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	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
2	Molecular analysis of severe hemophilia B in Indian families: Identification of mutational hotspot and novel variants. International Journal of Laboratory Hematology, 2022, 44, 186-192.	0.7	1
3	Pseudoachondroplasia: Phenotype and genotype in 11 Indian patients. American Journal of Medical Genetics, Part A, 2022, 188, 751-759.	0.7	1
4	Monosomy 1p36: Report of a cohort of 13 Asian Indian patients. American Journal of Medical Genetics, Part A, 2022, , .	0.7	0
5	Genotype-phenotype spectrum of 130 unrelated Indian families with Mucopolysaccharidosis type II. European Journal of Medical Genetics, 2022, 65, 104447.	0.7	3
6	Autosomal recessive spinocerebellar ataxiaâ€20 due to a novel <scp> SNX14 </scp> variant in an Indian girl. American Journal of Medical Genetics, Part A, 2022, , .	0.7	1
7	Congenital Hyperinsulinemia of Infancy: Role of Molecular Testing in Management and Genetic Counseling. Indian Journal of Pediatrics, 2022, 89, 395-398.	0.3	1
8	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
9	How Experts Make a Call: Copy Number Variation Analysis in Unusual/Rare Case Scenarios. Neurology India, 2022, 70, 148.	0.2	1
10	<scp> COASY </scp> related pontocerebellar hypoplasia type 12: A common Indian mutation with expansion of the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2022, , .	0.7	2
11	Deciphering the molecular landscape of microcephaly in 87 Indian families by exome sequencing. European Journal of Medical Genetics, 2022, 65, 104520.	0.7	2
12	Novel NLRP12 variant presenting with familial cold autoimmunity syndrome phenotype. Annals of the Rheumatic Diseases, 2021, 80, e117-e117.	0.5	9
13	Peters-Plus with Anal Atresia and a Novel Frameshift Mutation. Indian Journal of Pediatrics, 2021, 88, 184-185.	0.3	0
14	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
15	Koolenâ€de Vries syndrome: First report of two unrelated Indian patients. American Journal of Medical Genetics, Part A, 2021, 185, 982-985.	0.7	0
16	Clinical Sequencing Solves a Diagnostic Dilemma by Identifying a Novel Pathogenic Variant inÂUSB1ÂGene Causing Poikiloderma with Neutropenia. Indian Journal of Pediatrics, 2021, 88, 270-271.	0.3	1
17	Pearson Syndrome: Spontaneously Recovering Anemia and Hypoparathyroidism â€“ Correspondence. Indian Journal of Pediatrics, 2021, 88, 209-210.	0.3	0
18	Homozygous Missense Variation in <i>PNPLA8</i> Causes Prenatal-Onset Severe Neurodegeneration. Molecular Syndromology, 2021, 12, 174-178.	0.3	8

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19	Clinical and Mutation Spectra of Cockayne Syndrome in India. Neurology India, 2021, 69, 362.	0.2	4
20	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
21	Twins with PEX7 related intellectual disability and cataract: Highlighting phenotypes of peroxisome biogenesis disorder 9B. American Journal of Medical Genetics, Part A, 2021, 185, 1504-1508.	0.7	2
22	Homozygosity stretches around homozygous mutations in autosomal recessive disorders: patients from nonconsanguineous Indian families. Journal of Genetics, 2021, 100, 1.	0.4	8
23	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
24	Monogenic Lupus with IgA Nephropathy Caused by Spondyloenchondrodysplasia with Immune Dysregulation. Indian Journal of Pediatrics, 2021, 88, 819-823.	0.3	4
25	Partial Trisomy of Chromosome 8q and Partial Monosomy of Chromosome 6p with Robinow Syndrome-Like Phenotype. Indian Journal of Pediatrics, 2021, 88, 813-818.	0.3	0
26	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
27	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
28	Functional characterization of novel variants in SMPD1 in Indian patients with acid sphingomyelinase deficiency. Human Mutation, 2021, 42, 1336-1350.	1.1	6
29	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
30	Novel FOXP1 pathogenic variants in two Indian subjects with syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2021, 185, 1324-1327.	0.7	2
31	Kallmann Syndrome and X-linked Ichthyosis Caused by Translocation Between Chromosomes X and Y: A Case Report. Journal of Reproduction and Infertility, 2021, 22, 302-306.	1.0	2
32	Desbuquois dysplasia Kim variant: a rare case report syndrome. Clinical Dysmorphology, 2021, 30, 62-65.	0.1	0
33	Homozygosity stretches around homozygous mutations in autosomal recessive disorders: patients from nonconsanguineous Indian families. Journal of Genetics, 2021, 100, .	0.4	0
34	A mild phenotype of LGI4-Related arthrogryposis multiplex congenita with intrafamilial variability. European Journal of Medical Genetics, 2020, 63, 103756.	0.7	5
35	Renpenning syndrome in an Indian patient. American Journal of Medical Genetics, Part A, 2020, 182, 293-295.	0.7	2
36	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	0.7	15

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37	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
38	Untapped opportunities for rare disease gene discovery in India. American Journal of Medical Genetics, Part A, 2020, 182, 3056-3059.	0.7	2
39	Pearson Syndrome: Spontaneously Recovering Anemia and Hypoparathyroidism. Indian Journal of Pediatrics, 2020, 87, 1070-1072.	0.3	1
40	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
41	Distal Arthrogryposis: A Clue to the Etiology of Neonatal Cholestasis. Indian Journal of Pediatrics, 2020, 87, 869-870.	0.3	1
42	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
43	Hypotonic infant with Pallisterâ€“Killian syndrome diagnosed by cytogenetic microarray, without pigmentary skin changes and malformations. Journal of Genetics, 2020, 99, 1.	0.4	0
44	Hypotonic infant with Pallister-Killian syndrome diagnosed by cytogenetic microarray, without pigmentary skin changes and malformations. Journal of Genetics, 2020, 99, .	0.4	0
45	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
46	Vici Syndrome with a Novel Mutation in EPG5. Indian Pediatrics, 2019, 56, 603-605.	0.2	1
47	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
48	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
49	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
50	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
51	Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. American Journal of Human Genetics, 2019, 104, 439-453.	2.6	16
52	Computer-aided Facial Analysis in Diagnosing Dysmorphic Syndromes in Indian Children. Indian Pediatrics, 2019, 56, 1017-1019.	0.2	15
53	Expanding the phenotype in autosomal dominant mental retardation-24: a novel variation in DEAF1 gene. Clinical Dysmorphology, 2019, 28, 94-97.	0.1	4
54	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	0.7	40

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55	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
56	Concepts, Utility and Limitations of Cord Blood Banking: What Clinicians Need to Know. Indian Journal of Pediatrics, 2019, 86, 44-48.	0.3	6
57	Adrenoleukodystrophy: The Importance of Early MRI Findings and Serial Imaging. Neurology India, 2019, 67, 1559.	0.2	1
58	Cytogenetic microarray in structurally normal and abnormal foetuses: a five year experience elucidating increasing acceptance and clinical utility. Journal of Genetics, 2019, 98, .	0.4	2
59	Vici Syndrome with a Novel Mutation in EPG5. Indian Pediatrics, 2019, 56, 603-605.	0.2	0
60	Computer-aided Facial Analysis in Diagnosing Dysmorphic Syndromes in Indian Children. Indian Pediatrics, 2019, 56, 1017-1019.	0.2	4
61	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
62	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
63	Connexin 26 (GJB2) Mutations Associated with Non-Syndromic Hearing Loss (NSHL). Indian Journal of Pediatrics, 2018, 85, 1061-1066.	0.3	22
64	Phenotypic characterization of derivative 22 syndrome: case series and review. Journal of Genetics, 2018, 97, 205-211.	0.4	5
65	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
66	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
67	Socio-demographic Profile and Economic Burden of Treatment of Transfusion Dependent Thalassemia. Indian Journal of Pediatrics, 2018, 85, 102-107.	0.3	17
68	Malan syndrome: Extension of genotype and phenotype spectrum. American Journal of Medical Genetics, Part A, 2018, 176, 2896-2900.	0.7	4
69	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
70	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
71	Extending the phenotype and an ECEL1 gene mutation in distal arthrogryposis type 5D. Clinical Dysmorphology, 2018, 27, 130-134.	0.1	6
72	Gene expression profiling of coronary artery disease and its relation with different severities. Journal of Genetics, 2018, 97, 853-867.	0.4	2

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73	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
74	A large interstitial 11q deletion with isolated mild intellectual disability: review of the literature for genotypeâ€“phenotype correlation. Clinical Dysmorphology, 2018, 27, 142-144.	0.1	1
75	Spectrum of genomic variations in Indian patients with progressive familial intrahepatic cholestasis. BMC Gastroenterology, 2018, 18, 107.	0.8	9
76	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatrics, 2018, 55, 474-477.	0.2	3
77	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatrics, 2018, 55, 474-477.	0.2	1
78	Phenotypic characterization of derivative 22 syndrome: case series and review. Journal of Genetics, 2018, 97, 205-211.	0.4	2
79	Study of the association of forkhead box P3 () gene polymorphisms with unexplained recurrent spontaneous abortions in Indian population. Journal of Genetics, 2018, 97, 405-410.	0.4	3
80	Clinico-hematological Profile of Hb E-Î² Thalassemia-Prospective Analysis in a tertiary Care Centre. Journal of the Association of Physicians of India, The, 2018, 66, 42-45.	0.0	3
81	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
82	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
83	Cover Image, Volume 173A, Number 3, March 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
84	A novel variant in MED12 gene: Further delineation of phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 2257-2260.	0.7	4
85	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
86	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
87	Pyruvate Carboxylase Deficiency Mimicking Diabetic Ketoacidosis. Indian Journal of Pediatrics, 2017, 84, 959-960.	0.3	3
88	Hotspots in PTPN11 gene among Indian children with Noonan syndrome. Indian Pediatrics, 2017, 54, 638-640.	0.2	5
89	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
90	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	0.7	68

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91	KBG syndrome: 16q24.3 microdeletion in an Indian patient. Clinical Dysmorphology, 2017, 26, 161-166.	0.1	2
92	Expansion of the phenotypic spectrum in three families of methyl CpG-binding protein 2 duplication syndrome. Clinical Dysmorphology, 2017, 26, 73-77.	0.1	2
93	Double segment chromosomal imbalance due to inherited chromosomal translocation: Detection by cytogenetic microarray. Indian Pediatrics, 2017, 54, 879-881.	0.2	0
94	Fanconi-Bickel Syndrome: Another Novel Mutation in SLC2A2. Indian Journal of Pediatrics, 2017, 84, 236-237.	0.3	5
95	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
96	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
97	Prenatal screening for genetic disorders: Suggested guidelines for the Indian Scenario. Indian Journal of Medical Research, 2017, 146, 689.	0.4	21
98	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
99	Rheumatic Manifestations of Genetic Disorders and Hemophilia. , 2017, , 595-609.		0
100	Hyperekplexia: A forgotten diagnosis clinched by next-generation sequencing. Neurology India, 2017, 65, 1065.	0.2	4
101	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
102	Pycnodysostosis: mutation spectrum in five unrelated Indian children. Clinical Dysmorphology, 2016, 25, 113-120.	0.1	16
103	Floating Harbor Syndrome. Indian Journal of Pediatrics, 2016, 83, 896-897.	0.3	5
104	Complex chromosomal rearrangement involving five chromosomes. Clinical Dysmorphology, 2016, 25, 63-67.	0.1	0
105	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. American Journal of Medical Genetics, Part A, 2016, 170, 410-417.	0.7	31
106	Novel sequence variations in the thymidine phosphorylase gene causing mitochondrial neurogastrointestinal encephalopathy. Clinical Dysmorphology, 2016, 25, 156-162.	0.1	4
107	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
108	Worsening of callus hyperplasia after bisphosphonate treatment in type V osteogenesis imperfecta. Indian Pediatrics, 2016, 53, 250-252.	0.2	10

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109	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
110	Mutations in genes encoding condensin complex proteins cause microcephaly through decatenation failure at mitosis. Genes and Development, 2016, 30, 2158-2172.	2.7	106
111	Metatropic dysplasia with a novel mutation in TRPV4. Indian Pediatrics, 2016, 53, 735-737.	0.2	3
112	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
113	Unusual skin manifestations in a patient with menkes disease. American Journal of Medical Genetics, Part A, 2016, 170, 3039-3040.	0.7	1
114	Cartilage Hair Hypoplasia: Two Unrelated Cases with g.70 AÂ>ÂG Mutation in RMRP Gene. Indian Journal of Pediatrics, 2016, 83, 1003-1005.	0.3	2
115	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
116	Hunter syndrome in northern India: Clinical features and mutation spectrum. Indian Pediatrics, 2016, 53, 134-136.	0.2	3
117	Infantile Systemic Hyalinosis with Mutation in ANTXR2. Indian Journal of Pediatrics, 2016, 83, 1356-1357.	0.3	4
118	A Mutagenic Primer Assay for Genotyping of the CRHR1 Gene Rare Variant rs1876828 (A/G) in Asians: A Costâ€Effective SNP Typing. Journal of Clinical Laboratory Analysis, 2016, 30, 169-174.	0.9	0
119	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
120	Smith-Magenis Syndrome: Face Speaks. Indian Journal of Pediatrics, 2016, 83, 589-593.	0.3	5
121	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
122	Consanguinity as an Adjunct Diagnostic Tool. Indian Journal of Pediatrics, 2016, 83, 258-260.	0.3	3
123	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
124	Lysosomal storage disorders: Present and future. Indian Pediatrics, 2015, 52, 1025-1026.	0.2	1
125	Novel and recurrent mutations in WISP3 and an atypical phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 2481-2484.	0.7	21
126	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

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127	Chondrodysplasia punctata tibia metacarpal type. Clinical Dysmorphology, 2015, 24, 118-121.	0.1	4
128	Mutations in patients with osteogenesis imperfecta from consanguineous Indian families. European Journal of Medical Genetics, 2015, 58, 21-27.	0.7	37
129	Performance of QF-PCR in targeted prenatal aneuploidy diagnosis: Indian scenario. Gene, 2015, 562, 55-61.	1.0	8
130	Feasibility Study of an Outreach Program of Newborn Screening in Uttar Pradesh. Indian Journal of Pediatrics, 2015, 82, 427-432.	0.3	6
131	White matter changes in GM1 gangliosidosis. Indian Pediatrics, 2015, 52, 155-156.	0.2	11
132	Prenatal diagnosis in India is not limited to sex selection. Genetics in Medicine, 2015, 17, 88-88.	1.1	1
133	Recurrent and novel GLB1 mutations in India. Gene, 2015, 567, 173-181.	1.0	22
134	Medical genetics and genomic medicine in India: current status and opportunities ahead. Molecular Genetics & Genomic Medicine, 2015, 3, 160-171.	0.6	24
135	Symmetrical Terminal Transverse Limb Deficiencies. Indian Journal of Pediatrics, 2015, 82, 478-479.	0.3	2
136	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
137	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
138	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
139	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
140	Newborn screening for congenital hypothyroidism, galactosemia and biotinidase deficiency in Uttar Pradesh, India. Indian Pediatrics, 2014, 51, 701-705.	0.2	28
141	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
142	Distal arthrogryposis type 5D with a novel ECEL1 gene mutation. American Journal of Medical Genetics, Part A, 2014, 164, 2857-2862.	0.7	19
143	Severe short stature, profound microcephaly, developmental brain abnormality, agenesis of optic disc and retinal vessels, and bilateral cryptorchidism in two male siblings. Clinical Dysmorphology, 2014, 23, 117-120.	0.1	0
144	Fibrodysplasia Ossificans Progressiva: Three Indian Patients with Mutation in the ACVR1 Gene. Indian Journal of Pediatrics, 2014, 81, 617-619.	0.3	5

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145	A novel genotyping method for detection of the CRHR1 (rs1396862: C>T) gene variation among North Indian population. Molecular Biology Reports, 2014, 41, 2809-2813.	1.0	0
146	Cytogenetic microarray in prenatal and postnatal diagnosis. Molecular Cytogenetics, 2014, 7, I32.	0.4	3
147	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
148	Genetic insight of schizophrenia: past and future perspectives. Gene, 2014, 535, 97-100.	1.0	49
149	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
150	Platelet-specific collagen receptor glycoprotein VI gene variants affect recurrent pregnancy loss. Fertility and Sterility, 2014, 102, 1078-1084.e3.	0.5	10
151	GALNS mutations in Indian patients with mucopolysaccharidosis IVA. American Journal of Medical Genetics, Part A, 2014, 164, 2793-2801.	0.7	31
152	De novo SOX11 mutations cause Coffinâ€“Siris syndrome. Nature Communications, 2014, 5, 4011.	5.8	118
153	Research Letters. Indian Pediatrics, 2014, 51, 411-413.	0.2	4
154	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
155	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
156	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
157	STR Markers in clinics: a rapid prenatal diagnosis by quantitative fluorescent-pcr for aneuploidies. Molecular Cytogenetics, 2014, 7, I58.	0.4	1
158	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
159	S.S. Agarwal. The National Medical Journal of India, 2014, 27, 44-5.	0.1	0
160	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
161	Myotonic dystrophy type 1 (DM1): A triplet repeat expansion disorder. Gene, 2013, 522, 226-230.	1.0	30
162	Syndromic versus nonsyndromic atlantoaxial dislocation: do clinico-radiological differences have a bearing on management?. Acta Neurochirurgica, 2013, 155, 1157-1167.	0.9	20

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163	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	0.5	57
164	Genetic testing in children. Indian Pediatrics, 2013, 50, 823-827.	0.2	4
165	Utility of chromosomal microarray in five cases with cytogenetic abnormalities detected by traditional karyotype. Clinical Genetics, 2013, 84, 600-602.	1.0	2
166	Genetic variations of theFCER2gene and asthma susceptibility in north Indian children: a case-control study. Biomarkers, 2013, 18, 660-667.	0.9	4
167	Atlantoaxial dislocation in a child affected by warfarin embryopathy. Clinical Dysmorphology, 2013, 22, 124-126.	0.1	4
168	Neural tube defects: A need for population-based prevention program. Indian Journal of Human Genetics, 2012, 18, 145.	0.7	5
169	Multiplex Quantitative Fluorescent Polymerase Chain Reaction for Detection of Aneuploidies. Genetic Testing and Molecular Biomarkers, 2012, 16, 624-627.	0.3	12
170	Utility of MLPA in mutation analysis and carrier detection for Duchenne muscular dystrophy. Indian Journal of Human Genetics, 2012, 18, 91.	0.7	15
171	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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