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	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
2	Diagnostic yield of genetic testing in epileptic encephalopathy in childhood. Epilepsia, 2015, 56, 707-716.	5.1	223
3	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	21.4	164
4	Recessive Osteogenesis Imperfecta Caused by Missense Mutations in SPARC. American Journal of Human Genetics, 2015, 96, 979-985.	6.2	107
5	Clinical phenotypes associated with type II collagen mutations. Journal of Paediatrics and Child Health, 2012, 48, E38-43.	0.8	75
6	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
7	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	2.8	38
8	Buschke-Ollendorff syndrome: a novel case series and systematic review. British Journal of Dermatology, 2016, 174, 723-729.	1.5	37
9	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	3.3	35
10	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
11	Hypophosphatasia: Canadian update on diagnosis and management. Osteoporosis International, 2019, 30, 1713-1722.	3.1	28
12	<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
13	Mutations Preventing Regulated Exon Skipping in MET Cause Osteofibrous Dysplasia. American Journal of Human Genetics, 2015, 97, 837-847.	6.2	22
14	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
15	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
16	Haploinsufficiency of RREB1 causes a Noonan-like RASopathy via epigenetic reprogramming of RAS-MAPK pathway genes. Nature Communications, 2020, 11, 4673.	12.8	19
17	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
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	Unraveling incontinentia pigmenti: A comparison of phenotype and genotype variants. Journal of the American Academy of Dermatology, 2019, 81, 1142-1149.	1.2	15
20	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
21	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	6.2	14
22	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
23	The epileptology of GNB5 encephalopathy. Epilepsia, 2019, 60, e121-e127.	5.1	13
24	Î²â€Catenin modulation in neurofibromatosis type 1 bone repair: therapeutic implications. FASEB Journal, 2016, 30, 3227-3237.	0.5	12
25	Characteristic MR Imaging Findings of the Neonatal Brain in RASopathies. American Journal of Neuroradiology, 2018, 39, 1146-1152.	2.4	12
26	Parentsâ€™ Understanding of Genetics and Heritability. Journal of Genetic Counseling, 2017, 26, 541-547.	1.6	9
27	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
28	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
29	Genome-wide sequencing expands the phenotypic spectrum of EP300 variants. European Journal of Medical Genetics, 2018, 61, 125-129.	1.3	8
30	Lost bones: differential diagnosis of acro-osteolysis seen by the pediatric rheumatologist. Pediatric Rheumatology, 2021, 19, 113.	2.1	8
31	Perthes' disease. BMJ, The, 2014, 349, g5584-g5584.	6.0	7
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	Unique retinal signaling defect in GNB5-related disease. Documenta Ophthalmologica, 2020, 140, 273-277.	2.2	7
34	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
35	IFT80 mutations cause a novel complex ciliopathy phenotype with retinal degeneration. Clinical Genetics, 2018, 94, 368-372.	2.0	5
36	A novel ENPP1 mutation identified in a multigenerational family affected by Cole disease. Pediatric Dermatology, 2020, 37, 868-871.	0.9	5

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37	Aplasia cutis congenita associated with a heterozygous lossâ€ofâ€function UBA2 variant. British Journal of Dermatology, 2020, 182, 792-794.	1.5	4
38	<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
39	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
40	Epidermal growth factor receptor deficiency: Expanding the phenotype beyond infancy. Journal of Dermatology, 2020, 47, 898-902.	1.2	3
41	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
42	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
43	Evolving Management of Fibrodysplasia Ossificans Progressiva. Journal of Pediatrics, 2021, 232, S9-S15.	1.8	2
44	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
45	Improving the Diagnosis of Fibrodysplasia Ossificans Progressiva. Journal of Pediatrics, 2021, 232, S3-S8.	1.8	1
46	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
47	UNRAVELING INCONTINENTIA PIGMENTI: A COMPARISON OF PHENOTYPE AND GENOTYPE VARIANTS. Paediatrics and Child Health, 2018, 23, e32-e32.	0.6	0
48	SUN-093 Prospective Clinical Assessment Study in Children with Achondroplasia: The PROPEL Trial. Journal of the Endocrine Society, 2020, 4, .	0.2	0
49	Hardened Hope: Care Advances for Fibrodysplasia Ossificans Progressiva. Journal of Pediatrics, 2021, 232, S1-S2.	1.8	0
50	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