Katta Mohan Girisha

List of Publications by Year
in descending order

245 papers	5,238 citations	136885 32 h-index	128225 60 g-index
252 all docs	252 docs citations	252 times ranked	10379 citing authors

#	Article	IF	Citations
1	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. Blood, 2016, 127, 3154-3164.	0.6	465
2	Biallelic mutations in the death domain of PIDD1 impair caspase-2 activation and are associated with intellectual disability. Translational Psychiatry, 2021, 11, 1.	2.4	334
3	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. Nature Genetics, 2016, 48, 1071-1076.	9.4	314
4	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	4.5	234
5	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	1.5	141
6	RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. Nature, 2018, 557, 564-569.	13.7	141
7	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	2.3	134
8	The promise of discovering population-specific disease-associated genes in South Asia. Nature Genetics, 2017, 49, 1403-1407.	9.4	129
9	Cortical-Bone Fragility â€" Insights from sFRP4 Deficiency in Pyleâ€™s Disease. New England Journal of Medicine, 2016, 374, 2553-2562.	13.9	119
10	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	1.1	85
11	Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53.	0.7	75
12	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150.	2.6	74
13	Down syndrome:. Archives of Medical Research, 2004, 35, 31-35.	1.5	72
14	Microduplications encompassing the Sonic hedgehog limb enhancer <scp>ZRS</scp> are associated with Haasâ€type polysyndactyly and Laurinâ€Sandrow syndrome. Clinical Genetics, 2014, 86, 318-325.	1.0	72
15	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	2.6	71
16	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	0.7	69
17	A homozygous nonsense variant in <scp>IFT52</scp> is associated with a human skeletal ciliopathy. Clinical Genetics, 2016, 90, 536-539.	1.0	68
18	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	0.7	68

#	Article	IF	Citations
19	Biallelic Loss of Proprioception-Related PIEZO2 Causes Muscular Atrophy with Perinatal Respiratory Distress, Arthrogryposis, and Scoliosis. American Journal of Human Genetics, 2016, 99, 1206-1216.	2.6	65
20	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	1.4	64
21	Analysis of the WISP3 gene in Indian families with progressive pseudorheumatoid dysplasia. American Journal of Medical Genetics, Part A, 2012, 158A, 2820-2828.	0.7	63
22	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	2.6	62
23	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	0.5	57
24	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	0.7	55
25	The homozygous variant c.797G>A/p.(Cys266Tyr) inPISDis associated with a Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function. Human Mutation, 2019, 40, 299-309.	1.1	54
26	Homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. Journal of Human Genetics, 2017, 62, 723-727.	1.1	53
27	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	3.7	51
28	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	1.4	48
29	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	0.7	40
30	Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. Bone, 2018, 110, 368-377.	1.4	38
31	Mutations in patients with osteogenesis imperfecta from consanguineous Indian families. European Journal of Medical Genetics, 2015, 58, 21-27.	0.7	37
32	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. American Journal of Human Genetics, 2021, 108, 115-133.	2.6	37
33	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
34	Glycosaminoglycan levels in dried blood spots of patients with mucopolysaccharidoses and mucolipidoses. Molecular Genetics and Metabolism, 2017, 120, 247-254.	0.5	35
35	Clinical utility of fetal autopsy and its impact on genetic counseling. Prenatal Diagnosis, 2015, 35, 685-691.	1.1	34
36	Identification of a novel LRRK1 mutation in a family with osteosclerotic metaphyseal dysplasia. Journal of Human Genetics, 2017, 62, 437-441.	1.1	33

#	Article	IF	Citations
37	Mutations in SREBF1, Encoding Sterol Regulatory Element Binding Transcription Factor 1, Cause Autosomal-Dominant IFAP Syndrome. American Journal of Human Genetics, 2020, 107, 34-45.	2.6	33
38	GALNS mutations in Indian patients with mucopolysaccharidosis IVA. American Journal of Medical Genetics, Part A, 2014, 164, 2793-2801.	0.7	31
39	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. American Journal of Medical Genetics, Part A, 2016, 170, 410-417.	0.7	31
40	Organization for rare diseases India (ORDI)Ââ€“Âaddressing the challenges and opportunities for the Indian rare diseases' community. Genetical Research, 2014, 96, e009.	0.3	30
41	Expanding the phenotype associated with 17q12 duplication: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2013, 161, 352-359.	0.7	29
42	BGN Mutations in X-Linked Spondyloepimetaphyseal Dysplasia. American Journal of Human Genetics, 2016, 98, 1243-1248.	2.6	29
43	Autosomal recessive spinocerebellar ataxia 20: Report of a new patient and review of literature. European Journal of Medical Genetics, 2017, 60, 118-123.	0.7	29
44	Novel subtype of mucopolysaccharidosis caused by arylsulfatase K (ARSK) deficiency. Journal of Medical Genetics, 2022, 59, 957-964.	1.5	29
45	A novel sequence variant in SFRP4 causing Pyle disease. Journal of Human Genetics, 2017, 62, 575-576.	1.1	27
46	Gaucher disease: single gene molecular characterization of one-hundred Indian patients reveals novel variants and the most prevalent mutation. BMC Medical Genetics, 2019, 20, 31.	2.1	27
47	Homozygosity for a nonsense variant in AIMP2 is associated with a progressive neurodevelopmental disorder with microcephaly, seizures, and spastic quadriparesis. Journal of Human Genetics, 2018, 63, 19-25.	1.1	26
48	A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for earlyâ€onset monogenic disorders in Indians. Human Mutation, 2021, 42, e15-e61.	1.1	25
49	Mutation spectrum of COL1A1 and COL1A2 genes in Indian patients with osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2014, 164, 1482-1489.	0.7	24
50	Identification and characterization of 20 novel pathogenic variants in 60 unrelated Indian patients with mucopolysaccharidoses type I and type <scp>II</scp>. Clinical Genetics, 2016, 90, 496-508.	1.0	23
51	Human IFT52 mutations uncover a novel role for the protein in microtubule dynamics and centrosome cohesion. Human Molecular Genetics, 2019, 28, 2720-2737.	1.4	23
52	Costello syndrome with severe cutis laxa and mosaic HRAS G12S mutation. American Journal of Medical Genetics, Part A, 2010, 152A, 2861-2864.	0.7	22
53	Recurrent and novel GLB1 mutations in India. Gene, 2015, 567, 173-181.	1.0	22
54	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. European Journal of Human Genetics, 2018, 26, 695-708.	1.4	22

#	Article	IF	Citations
55	Novel and recurrent mutations in WISP3 and an atypical phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 2481-2484.	0.7	21
56	Novel KIAA0753 mutations extend the phenotype of skeletal ciliopathies. Scientific Reports, 2017, 7, 15585.	1.6	21
57	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. American Journal of Human Genetics, 2020, 107, 293-310.	2.6	21
58	T1 and M1 polymorphism in glutathione S-transferase gene and coronary artery disease in North Indian population. Indian Journal of Medical Sciences, 2004, 58, 520-6.	0.1	20
59	A neurodegenerative mitochondrial disease phenotype due to biallelic lossâ€ofâ€function variants in PNPLA8 encoding calciumâ€independent phospholipase A2Î³. American Journal of Medical Genetics, Part A, 2018, 176, 1232-1237.	0.7	19
60	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	1.1	19
61	Eight years experience from a skeletal dysplasia referral center in a tertiary hospital in Southern India: A model for the diagnosis and treatment of rare diseases in a developing country. American Journal of Medical Genetics, Part A, 2014, 164, 2317-2323.	0.7	18
62	Phenotype and genotype in patients with Larsen syndrome: clinical homogeneity and allelic heterogeneity in seven patients. BMC Medical Genetics, 2016, 17, 27.	2.1	18
63	Second family provides further evidence for causation of Steel syndrome by biallelic mutations in <scp>COL27A1</scp>. Clinical Genetics, 2017, 92, 323-326.	1.0	18
64	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	2.6	18
65	FLNA mutations in surviving males presenting with connective tissue findings: two new case reports and review of the literature. BMC Medical Genetics, 2018, 19, 140.	2.1	18
66	Locus and allelic heterogeneity and phenotypic variability in Waardenburg syndrome. Clinical Genetics, 2019, 95, 398-402.	1.0	18
67	Synpolydactyly and HOXD13 polyalanine repeat: addition of 2 alanine residues is without clinical consequences. BMC Medical Genetics, 2007, 8, 78.	2.1	17
68	Additional three patients with Smithâ€McCort dysplasia due to novel RAB33B mutations. American Journal of Medical Genetics, Part A, 2017, 173, 588-595.	0.7	17
69	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	2.6	17
70	A novel multiple joint dislocation syndrome associated with a homozygous nonsense variant in the EXOC6B gene. European Journal of Human Genetics, 2016, 24, 1206-1210.	1.4	16
71	Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. American Journal of Human Genetics, 2019, 104, 439-453.	2.6	16
72	Biâ€allelic missense variant, p.<scp>Ser35Leu</scp> in <scp>EXOSC1</scp> is associated with pontocerebellar hypoplasia. Clinical Genetics, 2021, 99, 594-600.	1.0	16

#	Article	IF	Citations
73	A novel mutation (g.106737G>T) in zone of polarizing activity regulatory sequence (ZRS) causes variable limb phenotypes in Werner mesomelia. American Journal of Medical Genetics, Part A, 2014, 164, 898-906.	0.7	15
74	Spectrum of SMPD1 mutations in Asianâ€Indian patients with acid sphingomyelinase (ASM)â€deficient Niemannâ€“Pick disease. American Journal of Medical Genetics, Part A, 2016, 170, 2719-2730.	0.7	15
75	Report of four novel variants in ASNS causing asparagine synthetase deficiency and review of literature. Congenital Anomalies (discontinued), 2018, 58, 181-182.	0.3	15
76	Genetic diversity of NDUFV1-dependent mitochondrial complex I deficiency. European Journal of Human Genetics, 2018, 26, 1582-1587.	1.4	15
77	Meckel syndrome: Clinical and mutation profile in six fetuses. Clinical Genetics, 2019, 96, 560-565.	1.0	15
78	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	0.7	15
79	Torgâ€“Winchester syndrome: lack of efficacy of pamidronate therapy. Clinical Dysmorphology, 2007, 16, 95-100.	0.1	14
80	A biallelic 36-bp insertion in PIBF1 is associated with Joubert syndrome. Journal of Human Genetics, 2018, 63, 935-939.	1.1	14
81	Identification of a novel homozygous variant confirms ITPA as a developmental and epileptic encephalopathy gene. American Journal of Medical Genetics, Part A, 2019, 179, 857-861.	0.7	14
82	Bain type of Xâ€linked syndromic mental retardation in a male with a pathogenic variant in HNRNPH2. American Journal of Medical Genetics, Part A, 2020, 182, 183-188.	0.7	14
83	Bi-allelic variants in IPO8 cause a connective tissue disorder associated with cardiovascular defects, skeletal abnormalities, and immune dysregulation. American Journal of Human Genetics, 2021, 108, 1126-1137.	2.6	14
84	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	1.1	14
85	Report of second case and clinical and molecular characterization of Eiken syndrome. Clinical Genetics, 2018, 94, 457-460.	1.0	13
86	Development, behaviour and autism in individuals with SMC1A variants. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2019, 60, 305-313.	3.1	13
87	Genome sequencing in families with congenital limb malformations. Human Genetics, 2021, 140, 1229-1239.	1.8	13
88	Fetal akinesia deformation sequence: Expanding the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 2643-2648.	0.7	12
89	A Novel Frameshift Mutation in TWIST2 Gene Causing Setleis Syndrome. Indian Journal of Pediatrics, 2014, 81, 302-304.	0.3	12
90	Clinical and genetic spectrum of 104 Indian families with central nervous system white matter abnormalities. Clinical Genetics, 2021, 100, 542-550.	1.0	12

#	Article	IF	Citations
91	Exome Sequencing Identifies a Dominant <i>TNNT3</i> Mutation in a Large Family with Distal Arthrogryposis. Molecular Syndromology, 2014, 5, 218-228.	0.3	11
92	White matter changes in GM1 gangliosidosis. Indian Pediatrics, 2015, 52, 155-156.	0.2	11
93	Spectrum of urorectal septum malformation sequence. Congenital Anomalies (discontinued), 2016, 56, 119-126.	0.3	11
94	Variants in the transcriptional corepressor BCORL1 are associated with an Xâ€linked disorder of intellectual disability, dysmorphic features, and behavioral abnormalities. American Journal of Medical Genetics, Part A, 2019, 179, 870-874.	0.7	11
95	Mucolipidosis type II Î±/Î² with a homozygous missense mutation in the GNPTAB gene. American Journal of Medical Genetics, Part A, 2012, 158A, 1225-1228.	0.7	10
96	Hyperphosphatasia with Mental Retardation Syndrome Due to a Novel Mutation in PGAP3. Journal of Pediatric Genetics, 2017, 06, 191-193.	0.3	10
97	Seven additional families with spondylocarpotarsal synostosis syndrome with novel biallelic deleterious variants in FLNB. Clinical Genetics, 2018, 94, 159-164.	1.0	10
98	Confirmation of a Rare Genetic Leukoencephalopathy due to a Novel Bi-allelic Variant in RPIA. European Journal of Medical Genetics, 2019, 62, 103708.	0.7	10
99	Phenotyping and genotyping of skeletal dysplasias: Evolution of a center and a decade of experience in India. Bone, 2019, 120, 204-211.	1.4	10
100	Preimplantation diagnosis of genetic diseases. Journal of Postgraduate Medicine, 2010, 56, 317-320.	0.2	10
101	Novel biallelic variants expand the SLC5A6-related phenotypic spectrum. European Journal of Human Genetics, 2022, 30, 439-449.	1.4	10
102	A novel EDARADD 5â€²â€splice site mutation resulting in activation of two alternate cryptic 5â€²â€splice sites causes autosomal recessive Hypohidrotic Ectodermal Dysplasia. American Journal of Medical Genetics, Part A, 2016, 170, 1639-1641.	0.7	9
103	Novel variant p.(Ala102Thr) in SDHB causes mitochondrial complex II deficiency: Case report and review of the literature. Annals of Human Genetics, 2020, 84, 345-351.	0.3	9
104	Rhizomelic chondrodysplasia punctata type 1: report of mutations in 3 children from India. Journal of Applied Genetics, 2010, 51, 107-110.	1.0	8
105	Placental Teratoma Presenting as a Lobulated Mass behind the Neck of Fetus: A Case Report. Case Reports in Obstetrics and Gynecology, 2012, 2012, 1-2.	0.2	8
106	Profile of patients with Von Gierke disease from India. Indian Pediatrics, 2012, 49, 228-230.	0.2	8
107	The novel EDAR p.L397H missense mutation causes autosomal dominant hypohidrotic ectodermal dysplasia. Journal of the European Academy of Dermatology and Venereology, 2017, 31, e17-e20.	1.3	8
108	Homozygous c.359del variant in MGME1 is associated with early onset cerebellar ataxia. European Journal of Medical Genetics, 2017, 60, 533-535.	0.7	8

#	Article	IF	Citations
109	Variable presentation of Fraser syndrome in two fetuses and a novel mutation in FRAS1. Congenital Anomalies (discontinued), 2017, 57, 83-85.	0.3	8
110	Report of the Third Family with Multiple Mitochondrial Dysfunctions Syndrome 5 Caused by the Founder Variant p.(Glu87Lys) in ISCA1. Journal of Pediatric Genetics, 2018, 07, 130-133.	0.3	8
111	Homozygous variant, p.(Arg643Trp) in VAC14 causes striatonigral degeneration: report of a novel variant and review of VAC14-related disorders. Journal of Human Genetics, 2019, 64, 1237-1242.	1.1	8
112	Phenotypic diversity and genetic complexity of PAX3 â€related Waardenburg syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2951-2958.	0.7	8
113	Recurrent bi-allelic splicing variant c.454+3A>G in TRAPPC4 is associated with progressive encephalopathy and muscle involvement. Brain, 2020, 143, e29.	3.7	8
114	LACC1 gene mutation in three sisters with polyarthritis without systemic features. Annals of the Rheumatic Diseases, 2020, 79, 425-426.	0.5	8
115	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	1.1	8
116	Diagnosis and Management of Global Development Delay: Consensus Guidelines of Growth, Development and Behavioral Pediatrics Chapter, Neurology Chapter and Neurodevelopment Pediatrics Chapter of the Indian Academy of Pediatrics. Indian Pediatrics, 2022, 59, 401-415.	0.2	8
117	Immunological response to two hepatitis B vaccines administered in two different schedules. Indian Journal of Pediatrics, 2006, 73, 489-491.	0.3	7
118	Novel mutation and atlantoaxial dislocation in two siblings from India with Dyggveâ€“Melchiorâ€“Clausen syndrome. European Journal of Medical Genetics, 2008, 51, 251-256.	0.7	7
119	Long-term efficacy of oral deferiprone in management of iron overload in beta thalassemia major. Hematology, 2008, 13, 77-82.	0.7	7
120	Massive cranial osteolysis, skin changes, growth retardation and developmental delay: Gorham syndrome with systemic manifestations?. American Journal of Medical Genetics, Part A, 2010, 152A, 759-763.	0.7	7
121	Second report of slipped capital femoral epiphysis in Rubinsteinâ€“Taybi syndrome. Clinical Dysmorphology, 2011, 20, 55-57.	0.1	7
122	Occurrence of Synpolydactyly and Omphalocele in a Fetus with a HOXD13 Mutation. Journal of Pediatric Genetics, 2017, 06, 194-197.	0.3	7
123	Bi-allelic c.181_183delTGT in BTB domain of KLHL7 is associated with overlapping phenotypes of Crisponi/CISS1-like and Bohring-Opitz like syndrome. European Journal of Medical Genetics, 2019, 62, 103528.	0.7	7
124	The third family with Eiken syndrome. Clinical Genetics, 2019, 96, 378-379.	1.0	7
125	A novel biâ€allelic lossâ€ofâ€function variant in MYOD1: Further evidence for geneâ€disease association and phenotypic variability in MYOD1â€related myopathy. Clinical Genetics, 2019, 96, 276-277.	1.0	7
126	Locus and allelic heterogeneity in five families with hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 17-21.	1.1	7

#	Article	IF	Citations
127	Clinical Characteristics, Molecular Profile, and Outcomes in Indian Patients with Glutaric Aciduria Type 1. Journal of Pediatric Genetics, 2021, 10, 213-221.	0.3	7
128	Genetic disorders with central nervous system white matter abnormalities: An update. Clinical Genetics, 2021, 99, 119-132.	1.0	7
129	Deficiency of TMEM53 causes a previously unknown sclerosing bone disorder by dysregulation of BMP-SMAD signaling. Nature Communications, 2021, 12, 2046.	5.8	7
130	Multilocus disease-causing genomic variations for Mendelian disorders: role of systematic phenotyping and implications on genetic counselling. European Journal of Human Genetics, 2021, 29, 1774-1780.	1.4	7
131	Clinically relevant variants in a large cohort of Indian patients with Marfan syndrome and related disorders identified by next-generation sequencing. Scientific Reports, 2021, 11, 764.	1.6	7
132	Familial 7q11.23 duplication with variable phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 2727-2730.	0.7	6
133	Clinical Variability in Familial X-Linked Ohdo Syndromeâ€“Maat-Kievit-Brunner Type with MED12 Mutation. Journal of Pediatric Genetics, 2017, 06, 198-204.	0.3	6
134	Pycnodysostosis: Novel Variants in CTSK and Occurrence of Giant Cell Tumor. Journal of Pediatric Genetics, 2018, 07, 009-013.	0.3	6
135	Biallelic variants p.Arg1133Cys and p.Arg1379Cys in COL2A1: Further delineation of phenotypic spectrum of recessive Type 2 collagenopathies. American Journal of Medical Genetics, Part A, 2020, 182, 338-347.	0.7	6
136	Late onset Pompe Disease in India â€“ Beyond the Caucasian phenotype. Neuromuscular Disorders, 2021, 31, 431-441.	0.3	6
137	Functional characterization of novel variants in SMPD1 in Indian patients with acid sphingomyelinase deficiency. Human Mutation, 2021, 42, 1336-1350.	1.1	6
138	Milder form of pachydermoperiostosis: a report of four cases. Clinical Dysmorphology, 2009, 18, 85-89.	0.1	5
139	A syndrome of facial dysmorphism, cubital pterygium, short distal phalanges, swan neck deformity of fingers, and scoliosis. American Journal of Medical Genetics, Part A, 2014, 164, 1035-1040.	0.7	5
140	Is coloboma a feature of fetal valproate syndrome?. Clinical Dysmorphology, 2014, 23, 24-25.	0.1	5
141	Hunter syndrome with late age of presentation: clinical description of a case and review of the literature. BMJ Case Reports, 2015, 2015, bcr2015209305-bcr2015209305.	0.2	5
142	Homozygous deletion of exons 2 and 3 of NPC2 associated with Niemannâ€“Pick disease type C. American Journal of Medical Genetics, Part A, 2016, 170, 2486-2489.	0.7	5
143	Intrafamilial variability in syndromic microphthalmia type 5 caused by a novel variation inOTX2. Ophthalmic Genetics, 2017, 38, 533-536.	0.5	5
144	Severe Form of Brachydactyly Type A1 in a Child with a c.298Gâ€‰>â€‰A Mutation in IHH Gene. Journal of Pediatric Genetics, 2017, 06, 177-180.	0.3	5

#	Article	IF	Citations
145	p.Arg69Trp in RNASEH2C is a founder variant in three Indian families with Aicardiâ€“GoutiÃ¨res syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 156-160.	0.7	5
146	Novel pathogenic variants in GBE1 causing fetal akinesia deformation sequence and severe neuromuscular form of glycogen storage disease type IV. Clinical Dysmorphology, 2019, 28, 17-21.	0.1	5
147	Identification of novel variants in a large cohort of children with Tayâ€“Sachs disease: An initiative of a multicentric task force on lysosomal storage disorders by Government of India. Journal of Human Genetics, 2019, 64, 985-994.	1.1	5
148	GATAD2B-related intellectual disability due to parental mosaicism and review of literature. Clinical Dysmorphology, 2019, 28, 190-194.	0.1	5
149	Recurrent 1q21.1 deletion syndrome: report on variable expression, nonpenetrance and review of literature. Clinical Dysmorphology, 2020, 29, 127-131.	0.1	5
150	Digital clubbing as the predominant manifestation of hypertrophic osteoarthropathy caused by pathogenic variants in HPGD in three Indian families. Clinical Dysmorphology, 2020, 29, 123-126.	0.1	5
151	Genomic Testing for Diagnosis of Genetic Disorders in Children: Chromosomal Microarray and Nextâ€”Generation Sequencing. Indian Pediatrics, 2020, 57, 549-554.	0.2	5
152	Congenital and inherited ophthalmologic abnormalities. Indian Journal of Pediatrics, 2003, 70, 549-552.	0.3	4
153	Research letters. Indian Pediatrics, 2015, 52, 73-76.	0.2	4
154	Phenotypic variability in patients with interstitial 6q21â€q22 microdeletion and Acroâ€“Cardioâ€“Facial syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2998-3003.	0.7	4
155	India Allele Finder: a web-based annotation tool for identifying common alleles in next-generation sequencing data of Indian origin. BMC Research Notes, 2017, 10, 233.	0.6	4
156	Introducing in AJMG Part A: Case reports in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1547-1548.	0.7	4
157	Shortâ€term response to phenytoin sodium in Andersenâ€Tawil syndromeâ€1 with a cardiacâ€dominant phenotype. PACE - Pacing and Clinical Electrophysiology, 2019, 42, 201-207.	0.5	4
158	Roberts syndrome in an Indian patient with humeroradial synostosis, congenital elbow contractures and a novel homozygous splice variant in ESCO2. American Journal of Medical Genetics, Part A, 2020, 182, 2793-2796.	0.7	4
159	Biallelic start loss variant, c.<scp>1A</scp>Â>â€‰G in <scp>GCSH</scp> is associated with variant nonketotic hyperglycinemia. Clinical Genetics, 2021, 100, 201-205.	1.0	4
160	TNFR2 gene polymorphism in coronary artery disease. Indian Journal of Medical Sciences, 2005, 59, 104.	0.1	4
161	Clinical and Genetic Spectrum of Inborn Errors of Immunity in a Tertiary Care Center in Southern India. Indian Journal of Pediatrics, 2022, 89, 233-242.	0.3	4
162	S252W mutation in Indian patients of Apert syndrome. Indian Pediatrics, 2006, 43, 733-5.	0.2	4

#	Article	IF	Citations
163	Anotia and facial palsy: Unusual features of cardiofacial syndrome. Indian Journal of Pediatrics, 2005, 72, 525-526.	0.3	3
164	Opsismodysplasia. Indian Journal of Pediatrics, 2010, 77, 567-568.	0.3	3
165	Novel mutation in an Indian patient with Methylmalonic Acidemia, cblA type. Indian Journal of Human Genetics, 2012, 18, 346.	0.7	3
166	Further characterization of acro-renal-uterine-mandibular syndrome. Clinical Dysmorphology, 2012, 21, 83-86.	0.1	3
167	Jejunal atresia and postaxial polydactyly. Clinical Dysmorphology, 2013, 22, 121-123.	0.1	3
168	Coâ€•occurrence of a de novo Williams and 22q11.2 microdeletion syndromes. American Journal of Medical Genetics, Part A, 2015, 167, 1927-1931.	0.7	3
169	Novel Mutation in an Indian Patient with Transcobalamin II Deficiency. Indian Journal of Pediatrics, 2015, 82, 1073-1074.	0.3	3
170	Metatropic dysplasia with a novel mutation in TRPV4. Indian Pediatrics, 2016, 53, 735-737.	0.2	3
171	A reply to a commentary on homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. Journal of Human Genetics, 2017, 62, 867-867.	1.1	3
172	Further evidence for causation of ischiospinal dysostosis by a pathogenic variant in BMPER and expansion of the phenotype. Congenital Anomalies (discontinued), 2019, 59, 26-27.	0.3	3
173	An emerging ribosomopathy affecting the skeleton due to biallelic variations in NEPRO. American Journal of Medical Genetics, Part A, 2019, 179, 1709-1717.	0.7	3
174	Cover Image, Volume 179A, Number 5, May 2019. , 2019, 179, i-i.		3
175	Whole exome sequencing identifies a novel pathogenic variation [p.(Gly194valfs*7)] in SLC45A2 in the homozygous state in multiple members of a family with oculocutaneous albinism in southern India. Clinical and Experimental Dermatology, 2020, 45, 409-413.	0.6	3
176	Identification and characterization of 30 novel pathogenic variations in 69 unrelated Indian patients with Mucolipidosis Type II and Type III. Journal of Human Genetics, 2020, 65, 971-984.	1.1	3
177	Bosleyâ€“Salihâ€“Alorainy syndrome in patients from India. American Journal of Medical Genetics, Part A, 2020, 182, 2699-2703.	0.7	3
178	Trichothiodystrophy type 4 in an Indian family. American Journal of Medical Genetics, Part A, 2020, 182, 2226-2229.	0.7	3
179	Spastic Paraplegia Type 56 in a Young Child. Indian Journal of Pediatrics, 2020, 87, 650-651.	0.3	3
180	Biallelic c.1263dupC in DOK7 results in fetal akinesia deformation sequence. American Journal of Medical Genetics, Part A, 2020, 182, 804-807.	0.7	3

#	Article	IF	Citations
181	Wiedemannâ€“Rautenstrauch syndrome in an Indian patient with biallelic pathogenic variants in POLR3A. American Journal of Medical Genetics, Part A, 2021, 185, 1602-1605.	0.7	3
182	Hedgehog acylâ€transferaseâ€related multiple congenital anomalies: Report of an additional family and delineation of the syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2756-2765.	0.7	3
183	Biallelic deep intronic variant c.5457+81T>A in TRIP11 causes loss of function and results in achondrogenesis 1A. Human Mutation, 2021, 42, 1005-1014.	1.1	3
184	Focal dermal hypoplasia with a de novo mutation p.e300FNx01 of porcn gene in a male infant. Indian Journal of Dermatology, 2016, 61, 700.	0.1	3
185	A new autosomal recessive disorder of bilateral frontotemporal pachygyria without microcephaly: Report of a case and review of literature. Neurology India, 2007, 55, 57.	0.2	3
186	Expanding the spectrum of syndromic PPP2R3Câ€related <scp>XY</scp> gonadal dysgenesis to <scp>XX</scp> gonadal dysgenesis. Clinical Genetics, 2022, 101, 221-232.	1.0	3
187	Issues in counseling for Down syndrome. Indian Pediatrics, 2007, 44, 131-3.	0.2	3
188	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	3.7	3
189	A homozygous hypomorphicBNIP1variant causes an increase in autophagosomes and reduced autophagic flux and results in a spondyloâ€epiphyseal dysplasia. Human Mutation, 2022, 43, 625-642.	1.1	3
190	Ectodysplasin pathogenic variants affecting the furinâ€cleavage site and unusual clinical features define Xâ€linked hypohidrotic ectodermal dysplasia in India. American Journal of Medical Genetics, Part A, 2022, 188, 788-805.	0.7	3
191	Compound heterozygosity for HbD Punjab and polyadenylation signal mutation causes clinically asymptomatic mild hypochromia and microcytosis. Annals of Hematology, 2010, 89, 625-626.	0.8	2
192	Analysis of cosegregation of intragenic DNA sequence variations as markers of maternal cell contamination in prenatal diagnosis of Î²-thalassemia. Translational Research, 2011, 157, 150-155.	2.2	2
193	Growth retardation, intellectual disability, facial anomalies, cataract, thoracic hypoplasia, and skeletal abnormalities: A novel phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2941-2945.	0.7	2
194	What does fetal autopsy unmask in oligohydramnios?. Journal of Maternal-Fetal and Neonatal Medicine, 2015, 29, 1-5.	0.7	2
195	Symmetrical Terminal Transverse Limb Deficiencies. Indian Journal of Pediatrics, 2015, 82, 478-479.	0.3	2
196	Congenital omphalocele and cleft palate in two fetuses. Congenital Anomalies (discontinued), 2016, 56, 190-191.	0.3	2
197	Novel splice site and nonsense variants in INVS cause infantile nephronophthisis. Gene, 2020, 729, 144229.	1.0	2
198	Untapped opportunities for rare disease gene discovery in India. American Journal of Medical Genetics, Part A, 2020, 182, 3056-3059.	0.7	2

#	Article	IF	Citations
199	A review of skeletal dysplasia research in India. Journal of Postgraduate Medicine, 2018, 64, 98-103.	0.2	2
200	Handless, footless fetus. Clinical Dysmorphology, 2006, 15, 233-234.	0.1	1
201	Loss of a condyle of the femur or tibia following septic arthritis in infancy: problems of management and testing of a hypothesis of pathogenesis. Journal of Children's Orthopaedics, 2012, 6, 319-325.	0.4	1
202	Prenatal diagnosis of absent pulmonary valve confirmed by autopsy. BMJ Case Reports, 2013, 2013, bcr2012007997-bcr2012007997.	0.2	1
203	CFTR mutations in India: Need to do more! GENETICISTâ€™S PERSPECTIVE. Indian Pediatrics, 2014, 51, 177-178.	0.2	1
204	Unusual skin manifestations in a patient with menkes disease. American Journal of Medical Genetics, Part A, 2016, 170, 3039-3040.	0.7	1
205	Spectrum of mutations in the SMPD1 gene in Asian Indian patients with acid sphingomyelinase deficient Niemannâ€“Pick disease. American Journal of Medical Genetics, Part A, 2017, 173, 829-829.	0.7	1
206	Ichthyosis congenita with biliary atresia: a rare association. Clinical Dysmorphology, 2017, 26, 179-180.	0.1	1
207	Complexities in Genotype-Phenotype Correlation and Genetic Counseling in Collagen VI - Related Myopathy. Indian Journal of Pediatrics, 2017, 84, 330-331.	0.3	1
208	Facial profile and additional features in fetuses with trisomy 21. Clinical Dysmorphology, 2018, 27, 126-129.	0.1	1
209	Mutation Spectrum of Tuberous Sclerosis Complex Patients in Indian Population. Journal of Pediatric Genetics, 2021, 10, 274-283.	0.3	1
210	Three M syndrome 2 in two Indian patients. American Journal of Medical Genetics, Part A, 2021, 185, 614-616.	0.7	1
211	Understanding Exome Sequencing: Tips for the Pediatrician. Indian Pediatrics, 2021, 58, 771-774.	0.2	1
212	Expanding the phenotype of PURA-related neurodevelopmental disorder: a close differential diagnosis of infantile hypotonia with psychomotor retardation and characteristic facies. Clinical Dysmorphology, 2021, 30, 1-5.	0.1	1
213	Middle Interhemispheric Variant of Holoprosencephaly â€“ Presenting as Non- Visualized Cavum Septum Pellucidum and An Interhemispheric Cyst in A 19-Weeks Fetus. Journal of Clinical and Diagnostic Research JCDR, 2015, 9, QD11-3.	0.8	1
214	Pseudoachondroplasia: Phenotype and genotype in 11 Indian patients. American Journal of Medical Genetics, Part A, 2022, 188, 751-759.	0.7	1
215	Mongolian spots in GM1 gangliosidosis: a pictorial report. Clinical Dysmorphology, 2021, 30, 6-9.	0.1	1
216	Unusual facial cleft in Fryns syndrome: defect of stomodeum?. Genetic Counseling, 2010, 21, 233-6.	0.1	1

#	Article	IF	Citations
217	Steel syndrome: Report of three patients, including monozygotic twins and review of clinical and mutation profiles. European Journal of Medical Genetics, 2022, 65, 104521.	0.7	1
218	Biallelic <scp> KITLG </scp> variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss. Journal of the European Academy of Dermatology and Venereology, 2022, , .	1.3	1
219	Hypoplasia/aplasia of pelvis, femora, fibulae, ulna, digits and nails. Clinical Dysmorphology, 2011, 20, 205-209.	0.1	0
220	Umbilical cyst due to patent urachus in a fetus with complete urorectal septum malformation sequence. Journal of Pediatric Genetics, 2015, 01, 059-061.	0.3	0
221	Editorial. Indian Pediatrics, 2016, 53, 115-117.	0.2	0
222	Cover Image, Volume 173A, Number 3, March 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
223	Glycosaminoglycan levels in dried blood spots of patients with mucopolyasccharidoses and mucolipidoses. Molecular Genetics and Metabolism, 2017, 120, S76-S77.	0.5	0
224	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
225	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0
226	Recurrence of Trisomic Pregnancies in Four Families: A Cytogenetic and Molecular Study. Journal of Fetal Medicine, 2019, 06, 81-87.	0.1	0
227	FRI0578â€…LACC1 MUTATION IN THREE SIBLINGS WITH POLYARTHRITIS WITHOUT SYSTEMIC MANIFESTATIONS. , 2019, , .		0
228	Response to Hall etÂal.. American Journal of Human Genetics, 2020, 107, 1188-1189.	2.6	0
229	Bi-allelic variant c.8638_8658dup in HYDIN causes bronchiectasis in two siblings. Gene Reports, 2020, 21, 100961.	0.4	0
230	Explanation for mild and severe osteogenesis imperfecta phenotypes due to splice variants at c.2029-1 in COL1A1. Gene Reports, 2020, 21, 100803.	0.4	0
231	Myelomonocytic leukaemia (JMML) in a child with intellectual disability and chromosome 4q deletion. Pediatric Hematology Oncology Journal, 2021, 6, 88-90.	0.1	0
232	A celebration in honor of John M. Graham, Jr, <scp>MD</scp>, <scp>ScD</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 2617-2619.	0.7	0
233	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	2.6	0
234	Pfeiffer Syndrome with Extreme Proptosis, Hypothyroidism and Tail like Appendage. Oman Medical Journal, 2014, 29, e080.	0.3	0

#	Article	IF	Citations
235	Prenatal Diagnosis of Intracranial Tumors and the Difficulties in Prognostication: A Report of Three Cases. International Journal of Infertility and Fetal Medicine, 2015, 6, 88-91.	0.0	0
236	Novel ALOX12B mutation identified in parents following single nucleotide polymorphism microarray testing of banked DNA from a fatal case of congenital ichthyosis. Indian Journal of Dermatology, 2016, 61, 122.	0.1	0
237	Classical cleidocranial dysplasia in an adult, due to a novel frameshift pathogenic variant inRUNX2. BMJ Case Reports, 2016, 2016, bcr2016215162.	0.2	0
238	Knobloch Syndrome in Siblings with Posterior Fossa Malformations Along with Cerebellar Midline Cleft Abnormality Caused by Biallelic COL18A1 Mutation: Case-Based Review. Journal of Pediatric Genetics, 0, , .	0.3	0
239	C18orf32 loss-of-function is associated with a neurodevelopmental disorder with hypotonia and contractures. Human Genetics, 2022, , 1.	1.8	0
240	Balanced translocation in mother leading to interchange trisomy 21?. Genetic Counseling, 2010, 21, 183-5.	0.1	0
241	Hunting for Mutations in Indian Patients with Hunter Syndrome. Indian Pediatrics, 2016, 53, 117.	0.2	0
242	Genomic Testing for Diagnosis of Genetic Disorders in Children: Chromosomal Microarray and Next-Generation Sequencing. Indian Pediatrics, 2020, 57, 549-554.	0.2	0
243	Understanding Exome Sequencing: Tips for the Pediatrician. Indian Pediatrics, 2021, 58, 771-774.	0.2	0
244	Homozygous variant p.(Arg163Trp) in PIGH causes glycosylphosphatidylinositol biosynthesis defect with epileptic encephalopathy and delayed myelination. Clinical Dysmorphology, 2022, Publish Ahead of Print, .	0.1	0
245	Clinical, radiological and molecular studies in 24 individuals with Dyggve-Melchior-Clausen dysplasia and Smith-McCort dysplasia from India. Journal of Medical Genetics, 2023, 60, 204-211.	1.5	0