Shubha R Phadke

List of Publications by Year
in descending order

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239 papers	2,763 citations	257357 24 h-index	302012 39 g-index
242 all docs	242 docs citations	242 times ranked	4450 citing authors

#	Article	IF	Citations
1	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	2.6	137
2	De novo SOX11 mutations cause Coffinâ€“Siris syndrome. Nature Communications, 2014, 5, 4011.	5.8	118
3	Mutations in genes encoding condensin complex proteins cause microcephaly through decatenation failure at mitosis. Genes and Development, 2016, 30, 2158-2172.	2.7	106
4	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	1.5	85
5	Molecular and clinical heterogeneity in CLCN7-dependent osteopetrosis: report of 20 novel mutations. Human Mutation, 2010, 31, E1071-E1080.	1.1	77
6	Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. Human Genetics, 2012, 131, 1761-1773.	1.8	73
7	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	0.7	68
8	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
9	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	0.5	57
10	Genetic insight of schizophrenia: past and future perspectives. Gene, 2014, 535, 97-100.	1.0	49
11	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	0.7	40
12	Mutations in patients with osteogenesis imperfecta from consanguineous Indian families. European Journal of Medical Genetics, 2015, 58, 21-27.	0.7	37
13	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
14	OEIS complex with craniofacial anomaliesâ€”defect of blastogenesis?. American Journal of Medical Genetics Part A, 1994, 53, 21-23.	2.4	32
15	Vascular endothelial growth factor gene polymorphisms in North Indian patients with recurrent miscarriages. Reproductive BioMedicine Online, 2011, 22, 59-64.	1.1	31
16	GALNS mutations in Indian patients with mucopolysaccharidosis IVA. American Journal of Medical Genetics, Part A, 2014, 164, 2793-2801.	0.7	31
17	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. American Journal of Medical Genetics, Part A, 2016, 170, 410-417.	0.7	31
18	Status of 25-hydroxyvitamin D deficiency and effect of vitamin D receptor gene polymorphisms on bone mineral density in thalassemia patients of North India. Hematology, 2012, 17, 291-296.	0.7	30

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19	Myotonic dystrophy type 1 (DM1): A triplet repeat expansion disorder. Gene, 2013, 522, 226-230.	1.0	30
20	Spectrum of lysosomal storage disorders at a medical genetics center in Northern India. Indian Pediatrics, 2012, 49, 799-804.	0.2	29
21	Morphometric analysis of face in dysmorphology. Computer Methods and Programs in Biomedicine, 2007, 85, 165-172.	2.6	28
22	Newborn screening for congenital hypothyroidism, galactosemia and biotinidase deficiency in Uttar Pradesh, India. Indian Pediatrics, 2014, 51, 701-705.	0.2	28
23	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
24	Genetic Analysis of the SRD5A2 Gene in Indian Patients with 5Î±-Reductase Deficiency. Journal of Pediatric Endocrinology and Metabolism, 2009, 22, 247-54.	0.4	25
25	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
26	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
27	Medical genetics and genomic medicine in India: current status and opportunities ahead. Molecular Genetics & Genomic Medicine, 2015, 3, 160-171.	0.6	24
28	Comparison of prenatal ultrasound findings and autopsy findings in fetuses terminated after prenatal diagnosis of malformations: An experience of a clinical genetics center. Journal of Clinical Ultrasound, 2010, 38, 244-249.	0.4	23
29	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
30	Recurrent and novel GLB1 mutations in India. Gene, 2015, 567, 173-181.	1.0	22
31	Connexin 26 (GJB2) Mutations Associated with Non-Syndromic Hearing Loss (NSHL). Indian Journal of Pediatrics, 2018, 85, 1061-1066.	0.3	22
32	Further delineation of a new (Van Den Ende-Gupta) syndrome of blepharophimosis, contractural arachnodactyly, and characteristic face. American Journal of Medical Genetics Part A, 1998, 77, 16-18.	2.4	21
33	Phenotype score to grade the severity of thalassemia intermedia. Indian Journal of Pediatrics, 2003, 70, 477-481.	0.3	21
34	Cutis Laxa Type II and Wrinkly Skin Syndrome: Distinct Phenotypes. Pediatric Dermatology, 2006, 23, 225-230.	0.5	21
35	Molecular and structural analysis of metachromatic leukodystrophy patients in Indian population. Journal of the Neurological Sciences, 2011, 301, 38-45.	0.3	21
36	Novel and recurrent mutations in WISP3 and an atypical phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 2481-2484.	0.7	21

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37	Implication of HLA-G 5â€² upstream regulatory region polymorphisms in idiopathic recurrent spontaneous abortions. Reproductive BioMedicine Online, 2015, 30, 82-91.	1.1	21
38	Prenatal screening for genetic disorders: Suggested guidelines for the Indian Scenario. Indian Journal of Medical Research, 2017, 146, 689.	0.4	21
39	Syndromic versus nonsyndromic atlantoaxial dislocation: do clinico-radiological differences have a bearing on management?. Acta Neurochirurgica, 2013, 155, 1157-1167.	0.9	20
40	Association of functional genetic variants of CTLA4 with reduced serum CTLA4 protein levels and increased risk of idiopathic recurrent miscarriages. Fertility and Sterility, 2016, 106, 1115-1123.e6.	0.5	20
41	Distal arthrogryposis type 5D with a novel ECEL1 gene mutation. American Journal of Medical Genetics, Part A, 2014, 164, 2857-2862.	0.7	19
42	Diagnosis and Management of Gaucher Disease in India â€“ Consensus Guidelines of the Gaucher Disease Task Force of the Society for Indian Academy of Medical Genetics and the Indian Academy of Pediatrics. Indian Pediatrics, 2018, 55, 143-153.	0.2	19
43	SMARCE1, a rare cause of Coffinâ€“Siris Syndrome: Clinical description of three additional cases. American Journal of Medical Genetics, Part A, 2016, 170, 1967-1973.	0.7	18
44	Hemihyperplasia syndromes. Indian Journal of Pediatrics, 2006, 73, 609-615.	0.3	17
45	Unbalanced X; autosome translocation. Indian Journal of Pediatrics, 2006, 73, 840-842.	0.3	17
46	Synpolydactyly and HOXD13 polyalanine repeat: addition of 2 alanine residues is without clinical consequences. BMC Medical Genetics, 2007, 8, 78.	2.1	17
47	Hemiconvulsionâ€“hemiplegiaâ€“epilepsy syndrome with 1q44 microdeletion: Causal or chance association. American Journal of Medical Genetics, Part A, 2014, 164, 186-189.	0.7	17
48	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
49	Socio-demographic Profile and Economic Burden of Treatment of Transfusion Dependent Thalassemia. Indian Journal of Pediatrics, 2018, 85, 102-107.	0.3	17
50	Pycnodysostosis: mutation spectrum in five unrelated Indian children. Clinical Dysmorphology, 2016, 25, 113-120.	0.1	16
51	Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. American Journal of Human Genetics, 2019, 104, 439-453.	2.6	16
52	Carrier analysis and prenatal diagnosis of haemophilia A in North India. International Journal of Molecular Medicine, 2002, 10, 661-4.	1.8	16
53	Novel mutations in Indian patients with autosomal recessive infantile malignant osteopetrosis. Indian Journal of Medical Research, 2010, 131, 508-14.	0.4	16
54	Urorectal septum malformation sequence: Ultrasound correlation with fetal examination. Indian Journal of Pediatrics, 2006, 73, 287-293.	0.3	15

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55	Utility of MLPA in mutation analysis and carrier detection for Duchenne muscular dystrophy. Indian Journal of Human Genetics, 2012, 18, 91.	0.7	15
56	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
57	Spondyloepiphyseal dysplasia Omani type: CHST3 mutation spectrum and phenotypes in three Indian families. American Journal of Medical Genetics, Part A, 2017, 173, 163-168.	0.7	15
58	Spectrum of ARSA variations in Asian Indian patients with Arylsulfatase A deficient metachromatic leukodystrophy. Journal of Human Genetics, 2019, 64, 323-331.	1.1	15
59	Computer-aided Facial Analysis in Diagnosing Dysmorphic Syndromes in Indian Children. Indian Pediatrics, 2019, 56, 1017-1019.	0.2	15
60	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	0.7	15
61	Torgâ€“Winchester syndrome: lack of efficacy of pamidronate therapy. Clinical Dysmorphology, 2007, 16, 95-100.	0.1	14
62	Diagnosis of Down Syndrome and Detection of Origin of Nondisjunction by Short Tandem Repeat Analysis. Genetic Testing and Molecular Biomarkers, 2010, 14, 489-491.	0.3	14
63	Recurrence of urorectal septum malformation sequence spectrum anomalies in siblings: Time to explore the genetics. American Journal of Medical Genetics, Part A, 2013, 161, 1718-1721.	0.7	14
64	Hemophilia Care in India: A Review and Experience from a Tertiary Care Centre in Uttar Pradesh. Indian Journal of Hematology and Blood Transfusion, 2011, 27, 121-126.	0.3	13
65	Late termination of pregnancy for fetal abnormalities: The perspective of Indian lay persons and medical practitioners. Prenatal Diagnosis, 2011, 31, 1286-1291.	1.1	13
66	Use of Multiplex Ligation-Dependent Probe Amplification (MLPA) in screening of subtelomeric regions in children with idiopathic mental retardation. Indian Journal of Pediatrics, 2009, 76, 1027-1031.	0.3	12
67	Multiplex Quantitative Fluorescent Polymerase Chain Reaction for Detection of Aneuploidies. Genetic Testing and Molecular Biomarkers, 2012, 16, 624-627.	0.3	12
68	Knowledge of Cord Blood Banking in General Population and Doctors: A Questionnaire Based Survey. Indian Journal of Pediatrics, 2016, 83, 238-241.	0.3	12
69	Next Generation Sequencing in Diagnosis of MLPA Negative Cases Presenting as Duchenne/ Becker Muscular Dystrophies. Indian Journal of Pediatrics, 2018, 85, 309-310.	0.3	12
70	Whole exome sequencing reveals a mutation in ARMC9 as a cause of mental retardation, ptosis, and polydactyly. American Journal of Medical Genetics, Part A, 2018, 176, 34-40.	0.7	12
71	Biochemical and molecular characterization of adult patients with type I Gaucher disease and carrier frequency analysis of Leu444Pro - a common Gaucher disease mutation in India. BMC Medical Genetics, 2018, 19, 178.	2.1	12
72	Polydactyly and genes. Indian Journal of Pediatrics, 2010, 77, 277-281.	0.3	11

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73	Recurrent Pregnancy Loss and Apolipoprotein E Gene Polymorphisms: A Caseâ€“Control Study from North India. American Journal of Reproductive Immunology, 2010, 64, 172-178.	1.2	11
74	White matter changes in GM1 gangliosidosis. Indian Pediatrics, 2015, 52, 155-156.	0.2	11
75	Complex Camptosynpolydactyly and Mesoaxial synostotic syndactyly with phalangeal reduction are allelic disorders. American Journal of Medical Genetics, Part A, 2016, 170, 1622-1625.	0.7	11
76	Sequence variations in TENM3 gene causing eye anomalies with intellectual disability: Expanding the phenotypic spectrum. European Journal of Medical Genetics, 2019, 62, 61-64.	0.7	11
77	GAPO syndrome in a child without dermal hyaline deposit. American Journal of Medical Genetics Part A, 1994, 51, 191-193.	2.4	10
78	COL1A1 Mutation in an Indian Child with Caffey Disease. Indian Journal of Pediatrics, 2011, 78, 877-879.	0.3	10
79	Platelet-specific collagen receptor glycoprotein VI gene variants affect recurrent pregnancy loss. Fertility and Sterility, 2014, 102, 1078-1084.e3.	0.5	10
80	Worsening of callus hyperplasia after bisphosphonate treatment in type V osteogenesis imperfecta. Indian Pediatrics, 2016, 53, 250-252.	0.2	10
81	Study of the association of forkhead box P3 (FOXP3) gene polymorphisms with unexplained recurrent spontaneous abortions in Indian population. Journal of Genetics, 2018, 97, 405-410.	0.4	10
82	Genetic counseling. Indian Journal of Pediatrics, 2004, 71, 151-156.	0.3	9
83	Analysis of Short Stature Cases Referred for Genetic Evaluation. Indian Journal of Pediatrics, 2012, 79, 1597-1600.	0.3	9
84	Spectrum of genomic variations in Indian patients with progressive familial intrahepatic cholestasis. BMC Gastroenterology, 2018, 18, 107.	0.8	9
85	Novel NLRP12 variant presenting with familial cold autoimmunity syndrome phenotype. Annals of the Rheumatic Diseases, 2021, 80, e117-e117.	0.5	9
86	Novel mutations of the arylsulphatase B (ARSB) gene in Indian patients with mucopolysaccharidosis type VI. Indian Journal of Medical Research, 2015, 142, 414.	0.4	9
87	Clinical utility of multiplex ligation-dependent probe amplification technique in identification of aetiology of unexplained mental retardation: a study in 203 Indian patients. Indian Journal of Medical Research, 2014, 139, 66-75.	0.4	9
88	Linear catch-up growth. Indian Journal of Pediatrics, 2000, 67, 225-230.	0.3	8
89	Profile of patients with Von Gierke disease from India. Indian Pediatrics, 2012, 49, 228-230.	0.2	8
90	Genetic variation of TBX21 gene increases risk of asthma and its severity in Indian children. Journal of Human Genetics, 2014, 59, 437-443.	1.1	8

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91	Study of Polymorphisms in CX3CR1, PLEKHA1 and VEGF Genes as Risk Factors for Age-related Macular Degeneration in Indian Patients. Archives of Medical Research, 2014, 45, 489-494.	1.5	8
92	Williams-Beuren Syndrome: Experience of 43 Patients and a Report of an Atypical Case from a Tertiary Care Center in India. Cytogenetic and Genome Research, 2015, 146, 187-194.	0.6	8
93	Performance of QF-PCR in targeted prenatal aneuploidy diagnosis: Indian scenario. Gene, 2015, 562, 55-61.	1.0	8
94	Sialidosis type II: Expansion of phenotypic spectrum and identification of a common mutation in seven patients. Molecular Genetics and Metabolism Reports, 2020, 22, 100561.	0.4	8
95	Homozygous Missense Variation in <i>PNPLA8</i> Causes Prenatal-Onset Severe Neurodegeneration. Molecular Syndromology, 2021, 12, 174-178.	0.3	8
96	Homozygosity stretches around homozygous mutations in autosomal recessive disorders: patients from nonconsanguineous Indian families. Journal of Genetics, 2021, 100, 1.	0.4	8
97	Partial trisomy 13 with features similar to C syndrome. Indian Pediatrics, 2004, 41, 614-7.	0.2	8
98	Mental retardation, ptosis and polydactyly: a new autosomal recessive syndrome?. Clinical Dysmorphology, 2002, 11, 289-292.	0.1	7
99	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
100	Long-term efficacy of oral deferiprone in management of iron overload in beta thalassemia major. Hematology, 2008, 13, 77-82.	0.7	7
101	Intracardiac echogenic focus and fetal outcome. Journal of Clinical Ultrasound, 2010, 38, 466-469.	0.4	7
102	Prenatal diagnosis of Pompe disease â€" Enzyme assay or molecular testing?. Indian Pediatrics, 2011, 48, 901-906.	0.2	7
103	The expanding spectrum of Elejalde syndrome. Clinical Dysmorphology, 2011, 20, 98-101.	0.1	7
104	Novel mutations in the transmembrane natriuretic peptide receptor NPR-B gene in four Indian families with acromesomelic dysplasia, type Maroteaux. Journal of Genetics, 2016, 95, 905-909.	0.4	7
105	Carrier frequency of <scp>SMN1</scp>â€related spinal muscular atrophy in north Indian population: The need for population based screening program. American Journal of Medical Genetics, Part A, 2021, 185, 274-277.	0.7	7
106	Use of strain, strain rate, tissue velocity imaging, and endothelial function for early detection of cardiovascular involvement in patients with beta-thalassemia. Annals of Pediatric Cardiology, 2017, 10, 158.	0.2	7
107	Aetiologic spectrum of mental retardation & developmental delay in India. Indian Journal of Medical Research, 2012, 136, 436-44.	0.4	7
108	Preaxial brachydactyly with abduction of thumbs and hallux varus: A distinct entity. American Journal of Medical Genetics Part A, 1994, 49, 274-277.	2.4	6

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109	Complex camptopolydactyly: An unusual hand malformation. American Journal of Medical Genetics Part A, 1999, 83, 191-192.	2.4	6
110	Recurrence of complex camptopolydactyly in a sibling suggestive of autosomal recessive mode of inheritance. American Journal of Medical Genetics Part A, 2003, 116A, 94-96.	2.4	6
111	Knowledge and Attitudes Towards Haemophilia: The Family Side and Role of Haemophilia Societies. Public Health Genomics, 2003, 6, 120-122.	0.6	6
112	Application of a reliable and rapid polymerase chain reaction based method in the diagnosis of myotonic dystrophy type 1 (DM1) in India. Meta Gene, 2014, 2, 106-113.	0.3	6
113	Feasibility Study of an Outreach Program of Newborn Screening in Uttar Pradesh. Indian Journal of Pediatrics, 2015, 82, 427-432.	0.3	6
114	Extending the phenotype and an ECEL1 gene mutation in distal arthrogryposis type 5D. Clinical Dysmorphology, 2018, 27, 130-134.	0.1	6
115	Concepts, Utility and Limitations of Cord Blood Banking: What Clinicians Need to Know. Indian Journal of Pediatrics, 2019, 86, 44-48.	0.3	6
116	Genetic heterogeneity of disorders with overgrowth and intellectual disability: Experience from a center in North India. American Journal of Medical Genetics, Part A, 2021, 185, 2345-2355.	0.7	6
117	Phenotypic and genotypic spectrum of CTSK variants in a cohort of twenty-five Indian patients with pycnodysostosis. European Journal of Medical Genetics, 2021, 64, 104235.	0.7	6
118	Functional characterization of novel variants in SMPD1 in Indian patients with acid sphingomyelinase deficiency. Human Mutation, 2021, 42, 1336-1350.	1.1	6
119	The Clinical Value of a â€˜Limited Fetal Autopsyâ€™ as Illustrated by 2 Cases. Australian and New Zealand Journal of Obstetrics and Gynaecology, 1994, 34, 111-113.	0.4	5
120	Short stature, ulnar deviation of hands with absent carpals and joint contractures: a new syndrome. Clinical Dysmorphology, 2007, 16, 55-57.	0.1	5
121	Spondylothoracic dysplasia: Prenatal diagnosis and the problems of nosologic overlap. American Journal of Medical Genetics, Part A, 2007, 143A, 899-902.	0.7	5
122	Angelman syndrome and prenatally diagnosed Praderâ€“Willi syndrome in first cousins. American Journal of Medical Genetics, Part A, 2011, 155, 2788-2790.	0.7	5
123	Neural tube defects: A need for population-based prevention program. Indian Journal of Human Genetics, 2012, 18, 145.	0.7	5
124	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
125	Fibrodysplasia Ossificans Progressiva: Three Indian Patients with Mutation in the ACVR1 Gene. Indian Journal of Pediatrics, 2014, 81, 617-619.	0.3	5
126	Floating Harbor Syndrome. Indian Journal of Pediatrics, 2016, 83, 896-897.	0.3	5

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127	Vascular endothelial growth factor gene polymorphisms and association with age related macular degeneration in Indian patients. Meta Gene, 2016, 9, 249-253.	0.3	5
128	Smith-Magenis Syndrome: Face Speaks. Indian Journal of Pediatrics, 2016, 83, 589-593.	0.3	5
129	Hotspots in PTPN11 gene among Indian children with Noonan syndrome. Indian Pediatrics, 2017, 54, 638-640.	0.2	5
130	Fanconi-Bickel Syndrome: Another Novel Mutation in SLC2A2. Indian Journal of Pediatrics, 2017, 84, 236-237.	0.3	5
131	Posterior reversible encephalopathy syndrome following blood transfusion in a patient with factor X deficiency: Is it an unusual systemic manifestation of an adverse transfusion reaction?. Transfusion and Apheresis Science, 2018, 57, 50-53.	0.5	5
132	Phenotypic characterization of derivative 22 syndrome: case series and review. Journal of Genetics, 2018, 97, 205-211.	0.4	5
133	A mild phenotype of LGI4-Related arthrogryposis multiplex congenita with intrafamilial variability. European Journal of Medical Genetics, 2020, 63, 103756.	0.7	5
134	Fabry disease: a treatable lysosomal storage disorder. The National Medical Journal of India, 2009, 22, 20-2.	0.1	5
135	Genetic counseling: The impact in Indian milieu. Indian Journal of Pediatrics, 2004, 71, 1079-1082.	0.3	4
136	Pericentric inversion causing duplication and deletion of chromosome region 13q22â€‰â†’â€‰qter in the offspring. American Journal of Medical Genetics, Part A, 2007, 143A, 82-84.	0.7	4
137	Severe form of congenital cerebral and cerebellar atrophy: A neurodegenerative disorder of fetal onset. Journal of Clinical Ultrasound, 2007, 35, 347-350.	0.4	4
138	Genetic testing in children. Indian Pediatrics, 2013, 50, 823-827.	0.2	4
139	Genetic variations of theFCER2gene and asthma susceptibility in north Indian children: a case-control study. Biomarkers, 2013, 18, 660-667.	0.9	4
140	Atlantoaxial dislocation in a child affected by warfarin embryopathy. Clinical Dysmorphology, 2013, 22, 124-126.	0.1	4
141	Research Letters. Indian Pediatrics, 2014, 51, 411-413.	0.2	4
142	Chondrodysplasia punctata tibia metacarpal type. Clinical Dysmorphology, 2015, 24, 118-121.	0.1	4
143	Novel sequence variations in the thymidine phosphorylase gene causing mitochondrial neurogastrointestinal encephalopathy. Clinical Dysmorphology, 2016, 25, 156-162.	0.1	4
144	Infantile Systemic Hyalinosis with Mutation in ANTXR2. Indian Journal of Pediatrics, 2016, 83, 1356-1357.	0.3	4

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145	A novel variant in MED12 gene: Further delineation of phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 2257-2260.	0.7	4
146	Malan syndrome: Extension of genotype and phenotype spectrum. American Journal of Medical Genetics, Part A, 2018, 176, 2896-2900.	0.7	4
147	Protein protein interaction network analysis of differentially expressed genes to understand involved biological processes in coronary artery disease and its different severity. Gene Reports, 2018, 12, 50-60.	0.4	4
148	Cytogenetic microarray in structurally normal and abnormal foetuses: a fiveÂyears experience elucidating increasing acceptance and clinical utility. Journal of Genetics, 2019, 98, 1.	0.4	4
149	Expanding the phenotype in autosomal dominant mental retardation-24: a novel variation in DEAF1 gene. Clinical Dysmorphology, 2019, 28, 94-97.	0.1	4
150	Clinical and Mutation Spectra of Cockayne Syndrome in India. Neurology India, 2021, 69, 362.	0.2	4
151	Clinical and molecular characterization of four patients with Robinow syndrome from different families. American Journal of Medical Genetics, Part A, 2021, 185, 1105-1112.	0.7	4
152	Monogenic Lupus with IgA Nephropathy Caused by Spondyloenchondrodysplasia with Immune Dysregulation. Indian Journal of Pediatrics, 2021, 88, 819-823.	0.3	4
153	Novel pathogenic variants in an Indian cohort with epidermolysis bullosa: Expanding the genotypic spectrum. European Journal of Medical Genetics, 2021, 64, 104345.	0.7	4
154	Fetal intra abdominal umbilical vein varix: Case series and review of literature. Indian Journal of Radiology and Imaging, 2017, 27, 59-61.	0.3	4
155	Exome sequencing & homozygosity mapping for identification of genetic aetiology for spastic ataxia in a consanguineous family. Indian Journal of Medical Research, 2015, 142, 220.	0.4	4
156	Recurrent benign copy number variants & issues in interpretation of variants of unknown significance identified by cytogenetic microarray in Indian patients with intellectual disability. Indian Journal of Medical Research, 2015, 142, 699.	0.4	4
157	Hyperekplexia: A forgotten diagnosis clinched by next-generation sequencing. Neurology India, 2017, 65, 1065.	0.2	4
158	Computer-aided Facial Analysis in Diagnosing Dysmorphic Syndromes in Indian Children. Indian Pediatrics, 2019, 56, 1017-1019.	0.2	4
159	Sociocultural and ethical dilemmas of genetic counseling?A suggested working approach. Journal of Genetic Counseling, 1994, 3, 81-83.	0.9	3
160	Pachygyria/hypogenitalism: A monogenic syndrome. American Journal of Medical Genetics Part A, 1999, 87, 254-257.	2.4	3
161	Further delineation of acro-renal-mandibular syndrome. Clinical Dysmorphology, 2006, 15, 119-120.	0.1	3
162	Identification of DKC1 gene mutation in an Indian patient. Indian Journal of Pediatrics, 2010, 77, 310-312.	0.3	3

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163	Report of two brothers with short stature, microcephaly, mental retardation, and retinoschisisâ€"A new mental retardation syndrome?. American Journal of Medical Genetics, Part A, 2011, 155, 9-13.	0.7	3
164	Cytogenetic microarray in prenatal and postnatal diagnosis. Molecular Cytogenetics, 2014, 7, I32.	0.4	3
165	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
166	Metatropic dysplasia with a novel mutation in TRPV4. Indian Pediatrics, 2016, 53, 735-737.	0.2	3
167	Hunter syndrome in northern India: Clinical features and mutation spectrum. Indian Pediatrics, 2016, 53, 134-136.	0.2	3
168	Consanguinity as an Adjunct Diagnostic Tool. Indian Journal of Pediatrics, 2016, 83, 258-260.	0.3	3
169	Pyruvate Carboxylase Deficiency Mimicking Diabetic Ketoacidosis. Indian Journal of Pediatrics, 2017, 84, 959-960.	0.3	3
170	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatrics, 2018, 55, 474-477.	0.2	3
171	Deletion 7q21.2-q22.1 in a case with split hand-split foot malformation, sensorineural hearing loss and intellectual disability: Phenotype subtypes and the correlation with genotypes. European Journal of Medical Genetics, 2019, 62, 103597.	0.7	3
172	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
173	Founder effects of the homogentisate 1,2-dioxygenase (HGD) gene in a gypsy population and mutation spectrum in the gene among alkaptonuria patients from India. Clinical Rheumatology, 2020, 39, 2743-2749.	1.0	3
174	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
175	Spectrum of prenatally detected central nervous system malformations: Neural tube defects continue to be the leading foetal malformation. Indian Journal of Medical Research, 2017, 145, 471-478.	0.4	3
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