Madhulika Kabra

List of Publications by Year
in descending order

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260	3,617	172457	243625
papers	citations	29 h-index	44 g-index

271	271	271	4757
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Poly (propyleneimine) dendrimer based nanocontainers for targeting of efavirenz to human monocytes/macrophagesin vitro. Journal of Drug Targeting, 2007, 15, 89-98.	4.4	145
2	Lead Encephalopathy in an Infant Mimicking a Neurometabolic Disorder. Journal of Child Neurology, 2010, 25, 390-392.	1.4	131
3	Idiopathic chronic pancreatitis in India: phenotypic characterisation and strong genetic susceptibility due to SPINK1 and CFTR gene mutations. Gut, 2010, 59, 800-807.	12.1	100
4	Hydroxyurea in thalassemia intermediaâ€"a promising therapy. Annals of Hematology, 2005, 84, 441-446.	1.8	93
5	Prevalence of the triple X syndrome in phenotypically normal women with premature ovarian failure and its association with autoimmune thyroid disorders. Fertility and Sterility, 2003, 80, 1052-1054.	1.0	92
6	Intravenous fluid regimen and hyponatraemia among children: a randomized controlled trial. Pediatric Nephrology, 2010, 25, 2303-2309.	1.7	79
7	Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86.	6.2	75
8	Association of SPINK1 Gene Mutation and CFTR Gene Polymorphisms in Patients With Pancreas Divisum Presenting With Idiopathic Pancreatitis. Journal of Clinical Gastroenterology, 2009, 43, 848-852.	2.2	72
9	Intranasal versus intravenous lorazepam for control of acute seizures in children: A randomized openâ€label study. Epilepsia, 2011, 52, 788-793.	5.1	72
10	Efficacy of 4:1 (classic) versus 2.5:1 ketogenic ratio diets in refractory epilepsy in young children: A randomized open labeled study. Epilepsy Research, 2011, 96, 96-100.	1.6	65
11	Incidence of acute kidney injury in hospitalized children. Indian Pediatrics, 2012, 49, 537-542.	0.4	59
12	Screening of families with autosomal recessive non-syndromic hearing impairment (ARNSHI) for mutations in GJB2 gene: Indian scenario. American Journal of Medical Genetics Part A, 2003, 120A, 180-184.	2.4	55
13	Efficacy of modified constraint induced movement therapy in improving upper limb function in children with hemiplegic cerebral palsy: A randomized controlled trial. Brain and Development, 2013, 35, 870-876.	1.1	51
14	Seizure control and biochemical profile on the ketogenic diet in young children with refractory epilepsyâ€"Indian experience. Seizure: the Journal of the British Epilepsy Association, 2009, 18, 446-449.	2.0	50
15	Diagnosis and Management of Down Syndrome. Indian Journal of Pediatrics, 2014, 81, 560-567.	0.8	41
16	Sequential Occurrence of Preneoplastic Lesions and Accumulation of Loss of Heterozygosity in Patients With Gallbladder Stones Suggest Causal Association With Gallbladder Cancer. Annals of Surgery, 2014, 260, 1073-1080.	4.2	40
17	Genetically Determined Chronic Pancreatitis but not Alcoholic Pancreatitis Is a Strong Risk Factor for Pancreatic Cancer. Pancreas, 2016, 45, 1478-1484.	1.1	40
18	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40

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19	Velaglucerase alfa (VPRIV) enzyme replacement therapy in patients with Gaucher disease: Longâ€term data from phase III clinical trials. American Journal of Hematology, 2015, 90, 584-591.	4.1	39
20	Carrier frequency of F508del mutation of cystic fibrosis in Indian population. Journal of Cystic Fibrosis, 2006, 5, 43-46.	0.7	37
21	Clinical profile and frequency of delta f508 mutation in Indian children with cystic fibrosis. Indian Pediatrics, 2003, 40, 612-9.	0.4	37
22	Is the spectrum of mutations in Indian patients with cystic fibrosis different?. American Journal of Medical Genetics Part A, 2000, 93, 161-163.	2.4	36
23	Targeted Deep Resequencing IdentifiesMID2Mutation for X-Linked Intellectual Disability with Varied Disease Severity in a Large Kindred from India. Human Mutation, 2014, 35, 41-44.	2.5	36
24	Live births in women with recurrent hydatidiform mole and two NLRP7 mutations. Reproductive BioMedicine Online, 2015, 31, 120-124.	2.4	36
25	Association of polymorphism in the thermolabile 5, 10-methylene tetrahydrofolate reductase gene and hyperhomocysteinemia with coronary artery disease. Molecular and Cellular Biochemistry, 2008, 310, 111-117.	3.1	34
26	Characterisation of mutations and genotypeâ€“phenotype correlation in cystic fibrosis: Experience from India. Journal of Cystic Fibrosis, 2008, 7, 110-115.	0.7	34
27	Griscelli syndrome. Journal of the American Academy of Dermatology, 2006, 55, 337-340.	1.2	33
28	Long-term daily high and low doses of azithromycin in children with cystic fibrosis: A randomized controlled trial. Journal of Cystic Fibrosis, 2010, 9, 17-23.	0.7	32
29	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. American Journal of Medical Genetics, Part A, 2016, 170, 410-417.	1.2	31
30	The impact of COVID-19 pandemic on the diagnosis and management of inborn errors of metabolism: A global perspective. Molecular Genetics and Metabolism, 2020, 131, 285-288.	1.1	31
31	Hypocalcemic heart failure masquerading as dilated cardiomyopathy. Indian Journal of Pediatrics, 2001, 68, 287-290.	0.8	30
32	Undergoing prenatal screening for Down's syndrome: presentation of choice and information in Europe and Asia. European Journal of Human Genetics, 2007, 15, 563-569.	2.8	30
33	Detection of fetomaternal hemorrhage following chorionic villus sampling by Kleihauer Betke test and rise in maternal serum alpha feto protein. Prenatal Diagnosis, 2007, 27, 139-142.	2.3	29
34	Newborn screening in India: Current perspectives. Indian Pediatrics, 2010, 47, 219-224.	0.4	29
35	Prevalence of Sleep Abnormalities in Indian Children With Autism Spectrum Disorder: A Cross-Sectional Study. Pediatric Neurology, 2017, 74, 62-67.	2.1	29
36	Prevalence of MTHFR C677T polymorphism in north Indian mothers having babies with Trisomy 21 Down syndrome. Down Syndrome Research and Practice, 2008, 12, 133-137.	0.3	29

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37	Mosaic 22q11.2 microdeletion syndrome: diagnosis and clinical manifestations of two cases. Molecular Cytogenetics, 2008, 1, 18.	0.9	27
38	Dietary management of inborn errors of metabolism. Indian Journal of Pediatrics, 2002, 69, 421-426.	0.8	25
39	Alpha 1 antitrypsin deficiency in children with chronic liver disease in North India. Indian Pediatrics, 2010, 47, 1015-1023.	0.4	25
40	Glutaric Acidemia Type 1-Clinico-Molecular Profile and Novel Mutations in GCDH Gene in Indian Patients. JIMD Reports, 2014, 21, 45-55.	1.5	25
41	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.	2.5	25
42	Poland anomaly with unusual associated anomalies: Case report of an apparent disorganization defect. American Journal of Medical Genetics Part A, 1994, 52, 402-405.	2.4	24
43	Active surveillance for intussusception in a phase III efficacy trial of an oral monovalent rotavirus vaccine in India. Vaccine, 2014, 32, A104-A109.	3.8	24
44	Mutation spectrum of COL1A1 and COL1A2 genes in Indian patients with osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2014, 164, 1482-1489.	1.2	24
45	Asparagine Synthetase deficiency-report of a novel mutation and review of literature. Metabolic Brain Disease, 2017, 32, 1889-1900.	2.9	24
46	Cardiovascular Autonomic Dysfunction in Children and Adolescents With Rett Syndrome. Pediatric Neurology, 2017, 70, 61-66.	2.1	23
47	Influence of MDR1 and CYP3A5 genetic polymorphisms on trough levels and therapeutic response of imatinib in newly diagnosed patients with chronic myeloid leukemia. Pharmacological Research, 2017, 120, 138-145.	7.1	23
48	Intravenous Pamidronate Therapy in Osteogenesis Imperfecta. Journal of Pediatric Orthopaedics, 2007, 27, 225-227.	1.2	22
49	VitaminÂA responsive night blindness in Dentâ€™s disease. Pediatric Nephrology, 2009, 24, 1765-1770.	1.7	22
50	Severe neuronopathic autosomal recessive osteopetrosis due to homozygous deletions affecting OSTM1. Bone, 2013, 55, 292-297.	2.9	22
51	Recurrent and novel GLB1 mutations in India. Gene, 2015, 567, 173-181.	2.2	22
52	Clinical and Molecular Disease Spectrum and Outcomes in Patients with Infantile-Onset Pompe Disease. Journal of Pediatrics, 2020, 216, 44-50.e5.	1.8	22
53	Molecular and structural analysis of metachromatic leukodystrophy patients in Indian population. Journal of the Neurological Sciences, 2011, 301, 38-45.	0.6	21
54	Neurodevelopmental and epilepsy outcome in children aged one to five years with infantile spasmsâ€”A North Indian cohort. Epilepsy Research, 2014, 108, 526-534.	1.6	21

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55	Carbimazole embryopathyâ€bilateral choanal atresia and patent vitelloâ€intestinal duct: A case report and review of literature. Birth Defects Research Part A: Clinical and Molecular Teratology, 2008, 82, 649-652.	1.6	20
56	Intellectual disability in Indian children: experience with a stratified approach for etiological diagnosis. Indian Pediatrics, 2013, 50, 1125-1130.	0.4	20
57	Effects of Exercise Intervention Program on Bone Mineral Accretion in Children and Adolescents with Cystic Fibrosis: A Randomized Controlled Trial. Indian Journal of Pediatrics, 2019, 86, 987-994.	0.8	20
58	Raine syndrome: a clinical, radiographic and genetic investigation of a case from the Indian subcontinent. Clinical Dysmorphology, 2010, 19, 153-156.	0.3	19
59	National newborn screening program â€" Still a hype or a hope now?. Indian Pediatrics, 2013, 50, 639-643.	0.4	19
60	Molecular Diagnosis of Hereditary Fructose Intolerance: Founder Mutation in a Community from India. JIMD Reports, 2014, 19, 85-93.	1.5	18
61	Cystic Fibrosis Presenting as Pseudo-Bartter Syndrome: An Important Diagnosis that is Missed!. Indian Journal of Pediatrics, 2020, 87, 726-732.	0.8	18
62	Prevalence of 22q11.2 microdeletion in 146 patients with cardiac malformation in a referral hospital of North India. BMC Medical Genetics, 2010, 11, 101.	2.1	17
63	Novel Genetic, Clinical, and Pathomechanistic Insights into TFG-Associated Hereditary Spastic Paraplegia. Human Mutation, 2016, 37, 1157-1161.	2.5	17
64	Natural history of non-lethal Raine syndrome during childhood. Orphanet Journal of Rare Diseases, 2020, 15, 93.	2.7	17
65	Peripheral neuropathy in cystic fibrosis: A prevalence study. Journal of Cystic Fibrosis, 2013, 12, 754-760.	0.7	16
66	Prevalence of UGT1A6 polymorphisms in children with epilepsy on valproate monotherapy. Neurology India, 2015, 63, 35.	0.4	16
67	Pycnodysostosis: mutation spectrum in five unrelated Indian children. Clinical Dysmorphology, 2016, 25, 113-120.	0.3	16
68	Application of whole exome sequencing in elucidating the phenotype and genotype spectrum of junctional epidermolysis bullosa: A preliminary experience of a tertiary care centre in India. Journal of Dermatological Science, 2017, 86, 30-36.	1.9	16
69	Association of Sleep Apnea With Development and Behavior in Down Syndrome: A Prospective Clinical and Polysomnographic Study. Pediatric Neurology, 2021, 116, 7-13.	2.1	16
70	Mental retardation. Indian Journal of Pediatrics, 2003, 70, 153-158.	0.8	15
71	Schwartz Jampel syndrome in children. Journal of Clinical Neuroscience, 2013, 20, 313-317.	1.5	15
72	Frequencies of CYP2C9 polymorphisms in North Indian population and their association with drug levels in children on phenytoin monotherapy. BMC Pediatrics, 2016, 16, 66.	1.7	15

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73	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.	1.2	15
74	Spectrum of ARSA variations in Asian Indian patients with Arylsulfatase A deficient metachromatic leukodystrophy. Journal of Human Genetics, 2019, 64, 323-331.	2.3	15
75	Pathogenic/likely pathogenic variants in the SHOX, GHR and IGFALS genes among Indian children with idiopathic short stature. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 79-88.	0.9	15
76	Group B Meningococcal Meningitis in India. Scandinavian Journal of Infectious Diseases, 1994, 26, 771-773.	1.5	14
77	Clinical profile and mutation analysis of xeroderma pigmentosum in Indian patients. Indian Journal of Dermatology, Venereology and Leprology, 2015, 81, 16.	0.6	14
78	Whole exome sequencing identifies a homozygous nonsense variation in ALMS1 gene in a patient with syndromic obesity. Obesity Research and Clinical Practice, 2017, 11, 241-246.	1.8	14
79	Prevalence of Celiac Disease in Indian Children with Down Syndrome and its Clinical and Laboratory Predictors. Indian Journal of Pediatrics, 2013, 80, 114-117.	0.8	13
80	Noninvasive screening for preclinical atherosclerosis in children on phenytoin or carbamazepine monotherapy: A cross sectional study. Epilepsy Research, 2013, 107, 121-126.	1.6	13
81	Adverse pregnancy outcome in patients with low pregnancyâ€associated plasma proteinâ€<scp>A</scp>: The <scp>I</scp>ndian <scp>E</scp>xperience. Journal of Obstetrics and Gynaecology Research, 2015, 41, 1003-1008.	1.3	13
82	Imatinib trough levels: a potential biomarker to predict cytogenetic and molecular response in newly diagnosed patients with chronic myeloid leukemia. Leukemia and Lymphoma, 2019, 60, 418-425.	1.3	13
83	Genetic polymorphisms associated with obesity and non-alcoholic fatty liver disease in Asian Indian adolescents. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 749-758.	0.9	13
84	The spectrum of leukodystrophies in children: Experience at a tertiary care centre from North India. Annals of Indian Academy of Neurology, 2016, 19, 332.	0.5	13
85	Frequency of primary mutations of Leber's hereditary optic neuropathy patients in North Indian population. Indian Journal of Ophthalmology, 2017, 65, 1156.	1.1	13
86	Pelvic radiograph in skeletal dysplasias: An approach. Indian Journal of Radiology and Imaging, 2017, 27, 187-199.	0.8	13
87	Hypothalamic hamartoma, gelastic epilepsy, precocious puberty â€“ a diffuse cerebral dysgenesis. Brain and Development, 2002, 24, 784-786.	1.1	12
88	Approach to inborn errors of metabolism presenting in the neonate. Indian Journal of Pediatrics, 2008, 75, 271-276.	0.8	12
89	Seventeen Novel Mutations in PCCA and PCCB Genes in Indian Propionic Acidemia Patients, and Their Outcomes. Genetic Testing and Molecular Biomarkers, 2016, 20, 373-382.	0.7	12
90	Congenital Cytomegalovirus Infection and Permanent Hearing Loss in Rural North Indian Children. Pediatric Infectious Disease Journal, 2017, 36, 670-673.	2.0	12

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91	Blepharophimosis, telecanthus, microstomia, and unusual ear anomaly (Simosa syndrome) in an infant. American Journal of Medical Genetics Part A, 1994, 51, 222-223.	2.4	11
92	Thrombocytopenic purpura as a presenting manifestation of tubercular lymphadenitis. Indian Journal of Pediatrics, 2003, 70, 993-994.	0.8	11
93	Prenatal diagnosis for a novel homozygous mutation inPKLR gene in an Indian family. Prenatal Diagnosis, 2007, 27, 117-118.	2.3	11
94	Use of HbA estimation by CE-HPLC for prenatal diagnosis of^{2}-thalassemia; experience from a tertiary care centre in north India: a brief report. Hematology, 2009, 14, 122-124.	1.5	11
95	Behavioral comorbidity in children and adolescents with epilepsy. Journal of Clinical Neuroscience, 2014, 21, 1337-1340.	1.5	11
96	Profile of prothrombotic factors in Indian children with ischemic stroke. Journal of Clinical Neuroscience, 2014, 21, 1315-1318.	1.5	11
97	Ghosal type hematodiaphyseal dysplasia. Indian Pediatrics, 2016, 53, 347-348.	0.4	11
98	Application of chromosomal microarrays in the evaluation of intellectual disability/global developmental delay patients â€“ A study from a tertiary care genetic centre in India. Gene, 2016, 590, 109-119.	2.2	11
99	Chanarin Dorfman syndrome: a case report with novel nonsense mutation. Gene, 2016, 575, 359-362.	2.2	11
100	A novel homozygous mutation in POLR3A gene causing 4H syndrome: a case report. BMC Pediatrics, 2018, 18, 126.	1.7	11
101	Fraser-Cryptophthalmos syndrome. Indian Journal of Pediatrics, 2000, 67, 775-778.	0.8	10
102	Biotinidase deficiencyâ€”A treatable entity. Indian Journal of Pediatrics, 2000, 67, 464-466.	0.8	10
103	Schinzel Acrocallosal syndrome. Indian Journal of Pediatrics, 2003, 70, 173-176.	0.8	10
104	Genetics of deafness in India. Indian Journal of Pediatrics, 2004, 71, 531-533.	0.8	10
105	Establishing national neonatal perinatal database and birth defects registry network â€” Need of the hour!. Indian Pediatrics, 2014, 51, 693-696.	0.4	10
106	Encephalocraniocutaneous Lipomatosis With Neurocutaneous Melanosis. Journal of Child Neurology, 2014, 29, 846-849.	1.4	10
107	Batten disease: biochemical and molecular characterization revealing novel PPT1 and TPP1 gene mutations in Indian patients. BMC Neurology, 2018, 18, 203.	1.8	10
108	Newborn Screening and Diagnosis of Infants with Congenital Adrenal Hyperplasia. Indian Pediatrics, 2020, 57, 49-55.	0.4	10

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109	Menkes disease â€“ An important cause of early onset refractory seizures. Journal of Pediatric Neurosciences, 2014, 9, 11.	0.3	10
110	Spondylo-megaepiphyseal-metaphyseal dysplasia: an unusual bone dysplasia. Pediatric Radiology, 2003, 33, 893-896.	2.0	9
111	Juvenile rheumatoid arthritis with myelofibrosis with myeloid metaplasia. Indian Journal of Pediatrics, 2005, 72, 789-791.	0.8	9
112	Multiplex PCR for rapid detection of exonal deletions in patients of duchenne muscular dystrophy. Indian Journal of Clinical Biochemistry, 2006, 21, 147-151.	1.9	9
113	Aquagenic Wrinkling of Skin: A Screening Test for Cystic Fibrosis. Indian Pediatrics, 2019, 56, 109-113.	0.4	9
114	Identification of a case of SRD5A3-congenital disorder of glycosylation (CDG1Q) by exome sequencing. Indian Journal of Medical Research, 2018, 147, 422.	1.0	9
115	Optimal care for children with down syndrome in India. Indian Journal of Pediatrics, 1996, 63, 121-126.	0.8	8
116	Infantile-onset leukoencephalopathy with discrepant mild clinical course. Indian Journal of Pediatrics, 2000, 67, 769-773.	0.8	8
117	Three Novel Variants in X-linked Adrenoleukodystrophy. Journal of Child Neurology, 2009, 24, 857-860.	1.4	8
118	Molecular Genetic Studies in Indian Patients With Megalencephalic Leukoencephalopathy. Pediatric Neurology, 2011, 44, 450-458.	2.1	8
119	Mutation analysis of Indian patients with urea cycle defects. Indian Pediatrics, 2012, 49, 585-586.	0.4	8
120	Prenatal Diagnosis of Fetal Petersâ€™ Plus Syndrome: A Case Report. Case Reports in Genetics, 2013, 2013, 1-3.	0.2	8
121	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.	1.1	8
122	ADRB2 polymorphism and salbutamol responsiveness in Northern Indian children with mild to moderate exacerbation of asthma. Indian Pediatrics, 2016, 53, 211-215.	0.4	8
123	Clinical profile and treatment status of subjects with cleft lip and palate anomaly in India: Preliminary report of a three-center study. Journal of Cleft Lip Palate and Craniofacial Anomalies, 2014, 1, 26.	0.0	8
124	Transethnic analysis of psoriasis susceptibility in South Asians and Europeans enhances fine mapping in the MHC and genome wide. Human Genetics and Genomics Advances, 2022, 3, 100069.	1.7	8
125	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.	0.4	8
126	Factor IX gene polymorphisms in Indian population. American Journal of Hematology, 2001, 68, 246-248.	4.1	7

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127	Wiedemannâ€“Rautenstrauch Syndrome: First Indian Case. Indian Journal of Pediatrics, 2011, 78, 1552-1555.	0.8	7
128	Imaging in Neonatal Maple Syrup Urine Disease. Indian Journal of Pediatrics, 2013, 80, 87-88.	0.8	7
129	Macrocephaly with Diffuse White Matter Changes Simulating a Leukodystrophy in Menkes Disease. Indian Journal of Pediatrics, 2013, 80, 160-162.	0.8	7
130	Status of iodine deficiency disorder in district Udham Singh Nagar, Uttarakhand state India. Indian Journal of Endocrinology and Metabolism, 2014, 18, 419.	0.4	7
131	Leukodystrophy Presenting as Acute-Onset Polyradiculoneuropathy. Pediatric Neurology, 2014, 50, 616-618.	2.1	7
132	Bone mineral density of Indian children and adolescents with cystic fibrosis. Indian Pediatrics, 2017, 54, 545-549.	0.4	7
133	Growth Pattern and Clinical Profile of Indian Children with Classical 21-Hydroxylase Deficiency Congenital Adrenal Hyperplasia on Treatment. Indian Journal of Pediatrics, 2019, 86, 496-502.	0.8	7
134	Rapid Eye Movement (REM) Sleep Behavior Disorder and REM Sleep with Atonia in the Young. Canadian Journal of Neurological Sciences, 2020, 47, 100-108.	0.5	7
135	Utility of fetal whole exome sequencing in the etiological evaluation and outcome of nonimmune hydrops fetalis. Prenatal Diagnosis, 2021, 41, 1414-1424.	2.3	7
136	Fragile X screening for FRAXA and FRAXE mutations using PCR based studies: Results of a five year study. Indian Journal of Human Genetics, 2006, 12, 17.	0.7	7
137	Cystic fibrosis-an Indian perspective on recent advances in diagnosis and management. Indian Journal of Pediatrics, 1996, 63, 189-198.	0.8	6
138	Detection of complex chromosomal rearrangements in a woman with repeated spontaneous abortions. Acta Obstetricia Et Gynecologica Scandinavica, 2001, 80, 478-479.	2.8	6
139	Prenatal diagnosis of megalencephalic leukodystrophy. Prenatal Diagnosis, 2008, 28, 357-359.	2.3	6
140	The mutation spectrum in Indian patients with Gaucher disease. Genome Biology, 2011, 12, .	9.6	6
141	Molecular analysis of ABCD1 gene in Indian patients with X-linked Adrenoleukodystrophy. Clinica Chimica Acta, 2011, 412, 2289-2295.	1.1	6
142	Acute Management of Sick Infants with Suspected Inborn Errors of Metabolism. Indian Journal of Pediatrics, 2011, 78, 854-859.	0.8	6
143	De novo deletion in MECP2 in a monozygotic twin pair: a case report. BMC Medical Genetics, 2011, 12, 113.	2.1	6
144	Report of Another Mutation Proven Case of Carbonic Anhydrase II Deficiency. Journal of Pediatric Genetics, 2019, 08, 091-094.	0.7	6

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145	First report of THOC6 related intellectual disability (Beaulieu Boycott Innes syndrome) in two siblings from India. European Journal of Medical Genetics, 2020, 63, 103742.	1.3	6
146	Spectrum of amyloglucosidase mutations in Asian Indian patients with Glycogen storage disease type III. American Journal of Medical Genetics, Part A, 2020, 182, 1190-1200.	1.2	6
147	Late onset Pompe Disease in India â€“ Beyond the Caucasian phenotype. Neuromuscular Disorders, 2021, 31, 431-441.	0.6	6
148	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.	1.3	6
149	Functional characterization of novel variants in SMPD1 in Indian patients with acid sphingomyelinase deficiency. Human Mutation, 2021, 42, 1336-1350.	2.5	6
150	Identification of GJB6 gene mutation in an Indian man with Clouston syndrome. Indian Journal of Dermatology, Venereology and Leprology, 2016, 82, 697.	0.6	6
151	Cystic fibrosis in India. The National Medical Journal of India, 2003, 16, 291-3.	0.3	6
152	Nephrogenic diabetes insipidus presenting with developmental delay and intracranial calcification. Indian Journal of Pediatrics, 2005, 72, 527-528.	0.8	5
153	A female with hemihypertrophy and chylous ascites â€“ Klippelâ€“Trenaunay syndrome or Proteus syndrome: a diagnostic dilemma. Clinical Dysmorphology, 2006, 15, 229-231.	0.3	5
154	Informed choice to undergo prenatal screening for thalassemia: a description of written information given to pregnant women in Europe and beyond. Prenatal Diagnosis, 2008, 28, 727-734.	2.3	5
155	Rapid Detection of Deletions in Hotspot C-Terminal Segment Region of MECP2 by Routine PCR Method: Report of Two Classical Rett Syndrome Patients of Indian Origin. Genetic Testing and Molecular Biomarkers, 2009, 13, 277-280.	0.7	5
156	Antenatal diagnosis of Lowe syndrome. Clinical and Experimental Nephrology, 2010, 14, 296-297.	1.6	5
157	Distinct De Novo deletions in a brotherâ€“sister pair with RTT: A case report. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 859-863.	1.7	5
158	Comparison of heart rate variability among children with well controlled versus refractory epilepsy: A cross-sectional study. Epilepsy Research, 2012, 101, 88-91.	1.6	5
159	Norrie Disease: First Mutation Report and Prenatal Diagnosis in an Indian Family. Indian Journal of Pediatrics, 2012, 79, 1529-1531.	0.8	5
160	Dystrophinopathy Diagnosis Made Easy. Journal of Child Neurology, 2014, 29, 469-474.	1.4	5
161	Skin Biopsy. Journal of Child Neurology, 2014, 29, NP5-NP8.	1.4	5
162	Prenatal screening: Perspective for the pediatrician. Indian Pediatrics, 2014, 51, 959-962.	0.4	5

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163	Smith-Magenis Syndrome: Face Speaks. Indian Journal of Pediatrics, 2016, 83, 589-593.	0.8	5
164	Do polymorphisms inMDR1andCYP3A5genes influence the risk of cytogenetic relapse in patients with chronic myeloid leukemia on imatinib therapy?. Leukemia and Lymphoma, 2017, 58, 2218-2226.	1.3	5
165	Biâ€allelic lossâ€ofâ€function novel variants in <scp>LTBP3</scp>â€related skeletal dysplasia: Report of first patient from India. American Journal of Medical Genetics, Part A, 2020, 182, 1944-1946.	1.2	5
166	Management of Infants with Congenital Adrenal Hyperplasia. Indian Pediatrics, 2020, 57, 159-164.	0.4	5
167	Hydrops fetalis in PKD1L1â€related heterotaxy: Report of two foetuses and expanding the phenotypic and molecular spectrum. Annals of Human Genetics, 2021, 85, 138-145.	0.8	5
168	Weak Ligaments and Sloping Joints: A New Hypothesis for Development of Congenital Atlantoaxial Dislocation and Basilar Invagination. Neurospine, 2020, 17, 843-856.	2.9	5
169	Novel non-identical MECP2 mutations in Rett syndrome family: A rare presentation. Brain and Development, 2012, 34, 28-31.	1.1	4
170	Iodine nutrition status amongst neonates in Kangra district, Himachal Pradesh. Journal of Trace Elements in Medicine and Biology, 2014, 28, 351-353.	3.0	4
171	At Least an Infantogram if not Perinatal Autopsy. Journal of Fetal Medicine, 2014, 01, 33-39.	0.1	4
172	Acrodermatitis Dysmetabolica - Report of Two Cases. Indian Journal of Pediatrics, 2015, 82, 869-870.	0.8	4
173	Spondylometaphyseal Dysplasia Corner Fracture (Sutcliffe) Type. Indian Journal of Pediatrics, 2016, 83, 1191-1194.	0.8	4
174	Prognostic Utility of Clinical Epilepsy Severity Score Versus Pretreatment Hypsarrhythmia Scoring in Children With West Syndrome. Clinical EEG and Neuroscience, 2017, 48, 280-287.	1.7	4
175	Decoding of novel missense TSC2 gene variants using in-silico methods. BMC Medical Genetics, 2019, 20, 164.	2.1	4
176	Enhanced Reprogramming Efficiency and Kinetics of Induced Pluripotent Stem Cells Derived from Human Duchenne Muscular Dystrophy. PLOS Currents, 2015, 7, .	1.4	4
177	Prenatal diagnosis of steroid 21-hydroxylase-deficient congenital adrenal hyperplasia: Experience from a tertiary care centre in India. Indian Journal of Medical Research, 2017, 145, 194-202.	1.0	4
178	Spectrum of GJB2 gene variants in Indian children with non-syndromic hearing loss. Indian Journal of Medical Research, 2018, 147, 615.	1.0	4
179	Impact of parental origin of X-chromosome on clinical and biochemical profile in Turner syndrome. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1155-1163.	0.9	4
180	Preserved umbilical cord facilitates antenatal diagnosis of spinal muscular atrophy. Indian Pediatrics, 2003, 40, 415-8.	0.4	4

#	Article	IF	Citations
181	Mitochondrial myopathy presenting as ataxia with dilated cardiomyopathy. Indian Journal of Pediatrics, 2001, 68, 347-350.	0.8	3
182	Recurrent pulmonary thromboembolism in a patient with heterozygous ?-thalassemia with Hb E state: A rare complication. American Journal of Hematology, 2003, 74, 85-85.	4.1	3
183	Intermittent hyperammonemic encephalopathy in a child with ornithine transcarbamylase deficiency. Indian Journal of Pediatrics, 2004, 71, 645-647.	0.8	3
184	Megalencephalic leukodystrophy with simple hemihyperplasia: a rare association. Clinical Dysmorphology, 2009, 18, 49-51.	0.3	3
185	Hyperekplexia Masquerading as Epilepsy. Indian Journal of Pediatrics, 2011, 78, 757-757.	0.8	3
186	Brachytelephalangic chondrodysplasia punctata. Clinical Dysmorphology, 2012, 21, 113-117.	0.3	3
187	Coping Strategies of Parents of Down Syndrome Children in India. Indian Journal of Pediatrics, 2013, 80, 534-535.	0.8	3
188	Increase in Iodine Deficiency Disorder due to Inadequate Sustainability of Supply of Iodized Salt in District Solan, Himachal Pradesh. Journal of Tropical Pediatrics, 2013, 59, 514-515.	1.5	3
189	Disseminated cryptococcosis. Indian Pediatrics, 2014, 51, 225-226.	0.4	3
190	Genetic Studies in Autism. Indian Journal of Pediatrics, 2016, 83, 1133-1140.	0.8	3
191	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatrics, 2018, 55, 474-477.	0.4	3
192	Report of Two Novel Mutations in Indian Patients with Rothmundâ€“Thomson Syndrome. Journal of Pediatric Genetics, 2019, 08, 163-167.	0.7	3
193	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.	2.3	3
194	â€˜Go for it, dream big, work hard and persistâ€™: A message to the next generation of CF leaders in recognition of International Women's Day 2020. Journal of Cystic Fibrosis, 2020, 19, 184-193.	0.7	3
195	Inherited 5p deletion syndrome due to paternal balanced translocation: Phenotypic heterogeneity due to duplication of 8q and 12p. Journal of Pediatric Genetics, 2013, 2, 163-9.	0.7	3
196	Identification of a novel homozygous mutation in transmembrane channel like 1 () gene, one of the second-tier hearing loss genes after in India. Indian Journal of Medical Research, 2017, 145, 492-497.	1.0	3
197	Genotype-phenotype spectrum of 130 unrelated Indian families with Mucopolysaccharidosis type II. European Journal of Medical Genetics, 2022, 65, 104447.	1.3	3
198	Detection of 22 q11.2 hemizygous deletion by interphase FISH in a patient with features of CATCH 22 syndrome. Indian Pediatrics, 2005, 42, 1236-9.	0.4	3

#	Article	IF	Citations
199	Newborn Screening and Diagnosis of Infants with Congenital Adrenal Hyperplasia. Indian Pediatrics, 2020, 57, 49-55.	0.4	3
200	Management of Wiskott-Aldrich syndrome. Indian Journal of Pediatrics, 1996, 63, 709-712.	0.8	2
201	Juvenile neuronal ceroid lipofuscinosis. Indian Journal of Pediatrics, 2000, 67, 689-691.	0.8	2
202	Prenatal diagnosis. Indian Journal of Pediatrics, 2003, 70, 81-85.	0.8	2
203	Inflicted neuro-trauma in infancy. Indian Journal of Pediatrics, 2010, 77, 318-320.	0.8	2
204	Familial Progressive Hypermelanosis in Indian Monozygotic Twins. Pediatric Dermatology, 2011, 28, 62-65.	0.9	2
205	Limb/Pelvis-Hypoplasia/Aplasia Syndrome - Further Delineation of Phenotype. Fetal and Pediatric Pathology, 2011, 30, 355-358.	0.7	2
206	Iodine Nutritional Status Among Neonates in the Solan District, Himachal Pradesh, India. Journal of Community Health, 2014, 39, 987-989.	3.8	2
207	Atypical late presentation in neonatal-onset multisystem inflammatory disease (NOMID). Journal of Pediatric Neurology, 2015, 07, 301-305.	0.2	2
208	Bilateral fronto-parietal polymicrogyria in an Indian infant. Journal of Pediatric Neurology, 2015, 09, 251-253.	0.2	2
209	Caffeyâ€™s Disease: Two Cases Presenting with Unexplained Fever. Indian Journal of Pediatrics, 2016, 83, 1499-1500.	0.8	2
210	Report of an Indian Family with Sengers Syndrome. Indian Journal of Pediatrics, 2021, 88, 92-92.	0.8	2
211	Spectrum of Movement Disorders of Late-Onset Niemann-Pick Disease Type C. Canadian Journal of Neurological Sciences, 2022, 49, 804-808.	0.5	2
212	Methylene Tetrahydrofolate Reductase Deficiency. Indian Journal of Pediatrics, 2020, 87, 951-953.	0.8	2
213	Fluorescence in situ hybridization (FISH) using non-commercial probes in the diagnosis of clinically suspected microdeletion syndromes. Indian Journal of Medical Research, 2013, 138, 135-42.	1.0	2
214	Management of Infants with Congenital Adrenal Hyperplasia. Indian Pediatrics, 2020, 57, 159-164.	0.4	2
215	I-cell disease (Mucolipidosis II). Indian Journal of Pediatrics, 2000, 67, 683-687.	0.8	1
216	Congenital adrenal hyperplasia presenting as hematuria and acute renal faliure. Indian Journal of Pediatrics, 2001, 68, 1161-1162.	0.8	1

#	Article	IF	Citations
217	Production of dysplastic platelets by peripheral blood megakaryoblasts in transient myeloproliferative disorder in Down syndrome. Platelets, 2008, 19, 160-161.	2.3	1
218	Prenatal diagnosis in India is not limited to sex selection. Genetics in Medicine, 2015, 17, 88-88.	2.4	1
219	Editorial: New Horizons in Genetic Diagnosis in Pediatric Practice: The Excitement and Challenges!. Indian Journal of Pediatrics, 2016, 83, 1131-1132.	0.8	1
220	Editorial. Indian Pediatrics, 2016, 53, 19-26.	0.4	1
221	Report of a Novel Homozygous Nonsense DDR2 Mutation in an Indian Adult Male with Spondylo-meta-epiphyseal Dysplasia, Short Limb-Abnormal Calcification Type. Journal of Pediatric Genetics, 2019, 08, 153-156.	0.7	1
222	Thenar Hypertrophy and Electrical Myotonia in Pompe Disease. Journal of Clinical Neuromuscular Disease, 2019, 20, 135-137.	0.7	1
223	Mutation Spectrum of Tuberous Sclerosis Complex Patients in Indian Population. Journal of Pediatric Genetics, 2021, 10, 274-283.	0.7	1
224	Combined Methylmalonic Aciduria and Homocystinuria Presenting as Pulmonary Hypertension. Indian Journal of Pediatrics, 2021, 88, 1244-1246.	0.8	1
225	Epigenetic Abnormalities of 11p15.5 Region in Beckwith-Wiedemann Syndrome - A Report of Eight Indian Cases. Indian Journal of Pediatrics, 2020, 87, 175-178.	0.8	1
226	. The Current Status of Medical Genetics in India. , 2012, , 1161-1163.		1
227	Genetic Causes of Recurrent Miscarriage. , 0, , 76-76.		1
228	Genetics of Birth Defects. Donald School Journal of Ultrasound in Obstetrics and Gynecology, 2010, 4, 327-331.	0.3	1
229	Spine radiograph in dysplasias: A pictorial essay. Indian Journal of Radiology and Imaging, 2020, 30, 436-447.	0.8	1
230	First case report of Penttinen syndrome from India. American Journal of Medical Genetics, Part A, 2021, , .	1.2	1
231	Post-mortem MRI in stillbirth: Normal imaging appearances. European Journal of Radiology, 2022, 148, 110166.	2.6	1
232	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatrics, 2018, 55, 474-477.	0.4	1
233	STAMBP gene mutation causing microcephalyâ€capillary malformation syndrome: a recognizable developmental and epileptic encephalopathy. Epileptic Disorders, 2022, 24, 602-605.	1.3	1
234	Prenatal diagnosis and treatment of steroid 21-hydroxylase deficiency (congenital adrenal) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 62 Td (	0.8	0

#	Article	IF	Citations
235	Congenital myotonic dystrophy. Indian Journal of Pediatrics, 2001, 68, 451-453.	0.8	0
236	Congenital synspondylism: report of two cases. European Journal of Radiology Extra, 2005, 53, 43-46.	0.1	0
237	T1387 Idiopathic Chronic Pancreatitis, Diabetes, and Smoking are Significant Risk Factors for Pancreatic Cancer: Results of Case-Control and Cohort Studies. Gastroenterology, 2010, 138, S-551.	1.3	0
238	471 High Prevalence of Pre-Neoplastic Lesions and Loss of Heterozygosity at Tumour Suppressor Genes in Patients With Gallbladder Stones: Implications for Etiopathogenesis of Gallbladder Cancer. Gastroenterology, 2012, 142, S-99.	1.3	0
239	TMC1 may be a common gene for nonsyndromic hereditary hearing loss in Indian population. Molecular Cytogenetics, 2014, 7, P70.	0.9	0
240	Molecular analysis of mucopolysaccharidoses: identification and characterization of pathogenic mutations in Indian population. Molecular Cytogenetics, 2014, 7, P60.	0.9	0
241	Application of Chromosomal Microarray and Multiplex Ligation-dependent Probe Amplification in prenatal diagnosis. Molecular Cytogenetics, 2014, 7, P127.	0.9	0
242	Neurofibromatosis type II (Wishart type). Journal of Pediatric Neurology, 2015, 07, 333-335.	0.2	0
243	Validation of Polymerase Chain Reactionâ€“Based Assay to Detect Actual Number of CGG Repeats in FMR1 Gene in Indian Fragile X Syndrome Patients. Journal of Child Neurology, 2017, 32, 371-378.	1.4	0
244	Echogenic KidneysÂas an Antenatal Clue to the Metabolic Etiology: A Case Report. Journal of Fetal Medicine, 2019, 6, 95-97.	0.1	0
245	Stippled keratoderma and nail dystrophy associated with hyperkeratotic pustular lesions in a 2â€yearâ€old boy. Pediatric Dermatology, 2020, 37, e64-e66.	0.9	0
246	Duchenne Muscular Dystrophy- Where Genetic Testing is Inevitable and Vital!. Indian Journal of Pediatrics, 2020, 87, 487-488.	0.8	0
247	Corrigendum to â€œSpectrum of amyloglucosidase mutations in Asian Indian patients with Glycogen storage disease type <scp>III</scp>. Am J Med Genet Part A. 2020;<scp>182A</scp>:1190â€“1,200â€. American Journal of Medical Genetics, Part A, 2021, 185, 1008-1010.	1.2	0
248	Physical Growth and Its Determinants in Indian Children with Down Syndrome, from 3 Months to 5 Years of Age. Indian Journal of Pediatrics, 2021, , 1.	0.8	0
249	TORCH Infections and Mental Retardation. , 0, , 99-99.		0
250	Congenital Infections and Mental Retardation. , 0, , 30-30.		0
251	Laboratory Techniques in Prenatal Diagnosis of Genetic Disorders. , 0, , 95-95.		0
252	Congenital TORCH Infections and Mental Retardation. , 0, , 81-81.		0

#	Article	IF	Citations
253	Common Genetic Disorders. , 0, , 855-855.		0
254	Cystic Fibrosis: Clinical Manifestations and Treatment. , 0, , 360-360.		0
255	Genetic/Syndromic Obesity: Praderâ€“Willi Syndrome. , 0, , 142-142.		0
256	Does Diet Offset the Effect of Veiling on Bone Mineral Density of premenopausal Indian women. MAMC Journal of Medical Sciences, 2015, 1, 12.	0.2	0
257	Antenatal Diagnosis of Hemoglobinopathies. , 0, , 204-204.		0
258	Monosomy 1p36: Report of a cohort of 13 Asian Indian patients. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0
259	Aquagenic Wrinkling of Skin: A Screening Test for Cystic Fibrosis. Indian Pediatrics, 2019, 56, 109-113.	0.4	0
260	A novel leaky splice variant in centromere protein J (CENPJ)â€associated Seckel syndrome. Annals of Human Genetics, 2022, , .	0.8	0