYPE AND GENOTYPE VARIANTS. Paediatrics and Child Health, 2018, 23, e32-e32.	0.6	0
28	Genotype-phenotype data from a case series of patients with mosaic neurofibromatosis type 1. British Journal of Dermatology, 2018, 179, 1216-1217.	1.5	6
29	IFT80 mutations cause a novel complex ciliopathy phenotype with retinal degeneration. Clinical Genetics, 2018, 94, 368-372.	2.0	5
30	Interferon-Stimulated Gene Expression as a Preferred Biomarker for Disease Activity in Aicardiâ€“GoutiÃ¨res Syndrome. Journal of Interferon and Cytokine Research, 2017, 37, 147-152.	1.2	8
31	Homozygous mutation in PRUNE1 in an Ojiâ€Cree male with a complex neurological phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 740-743.	1.2	20
32	Mosaic Neurofibromatosis Type 1 in Children: A Single-Institution Experience. Journal of Cutaneous Medicine and Surgery, 2017, 21, 379-382.	1.2	7
33	Parentsâ€™ Understanding of Genetics and Heritability. Journal of Genetic Counseling, 2017, 26, 541-547.	1.6	9
34	<i>PLS3</i> Mutations in X-Linked Osteoporosis: Clinical and Bone Characteristics of Two Novel Mutations. Hormone Research in Paediatrics, 2017, 88, 298-304.	1.8	27
35	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	21.4	164
36	Mosaicism for a SPRED1 deletion revealed in a patient with clinically suspected mosaic neurofibromatosis. British Journal of Dermatology, 2017, 176, 1077-1078.	1.5	8

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37	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
38	Î²â€Catenin modulation in neurofibromatosis type 1 bone repair: therapeutic implications. FASEB Journal, 2016, 30, 3227-3237.	0.5	12
39	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	3.3	35
40	Buschke-Ollendorff syndrome: a novel case series and systematic review. British Journal of Dermatology, 2016, 174, 723-729.	1.5	37
41	MG-109â€...Revisiting a clinical diagnosis 15 years later with the aid of whole exome sequencing: Osteopetrosis versus harderophorphyria. Journal of Medical Genetics, 2015, 52, A4.1-A4.	3.2	0
42	Recessive Osteogenesis Imperfecta Caused by Missense Mutations in SPARC. American Journal of Human Genetics, 2015, 96, 979-985.	6.2	107
43	Mutations Preventing Regulated Exon Skipping in MET Cause Osteofibrous Dysplasia. American Journal of Human Genetics, 2015, 97, 837-847.	6.2	22
44	Diagnostic yield of genetic testing in epileptic encephalopathy in childhood. Epilepsia, 2015, 56, 707-716.	5.1	223
45	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	6.2	14
46	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	2.8	38
47	Perthes' disease. BMJ, The, 2014, 349, g5584-g5584.	6.0	7
48	Post-axial polydactyly type A2, overgrowth and autistic traits associated with a chromosome 13q31.3 microduplication encompassing miR-17-92 and GPC5. European Journal of Medical Genetics, 2013, 56, 452-457.	1.3	19
49	Clinical phenotypes associated with type II collagen mutations. Journal of Paediatrics and Child Health, 2012, 48, E38-43.	0.8	75
50	Two Novel COL2A1 Mutations Associated with a Legg-CalvÃ©-Perthes Disease-like Presentation. Clinical Orthopaedics and Related Research, 2011, 469, 1785-1790.	1.5	33