

# Madhulika Kabra

## List of Publications by Year in descending order

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Version: 2024-02-01

260  
papers

3,617  
citations

172207

29  
h-index

243296

44  
g-index

271  
all docs

271  
docs citations

271  
times ranked

4757  
citing authors

#	ARTICLE	IF	CITATIONS
1	Spectrum of Movement Disorders of Late-Onset Niemann-Pick Disease Type C. Canadian Journal of Neurological Sciences, 2022, 49, 804-808.	0.3	2
2	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.	1.0	8
3	Monosomy 1p36: Report of a cohort of 13 Asian Indian patients. American Journal of Medical Genetics, Part A, 2022, , .	0.7	0
4	Post-mortem MRI in stillbirth: Normal imaging appearances. European Journal of Radiology, 2022, 148, 110166.	1.2	1
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	A novel leaky splice variant in centromere protein J ( <i>CENPJ</i> ) associated Seckel syndrome. Annals of Human Genetics, 2022, , .	0.3	0
7	<i>STAMBP</i> gene mutation causing microcephaly capillary malformation syndrome: a recognizable developmental and epileptic encephalopathy. Epileptic Disorders, 2022, 24, 602-605.	0.7	1
8	Diagnosis and Management of Global Development Delay: Consensus Guidelines of Growth, Development and Behavioral Pediatrics Chapter, Neurology Chapter and Neurodevelopment Pediatrics Chapter of the Indian Academy of Pediatrics. Indian Pediatrics, 2022, 59, 401-415.	0.2	8
9	Mutation Spectrum of Tuberous Sclerosis Complex Patients in Indian Population. Journal of Pediatric Genetics, 2021, 10, 274-283.	0.3	1
10	Corrigendum to "Spectrum of amyloglucosidase mutations in Asian Indian patients with Glycogen storage disease type III". Am J Med Genet Part A. 2020;182A:1190-1200. American Journal of Medical Genetics, Part A, 2021, 185, 1008-1010.	0.7	0
11	Report of an Indian Family with Sengers Syndrome. Indian Journal of Pediatrics, 2021, 88, 92-92.	0.3	2
12	Late onset Pompe Disease in India " Beyond the Caucasian phenotype. Neuromuscular Disorders, 2021, 31, 431-441.	0.3	6
13	A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for early-onset monogenic disorders in Indians. Human Mutation, 2021, 42, e15-e61.	1.1	25
14	Hydrops fetalis in <i>PKD1L1</i> related heterotaxy: Report of two fetuses and expanding the phenotypic and molecular spectrum. Annals of Human Genetics, 2021, 85, 138-145.	0.3	5
15	Association of Sleep Apnea With Development and Behavior in Down Syndrome: A Prospective Clinical and Polysomnographic Study. Pediatric Neurology, 2021, 116, 7-13.	1.0	16
16	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
17	Utility of fetal whole exome sequencing in the etiological evaluation and outcome of nonimmune hydrops fetalis. Prenatal Diagnosis, 2021, 41, 1414-1424.	1.1	7
18	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.	0.3	0

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19	Functional characterization of novel variants in <i>SMPD1</i> in Indian patients with acid sphingomyelinase deficiency. <i>Human Mutation</i> , 2021, 42, 1336-1350.	1.1	6
20	Combined Methylmalonic Aciduria and Homocystinuria Presenting as Pulmonary Hypertension. <i>Indian Journal of Pediatrics</i> , 2021, 88, 1244-1246.	0.3	1
21	First case report of Penttinen syndrome from India. <i>American Journal of Medical Genetics, Part A</i> , 2021, , .	0.7	1
22	Rapid Eye Movement (REM) Sleep Behavior Disorder and REM Sleep with Atonia in the Young. <i>Canadian Journal of Neurological Sciences</i> , 2020, 47, 100-108.	0.3	7
23	Clinical and Molecular Disease Spectrum and Outcomes in Patients with Infantile-Onset Pompe Disease. <i>Journal of Pediatrics</i> , 2020, 216, 44-50.e5.	0.9	22
24	First report of THOC6 related intellectual disability (Beaulieu Boycott Innes syndrome) in two siblings from India. <i>European Journal of Medical Genetics</i> , 2020, 63, 103742.	0.7	6
25	Pathogenic/likely pathogenic variants in the <i>SHOX</i> , <i>GHR</i> and <i>IGFALS</i> genes among Indian children with idiopathic short stature. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2020, 33, 79-88.	0.4	15
26	Stippled keratoderma and nail dystrophy associated with hyperkeratotic pustular lesions in a 2-year-old boy. <i>Pediatric Dermatology</i> , 2020, 37, e64-e66.	0.5	0
27	The impact of COVID-19 pandemic on the diagnosis and management of inborn errors of metabolism: A global perspective. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 285-288.	0.5	31
28	Identification and characterization of 30 novel pathogenic variations in 69 unrelated Indian patients with Mucopolidosis Type II and Type III. <i>Journal of Human Genetics</i> , 2020, 65, 971-984.	1.1	3
29	Duchenne Muscular Dystrophy- Where Genetic Testing is Inevitable and Vital!. <i>Indian Journal of Pediatrics</i> , 2020, 87, 487-488.	0.3	0
30	Biallelic loss-of-function novel variants in <i>LTBP3</i> related skeletal dysplasia: Report of first patient from India. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1944-1946.	0.7	5
31	Cystic Fibrosis Presenting as Pseudo-Bartter Syndrome: An Important Diagnosis that is Missed!. <i>Indian Journal of Pediatrics</i> , 2020, 87, 726-732.	0.3	18
32	“Go for it, dream big, work hard and persist”: A message to the next generation of CF leaders in recognition of International Women's Day 2020. <i>Journal of Cystic Fibrosis</i> , 2020, 19, 184-193.	0.3	3
33	Spectrum of amyloglucosidase mutations in Asian Indian patients with Glycogen storage disease type III. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1190-1200.	0.7	6
34	Management of Infants with Congenital Adrenal Hyperplasia. <i>Indian Pediatrics</i> , 2020, 57, 159-164.	0.2	5
35	Natural history of non-lethal Raine syndrome during childhood. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 93.	1.2	17
36	Epigenetic Abnormalities of 11p15.5 Region in Beckwith-Wiedemann Syndrome - A Report of Eight Indian Cases. <i>Indian Journal of Pediatrics</i> , 2020, 87, 175-178.	0.3	1

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37	Newborn Screening and Diagnosis of Infants with Congenital Adrenal Hyperplasia. <i>Indian Pediatrics</i> , 2020, 57, 49-55.	0.2	10
38	Methylene Tetrahydrofolate Reductase Deficiency. <i>Indian Journal of Pediatrics</i> , 2020, 87, 951-953.	0.3	2
39	Impact of parental origin of X-chromosome on clinical and biochemical profile in Turner syndrome. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2020, 33, 1155-1163.	0.4	4
40	Weak Ligaments and Sloping Joints: A New Hypothesis for Development of Congenital Atlantoaxial Dislocation and Basilar Invagination. <i>Neurospine</i> , 2020, 17, 843-856.	1.1	5
41	Spine radiograph in dysplasias: A pictorial essay. <i>Indian Journal of Radiology and Imaging</i> , 2020, 30, 436-447.	0.3	1
42	Newborn Screening and Diagnosis of Infants with Congenital Adrenal Hyperplasia. <i>Indian Pediatrics</i> , 2020, 57, 49-55.	0.2	3
43	Management of Infants with Congenital Adrenal Hyperplasia. <i>Indian Pediatrics</i> , 2020, 57, 159-164.	0.2	2
44	Imatinib trough levels: a potential biomarker to predict cytogenetic and molecular response in newly diagnosed patients with chronic myeloid leukemia. <i>Leukemia and Lymphoma</i> , 2019, 60, 418-425.	0.6	13
45	Effects of Exercise Intervention Program on Bone Mineral Accretion in Children and Adolescents with Cystic Fibrosis: A Randomized Controlled Trial. <i>Indian Journal of Pediatrics</i> , 2019, 86, 987-994.	0.3	20
46	Decoding of novel missense TSC2 gene variants using in-silico methods. <i>BMC Medical Genetics</i> , 2019, 20, 164.	2.1	4
47	Report of Two Novel Mutations in Indian Patients with Rothmundâ€“Thomson Syndrome. <i>Journal of Pediatric Genetics</i> , 2019, 08, 163-167.	0.3	3
48	Report of a Novel Homozygous Nonsense DDR2 Mutation in an Indian Adult Male with Spondylo-meta-epiphyseal Dysplasia, Short Limb-Abnormal Calcification Type. <i>Journal of Pediatric Genetics</i> , 2019, 08, 153-156.	0.3	1
49	Growth Pattern and Clinical Profile of Indian Children with Classical 21-Hydroxylase Deficiency Congenital Adrenal Hyperplasia on Treatment. <i>Indian Journal of Pediatrics</i> , 2019, 86, 496-502.	0.3	7
50	Spectrum of ARSA variations in Asian Indian patients with Arylsulfatase A deficient metachromatic leukodystrophy. <i>Journal of Human Genetics</i> , 2019, 64, 323-331.	1.1	15
51	Genetic polymorphisms associated with obesity and non-alcoholic fatty liver disease in Asian Indian adolescents. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2019, 32, 749-758.	0.4	13
52	Echogenic Kidneysâ€“as an Antenatal Clue to the Metabolic Etiology: A Case Report. <i>Journal of Fetal Medicine</i> , 2019, 6, 95-97.	0.1	0
53	Report of Another Mutation Proven Case of Carbonic Anhydrase II Deficiency. <i>Journal of Pediatric Genetics</i> , 2019, 08, 091-094.	0.3	6
54	Aquagenic Wrinkling of Skin: A Screening Test for Cystic Fibrosis. <i>Indian Pediatrics</i> , 2019, 56, 109-113.	0.2	9

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55	Thenar Hypertrophy and Electrical Myotonia in Pompe Disease. <i>Journal of Clinical Neuromuscular Disease</i> , 2019, 20, 135-137.	0.3	1
56	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 150-158.	0.7	40
57	Aquagenic Wrinkling of Skin: A Screening Test for Cystic Fibrosis. <i>Indian Pediatrics</i> , 2019, 56, 109-113.	0.2	0
58	A novel homozygous mutation in POLR3A gene causing 4H syndrome: a case report. <i>BMC Pediatrics</i> , 2018, 18, 126.	0.7	11
59	Batten disease: biochemical and molecular characterization revealing novel PPT1 and TPP1 gene mutations in Indian patients. <i>BMC Neurology</i> , 2018, 18, 203.	0.8	10
60	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. <i>Indian Pediatrics</i> , 2018, 55, 474-477.	0.2	3
61	Spectrum of GJB2 gene variants in Indian children with non-syndromic hearing loss. <i>Indian Journal of Medical Research</i> , 2018, 147, 615.	0.4	4
62	Identification of a case of SRD5A3-congenital disorder of glycosylation (CDG1Q) by exome sequencing. <i>Indian Journal of Medical Research</i> , 2018, 147, 422.	0.4	9
63	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. <i>Indian Pediatrics</i> , 2018, 55, 474-477.	0.2	1
64	Cardiovascular Autonomic Dysfunction in Children and Adolescents With Rett Syndrome. <i>Pediatric Neurology</i> , 2017, 70, 61-66.	1.0	23
65	Validation of Polymerase Chain Reaction-Based Assay to Detect Actual Number of CCG Repeats in FMR1 Gene in Indian Fragile X Syndrome Patients. <i>Journal of Child Neurology</i> , 2017, 32, 371-378.	0.7	0
66	Prevalence of Sleep Abnormalities in Indian Children With Autism Spectrum Disorder: A Cross-Sectional Study. <i>Pediatric Neurology</i> , 2017, 74, 62-67.	1.0	29
67	Influence of MDR1 and CYP3A5 genetic polymorphisms on trough levels and therapeutic response of imatinib in newly diagnosed patients with chronic myeloid leukemia. <i>Pharmacological Research</i> , 2017, 120, 138-145.	3.1	23
68	Do polymorphisms in <i>MDR1</i> and <i>CYP3A5</i> genes influence the risk of cytogenetic relapse in patients with chronic myeloid leukemia on imatinib therapy?. <i>Leukemia and Lymphoma</i> , 2017, 58, 2218-2226.	0.6	5
69	Congenital Cytomegalovirus Infection and Permanent Hearing Loss in Rural North Indian Children. <i>Pediatric Infectious Disease Journal</i> , 2017, 36, 670-673.	1.1	12
70	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. <i>Journal of Dermatological Science</i> , 2017, 86, 30-36.	1.0	16
71	Asparagine Synthetase deficiency-report of a novel mutation and review of literature. <i>Metabolic Brain Disease</i> , 2017, 32, 1889-1900.	1.4	24
72	Prognostic Utility of Clinical Epilepsy Severity Score Versus Pretreatment Hypsarrhythmia Scoring in Children With West Syndrome. <i>Clinical EEG and Neuroscience</i> , 2017, 48, 280-287.	0.9	4

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73	Whole exome sequencing identifies a homozygous nonsense variation in ALMS1 gene in a patient with syndromic obesity. <i>Obesity Research and Clinical Practice</i> , 2017, 11, 241-246.	0.8	14
74	Bone mineral density of Indian children and adolescents with cystic fibrosis. <i>Indian Pediatrics</i> , 2017, 54, 545-549.	0.2	7
75	Prenatal diagnosis of steroid 21-hydroxylase-deficient congenital adrenal hyperplasia: Experience from a tertiary care centre in India. <i>Indian Journal of Medical Research</i> , 2017, 145, 194-202.	0.4	4
76	Frequency of primary mutations of Leber's hereditary optic neuropathy patients in North Indian population. <i>Indian Journal of Ophthalmology</i> , 2017, 65, 1156.	0.5	13
77	Pelvic radiograph in skeletal dysplasias: An approach. <i>Indian Journal of Radiology and Imaging</i> , 2017, 27, 187-199.	0.3	13
78	Identification of a novel homozygous mutation in transmembrane channel like 1 () gene, one of the second-tier hearing loss genes after in India. <i>Indian Journal of Medical Research</i> , 2017, 145, 492-497.	0.4	3
79	Pycnodysostosis: mutation spectrum in five unrelated Indian children. <i>Clinical Dysmorphology</i> , 2016, 25, 113-120.	0.1	16
80	Caffey's Disease: Two Cases Presenting with Unexplained Fever. <i>Indian Journal of Pediatrics</i> , 2016, 83, 1499-1500.	0.3	2
81	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 410-417.	0.7	31
82	Frequencies of CYP2C9 polymorphisms in North Indian population and their association with drug levels in children on phenytoin monotherapy. <i>BMC Pediatrics</i> , 2016, 16, 66.	0.7	15
83	Ghosal type hematodiaphyseal dysplasia. <i>Indian Pediatrics</i> , 2016, 53, 347-348.	0.2	11
84	Spondylometaphyseal Dysplasia Corner Fracture (Sutcliffe) Type. <i>Indian Journal of Pediatrics</i> , 2016, 83, 1191-1194.	0.3	4
85	Editorial: New Horizons in Genetic Diagnosis in Pediatric Practice: The Excitement and Challenges!. <i>Indian Journal of Pediatrics</i> , 2016, 83, 1131-1132.	0.3	1
86	Novel Genetic, Clinical, and Pathomechanistic Insights into TFG-Associated Hereditary Spastic Paraplegia. <i>Human Mutation</i> , 2016, 37, 1157-1161.	1.1	17
87	Spectrum of <i>SMPD1</i> mutations in Asian Indian patients with acid sphingomyelinase (ASM)-deficient Niemann-Pick disease. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2719-2730.	0.7	15
88	Genetically Determined Chronic Pancreatitis but not Alcoholic Pancreatitis Is a Strong Risk Factor for Pancreatic Cancer. <i>Pancreas</i> , 2016, 45, 1478-1484.	0.5	40
89	Editorial. <i>Indian Pediatrics</i> , 2016, 53, 19-26.	0.2	1
90	ADRB2 polymorphism and salbutamol responsiveness in Northern Indian children with mild to moderate exacerbation of asthma. <i>Indian Pediatrics</i> , 2016, 53, 211-215.	0.2	8

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91	Seventeen Novel Mutations in <i>PCCA</i> and <i>PCCB</i> Genes in Indian Propionic Acidemia Patients, and Their Outcomes. <i>Genetic Testing and Molecular Biomarkers</i> , 2016, 20, 373-382.	0.3	12
92	Application of chromosomal microarrays in the evaluation of intellectual disability/global developmental delay patients – A study from a tertiary care genetic centre in India. <i>Gene</i> , 2016, 590, 109-119.	1.0	11
93	Genetic Studies in Autism. <i>Indian Journal of Pediatrics</i> , 2016, 83, 1133-1140.	0.3	3
94	Smith-Magenis Syndrome: Face Speaks. <i>Indian Journal of Pediatrics</i> , 2016, 83, 589-593.	0.3	5
95	Chanarin Dorfman syndrome: a case report with novel nonsense mutation. <i>Gene</i> , 2016, 575, 359-362.	1.0	11
96	Identification of GJB6 gene mutation in an Indian man with Clouston syndrome. <i>Indian Journal of Dermatology, Venereology and Leprology</i> , 2016, 82, 697.	0.2	6
97	The spectrum of leukodystrophies in children: Experience at a tertiary care centre from North India. <i>Annals of Indian Academy of Neurology</i> , 2016, 19, 332.	0.2	13
98	Williams-Beuren Syndrome: Experience of 43 Patients and a Report of an Atypical Case from a Tertiary Care Center in India. <i>Cytogenetic and Genome Research</i> , 2015, 146, 187-194.	0.6	8
99	Adverse pregnancy outcome in patients with low pregnancy-associated plasma protein-A: The Indian experience. <i>Journal of Obstetrics and Gynaecology Research</i> , 2015, 41, 1003-1008.	0.6	13
100	Live births in women with recurrent hydatidiform mole and two NLRP7 mutations. <i>Reproductive BioMedicine Online</i> , 2015, 31, 120-124.	1.1	36
101	Acrodermatitis Dysmetabolica - Report of Two Cases. <i>Indian Journal of Pediatrics</i> , 2015, 82, 869-870.	0.3	4
102	Clinical profile and mutation analysis of xeroderma pigmentosum in Indian patients. <i>Indian Journal of Dermatology, Venereology and Leprology</i> , 2015, 81, 16.	0.2	14
103	Prevalence of UGT1A6 polymorphisms in children with epilepsy on valproate monotherapy. <i>Neurology India</i> , 2015, 63, 35.	0.2	16
104	Prenatal diagnosis in India is not limited to sex selection. <i>Genetics in Medicine</i> , 2015, 17, 88-88.	1.1	1
105	Recurrent and novel GLB1 mutations in India. <i>Gene</i> , 2015, 567, 173-181.	1.0	22
106	Velaglucerase alfa (VPRIV) enzyme replacement therapy in patients with Gaucher disease: Long-term data from phase III clinical trials. <i>American Journal of Hematology</i> , 2015, 90, 584-591.	2.0	39
107	Atypical late presentation in neonatal-onset multisystem inflammatory disease (NOMID). <i>Journal of Pediatric Neurology</i> , 2015, 07, 301-305.	0.0	2
108	Neurofibromatosis type II (Wishart type). <i>Journal of Pediatric Neurology</i> , 2015, 07, 333-335.	0.0	0

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109	Bilateral fronto-parietal polymicrogyria in an Indian infant. <i>Journal of Pediatric Neurology</i> , 2015, 09, 251-253.	0.0	2
110	Enhanced Reprogramming Efficiency and Kinetics of Induced Pluripotent Stem Cells Derived from Human Duchenne Muscular Dystrophy. <i>PLOS Currents</i> , 2015, 7, .	1.4	4
111	Does Diet Offset the Effect of Veiling on Bone Mineral Density of premenopausal Indian women. <i>MAMC Journal of Medical Sciences</i> , 2015, 1, 12.	0.2	0
112	Dystrophinopathy Diagnosis Made Easy. <i>Journal of Child Neurology</i> , 2014, 29, 469-474.	0.7	5
113	Skin Biopsy. <i>Journal of Child Neurology</i> , 2014, 29, NP5-NP8.	0.7	5
114	Molecular Diagnosis of Hereditary Fructose Intolerance: Founder Mutation in a Community from India. <i>JIMD Reports</i> , 2014, 19, 85-93.	0.7	18
115	Prenatal screening: Perspective for the pediatrician. <i>Indian Pediatrics</i> , 2014, 51, 959-962.	0.2	5
116	Establishing national neonatal perinatal database and birth defects registry network – Need of the hour!. <i>Indian Pediatrics</i> , 2014, 51, 693-696.	0.2	10
117	Status of iodine deficiency disorder in district Udham Singh Nagar, Uttarakhand state India. <i>Indian Journal of Endocrinology and Metabolism</i> , 2014, 18, 419.	0.2	7
118	Encephalocraniocutaneous Lipomatosis With Neurocutaneous Melanosis. <i>Journal of Child Neurology</i> , 2014, 29, 846-849.	0.7	10
119	Glutaric Acidemia Type 1-Clinico-Molecular Profile and Novel Mutations in GCDH Gene in Indian Patients. <i>JIMD Reports</i> , 2014, 21, 45-55.	0.7	25
120	Mutations in CSPP1 Lead to Classical Joubert Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 80-86.	2.6	75
121	Neurodevelopmental and epilepsy outcome in children aged one to five years with infantile spasms – A North Indian cohort. <i>Epilepsy Research</i> , 2014, 108, 526-534.	0.8	21
122	Behavioral comorbidity in children and adolescents with epilepsy. <i>Journal of Clinical Neuroscience</i> , 2014, 21, 1337-1340.	0.8	11
123	Iodine Nutritional Status Among Neonates in the Solan District, Himachal Pradesh, India. <i>Journal of Community Health</i> , 2014, 39, 987-989.	1.9	2
124	TMC1 may be a common gene for nonsyndromic hereditary hearing loss in Indian population. <i>Molecular Cytogenetics</i> , 2014, 7, P70.	0.4	0
125	Molecular analysis of mucopolysaccharidoses: identification and characterization of pathogenic mutations in Indian population. <i>Molecular Cytogenetics</i> , 2014, 7, P60.	0.4	0
126	Application of Chromosomal Microarray and Multiplex Ligation-dependent Probe Amplification in prenatal diagnosis. <i>Molecular Cytogenetics</i> , 2014, 7, P127.	0.4	0



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127	Iodine nutrition status amongst neonates in Kangra district, Himachal Pradesh. <i>Journal of Trace Elements in Medicine and Biology</i> , 2014, 28, 351-353.	1.5	4
128	Targeted Deep Resequencing Identifies <i>MID2</i> Mutation for X-Linked Intellectual Disability with Varied Disease Severity in a Large Kindred from India. <i>Human Mutation</i> , 2014, 35, 41-44.	1.1	36
129	Active surveillance for intussusception in a phase III efficacy trial of an oral monovalent rotavirus vaccine in India. <i>Vaccine</i> , 2014, 32, A104-A109.	1.7	24
130	At Least an Infantogram if not Perinatal Autopsy. <i>Journal of Fetal Medicine</i> , 2014, 01, 33-39.	0.1	4
131	Diagnosis and Management of Down Syndrome. <i>Indian Journal of Pediatrics</i> , 2014, 81, 560-567.	0.3	41
132	Disseminated cryptococcosis. <i>Indian Pediatrics</i> , 2014, 51, 225-226.	0.2	3
133	Mutation spectrum of <i>COL1A1</i> and <i>COL1A2</i> genes in Indian patients with osteogenesis imperfecta. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1482-1489.	0.7	24
134	Leukodystrophy Presenting as Acute-Onset Polyradiculoneuropathy. <i>Pediatric Neurology</i> , 2014, 50, 616-618.	1.0	7
135	Profile of prothrombotic factors in Indian children with ischemic stroke. <i>Journal of Clinical Neuroscience</i> , 2014, 21, 1315-1318.	0.8	11
136	Sequential Occurrence of Preneoplastic Lesions and Accumulation of Loss of Heterozygosity in Patients With Gallbladder Stones Suggest Causal Association With Gallbladder Cancer. <i>Annals of Surgery</i> , 2014, 260, 1073-1080.	2.1	40
137	Menkes disease – An important cause of early onset refractory seizures. <i>Journal of Pediatric Neurosciences</i> , 2014, 9, 11.	0.2	10
138	Clinical profile and treatment status of subjects with cleft lip and palate anomaly in India: Preliminary report of a three-center study. <i>Journal of Cleft Lip Palate and Craniofacial Anomalies</i> , 2014, 1, 26.	0.1	8
139	Imaging in Neonatal Maple Syrup Urine Disease. <i>Indian Journal of Pediatrics</i> , 2013, 80, 87-88.	0.3	7
140	National newborn screening program – Still a hype or a hope now?. <i>Indian Pediatrics</i> , 2013, 50, 639-643.	0.2	19
141	Coping Strategies of Parents of Down Syndrome Children in India. <i>Indian Journal of Pediatrics</i> , 2013, 80, 534-535.	0.3	3
142	Prevalence of Celiac Disease in Indian Children with Down Syndrome and its Clinical and Laboratory Predictors. <i>Indian Journal of Pediatrics</i> , 2013, 80, 114-117.	0.3	13
143	Macrocephaly with Diffuse White Matter Changes Simulating a Leukodystrophy in Menkes Disease. <i>Indian Journal of Pediatrics</i> , 2013, 80, 160-162.	0.3	7
144	Intellectual disability in Indian children: experience with a stratified approach for etiological diagnosis. <i>Indian Pediatrics</i> , 2013, 50, 1125-1130.	0.2	20

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145	Schwartz Jampel syndrome in children. <i>Journal of Clinical Neuroscience</i> , 2013, 20, 313-317.	0.8	15
146	Noninvasive screening for preclinical atherosclerosis in children on phenytoin or carbamazepine monotherapy: A cross sectional study. <i>Epilepsy Research</i> , 2013, 107, 121-126.	0.8	13
147	Severe neuronopathic autosomal recessive osteopetrosis due to homozygous deletions affecting OSTM1. <i>Bone</i> , 2013, 55, 292-297.	1.4	22
148	Peripheral neuropathy in cystic fibrosis: A prevalence study. <i>Journal of Cystic Fibrosis</i> , 2013, 12, 754-760.	0.3	16
149	Efficacy of modified constraint induced movement therapy in improving upper limb function in children with hemiplegic cerebral palsy: A randomized controlled trial. <i>Brain and Development</i> , 2013, 35, 870-876.	0.6	51
150	Increase in Iodine Deficiency Disorder due to Inadequate Sustainability of Supply of Iodized Salt in District Solan, Himachal Pradesh. <i>Journal of Tropical Pediatrics</i> , 2013, 59, 514-515.	0.7	3
151	Prenatal Diagnosis of Fetal Peters <sup>™</sup> Plus Syndrome: A Case Report. <i>Case Reports in Genetics</i> , 2013, 2013, 1-3.	0.1	8
152	Fluorescence in situ hybridization (FISH) using non-commercial probes in the diagnosis of clinically suspected microdeletion syndromes. <i>Indian Journal of Medical Research</i> , 2013, 138, 135-42.	0.4	2
153	Inherited 5p deletion syndrome due to paternal balanced translocation: Phenotypic heterogeneity due to duplication of 8q and 12p. <i>Journal of Pediatric Genetics</i> , 2013, 2, 163-9.	0.3	3
154	Brachytelephalangic chondrodysplasia punctata. <i>Clinical Dysmorphology</i> , 2012, 21, 113-117.	0.1	3
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