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	Spectrum of Movement Disorders of Late-Onset Niemann-Pick Disease Type C. Canadian Journal of Neurological Sciences, 2022, 49, 804-808.	0.5	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.	1.7	8
3	Monosomy 1p36: Report of a cohort of 13 Asian Indian patients. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0
4	Post-mortem MRI in stillbirth: Normal imaging appearances. European Journal of Radiology, 2022, 148, 110166.	2.6	1
5	Genotype-phenotype spectrum of 130 unrelated Indian families with Mucopolysaccharidosis type II. European Journal of Medical Genetics, 2022, 65, 104447.	1.3	3
6	A novel leaky splice variant in centromere protein J (CENPJ)â€associated Seckel syndrome. Annals of Human Genetics, 2022, , .	0.8	0
7	STAMBP gene mutation causing microcephalyâ€capillary malformation syndrome: a recognizable developmental and epileptic encephalopathy. Epileptic Disorders, 2022, 24, 602-605.	1.3	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.	0.4	8
9	Mutation Spectrum of Tuberous Sclerosis Complex Patients in Indian Population. Journal of Pediatric Genetics, 2021, 10, 274-283.	0.7	1
10	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
11	Report of an Indian Family with Sengers Syndrome. Indian Journal of Pediatrics, 2021, 88, 92-92.	0.8	2
12	Late onset Pompe Disease in India â€“ Beyond the Caucasian phenotype. Neuromuscular Disorders, 2021, 31, 431-441.	0.6	6
13	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
14	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
15	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
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.	1.3	6
17	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
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.	0.8	0

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19	Functional characterization of novel variants in SMPD1 in Indian patients with acid sphingomyelinase deficiency. Human Mutation, 2021, 42, 1336-1350.	2.5	6
20	Combined Methylmalonic Aciduria and Homocystinuria Presenting as Pulmonary Hypertension. Indian Journal of Pediatrics, 2021, 88, 1244-1246.	0.8	1
21	First case report of Penttinen syndrome from India. American Journal of Medical Genetics, Part A, 2021, , .	1.2	1
22	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
23	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
24	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
25	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
26	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
27	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
28	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
29	Duchenne Muscular Dystrophy- Where Genetic Testing is Inevitable and Vital!. Indian Journal of Pediatrics, 2020, 87, 487-488.	0.8	0
30	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
31	Cystic Fibrosis Presenting as Pseudo-Bartter Syndrome: An Important Diagnosis that is Missed!. Indian Journal of Pediatrics, 2020, 87, 726-732.	0.8	18
32	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
33	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
34	Management of Infants with Congenital Adrenal Hyperplasia. Indian Pediatrics, 2020, 57, 159-164.	0.4	5
35	Natural history of non-lethal Raine syndrome during childhood. Orphanet Journal of Rare Diseases, 2020, 15, 93.	2.7	17
36	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

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37	Newborn Screening and Diagnosis of Infants with Congenital Adrenal Hyperplasia. Indian Pediatrics, 2020, 57, 49-55.	0.4	10
38	Methylene Tetrahydrofolate Reductase Deficiency. Indian Journal of Pediatrics, 2020, 87, 951-953.	0.8	2
39	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
40	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
41	Spine radiograph in dysplasias: A pictorial essay. Indian Journal of Radiology and Imaging, 2020, 30, 436-447.	0.8	1
42	Newborn Screening and Diagnosis of Infants with Congenital Adrenal Hyperplasia. Indian Pediatrics, 2020, 57, 49-55.	0.4	3
43	Management of Infants with Congenital Adrenal Hyperplasia. Indian Pediatrics, 2020, 57, 159-164.	0.4	2
44	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
45	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
46	Decoding of novel missense TSC2 gene variants using in-silico methods. BMC Medical Genetics, 2019, 20, 164.	2.1	4
47	Report of Two Novel Mutations in Indian Patients with Rothmundâ€“Thomson Syndrome. Journal of Pediatric Genetics, 2019, 08, 163-167.	0.7	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. Journal of Pediatric Genetics, 2019, 08, 153-156.	0.7	1
49	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
50	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
51	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
52	Echogenic KidneysÂas an Antenatal Clue to the Metabolic Etiology: A Case Report. Journal of Fetal Medicine, 2019, 6, 95-97.	0.1	0
53	Report of Another Mutation Proven Case of Carbonic Anhydrase II Deficiency. Journal of Pediatric Genetics, 2019, 08, 091-094.	0.7	6
54	Aquagenic Wrinkling of Skin: A Screening Test for Cystic Fibrosis. Indian Pediatrics, 2019, 56, 109-113.	0.4	9

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55	Thenar Hypertrophy and Electrical Myotonia in Pompe Disease. Journal of Clinical Neuromuscular Disease, 2019, 20, 135-137.	0.7	1
56	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
57	Aquagenic Wrinkling of Skin: A Screening Test for Cystic Fibrosis. Indian Pediatrics, 2019, 56, 109-113.	0.4	0
58	A novel homozygous mutation in POLR3A gene causing 4H syndrome: a case report. BMC Pediatrics, 2018, 18, 126.	1.7	11
59	Batten disease: biochemical and molecular characterization revealing novel PPT1 and TPP1 gene mutations in Indian patients. BMC Neurology, 2018, 18, 203.	1.8	10
60	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatrics, 2018, 55, 474-477.	0.4	3
61	Spectrum of GJB2 gene variants in Indian children with non-syndromic hearing loss. Indian Journal of Medical Research, 2018, 147, 615.	1.0	4
62	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
63	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatrics, 2018, 55, 474-477.	0.4	1
64	Cardiovascular Autonomic Dysfunction in Children and Adolescents With Rett Syndrome. Pediatric Neurology, 2017, 70, 61-66.	2.1	23
65	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
66	Prevalence of Sleep Abnormalities in Indian Children With Autism Spectrum Disorder: A Cross-Sectional Study. Pediatric Neurology, 2017, 74, 62-67.	2.1	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. Pharmacological Research, 2017, 120, 138-145.	7.1	23
68	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
69	Congenital Cytomegalovirus Infection and Permanent Hearing Loss in Rural North Indian Children. Pediatric Infectious Disease Journal, 2017, 36, 670-673.	2.0	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. Journal of Dermatological Science, 2017, 86, 30-36.	1.9	16
71	Asparagine Synthetase deficiency-report of a novel mutation and review of literature. Metabolic Brain Disease, 2017, 32, 1889-1900.	2.9	24
72	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

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73	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
74	Bone mineral density of Indian children and adolescents with cystic fibrosis. Indian Pediatrics, 2017, 54, 545-549.	0.4	7
75	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
76	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
77	Pelvic radiograph in skeletal dysplasias: An approach. Indian Journal of Radiology and Imaging, 2017, 27, 187-199.	0.8	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. Indian Journal of Medical Research, 2017, 145, 492-497.	1.0	3
79	Pycnodysostosis: mutation spectrum in five unrelated Indian children. Clinical Dysmorphology, 2016, 25, 113-120.	0.3	16
80	Caffeyâ€™s Disease: Two Cases Presenting with Unexplained Fever. Indian Journal of Pediatrics, 2016, 83, 1499-1500.	0.8	2
81	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. American Journal of Medical Genetics, Part A, 2016, 170, 410-417.	1.2	31
82	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
83	Ghosal type hematodiaphyseal dysplasia. Indian Pediatrics, 2016, 53, 347-348.	0.4	11
84	Spondylometaphyseal Dysplasia Corner Fracture (Sutcliffe) Type. Indian Journal of Pediatrics, 2016, 83, 1191-1194.	0.8	4
85	Editorial: New Horizons in Genetic Diagnosis in Pediatric Practice: The Excitement and Challenges!. Indian Journal of Pediatrics, 2016, 83, 1131-1132.	0.8	1
86	Novel Genetic, Clinical, and Pathomechanistic Insights into TFG-Associated Hereditary Spastic Paraplegia. Human Mutation, 2016, 37, 1157-1161.	2.5	17
87	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
88	Genetically Determined Chronic Pancreatitis but not Alcoholic Pancreatitis Is a Strong Risk Factor for Pancreatic Cancer. Pancreas, 2016, 45, 1478-1484.	1.1	40
89	Editorial. Indian Pediatrics, 2016, 53, 19-26.	0.4	1
90	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

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91	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
92	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
93	Genetic Studies in Autism. Indian Journal of Pediatrics, 2016, 83, 1133-1140.	0.8	3
94	Smith-Magenis Syndrome: Face Speaks. Indian Journal of Pediatrics, 2016, 83, 589-593.	0.8	5
95	Chanarin Dorfman syndrome: a case report with novel nonsense mutation. Gene, 2016, 575, 359-362.	2.2	11
96	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
97	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
98	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
99	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
100	Live births in women with recurrent hydatidiform mole and two NLRP7 mutations. Reproductive BioMedicine Online, 2015, 31, 120-124.	2.4	36
101	Acrodermatitis Dysmetabolica - Report of Two Cases. Indian Journal of Pediatrics, 2015, 82, 869-870.	0.8	4
102	Clinical profile and mutation analysis of xeroderma pigmentosum in Indian patients. Indian Journal of Dermatology, Venereology and Leprology, 2015, 81, 16.	0.6	14
103	Prevalence of UGT1A6 polymorphisms in children with epilepsy on valproate monotherapy. Neurology India, 2015, 63, 35.	0.4	16
104	Prenatal diagnosis in India is not limited to sex selection. Genetics in Medicine, 2015, 17, 88-88.	2.4	1
105	Recurrent and novel GLB1 mutations in India. Gene, 2015, 567, 173-181.	2.2	22
106	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
107	Atypical late presentation in neonatal-onset multisystem inflammatory disease (NOMID). Journal of Pediatric Neurology, 2015, 07, 301-305.	0.2	2
108	Neurofibromatosis type II (Wishart type). Journal of Pediatric Neurology, 2015, 07, 333-335.	0.2	0

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109	Bilateral fronto-parietal polymicrogyria in an Indian infant. Journal of Pediatric Neurology, 2015, 09, 251-253.	0.2	2
110	Enhanced Reprogramming Efficiency and Kinetics of Induced Pluripotent Stem Cells Derived from Human Duchenne Muscular Dystrophy. PLOS Currents, 2015, 7, .	1.4	4
111	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
112	Dystrophinopathy Diagnosis Made Easy. Journal of Child Neurology, 2014, 29, 469-474.	1.4	5
113	Skin Biopsy. Journal of Child Neurology, 2014, 29, NP5-NP8.	1.4	5
114	Molecular Diagnosis of Hereditary Fructose Intolerance: Founder Mutation in a Community from India. JIMD Reports, 2014, 19, 85-93.	1.5	18
115	Prenatal screening: Perspective for the pediatrician. Indian Pediatrics, 2014, 51, 959-962.	0.4	5
116	Establishing national neonatal perinatal database and birth defects registry network â€" Need of the hour!. Indian Pediatrics, 2014, 51, 693-696.	0.4	10
117	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
118	Encephalocraniocutaneous Lipomatosis With Neurocutaneous Melanosis. Journal of Child Neurology, 2014, 29, 846-849.	1.4	10
119	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
120	Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86.	6.2	75
121	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
122	Behavioral comorbidity in children and adolescents with epilepsy. Journal of Clinical Neuroscience, 2014, 21, 1337-1340.	1.5	11
123	Iodine Nutritional Status Among Neonates in the Solan District, Himachal Pradesh, India. Journal of Community Health, 2014, 39, 987-989.	3.8	2
124	TMC1 may be a common gene for nonsyndromic hereditary hearing loss in Indian population. Molecular Cytogenetics, 2014, 7, P70.	0.9	0
125	Molecular analysis of mucopolysaccharidoses: identification and characterization of pathogenic mutations in Indian population. Molecular Cytogenetics, 2014, 7, P60.	0.9	0
126	Application of Chromosomal Microarray and Multiplex Ligation-dependent Probe Amplification in prenatal diagnosis. Molecular Cytogenetics, 2014, 7, P127.	0.9	0

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127	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
128	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
129	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
130	At Least an Infantogram if not Perinatal Autopsy. Journal of Fetal Medicine, 2014, 01, 33-39.	0.1	4
131	Diagnosis and Management of Down Syndrome. Indian Journal of Pediatrics, 2014, 81, 560-567.	0.8	41
132	Disseminated cryptococcosis. Indian Pediatrics, 2014, 51, 225-226.	0.4	3
133	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
134	Leukodystrophy Presenting as Acute-Onset Polyradiculoneuropathy. Pediatric Neurology, 2014, 50, 616-618.	2.1	7
135	Profile of prothrombotic factors in Indian children with ischemic stroke. Journal of Clinical Neuroscience, 2014, 21, 1315-1318.	1.5	11
136	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
137	Menkes disease â€“ An important cause of early onset refractory seizures. Journal of Pediatric Neurosciences, 2014, 9, 11.	0.3	10
138	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
139	Imaging in Neonatal Maple Syrup Urine Disease. Indian Journal of Pediatrics, 2013, 80, 87-88.	0.8	7
140	National newborn screening program â€” Still a hype or a hope now?. Indian Pediatrics, 2013, 50, 639-643.	0.4	19
141	Coping Strategies of Parents of Down Syndrome Children in India. Indian Journal of Pediatrics, 2013, 80, 534-535.	0.8	3
142	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
143	Macrocephaly with Diffuse White Matter Changes Simulating a Leukodystrophy in Menkes Disease. Indian Journal of Pediatrics, 2013, 80, 160-162.	0.8	7
144	Intellectual disability in Indian children: experience with a stratified approach for etiological diagnosis. Indian Pediatrics, 2013, 50, 1125-1130.	0.4	20

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145	Schwartz Jampel syndrome in children. Journal of Clinical Neuroscience, 2013, 20, 313-317.	1.5	15
146	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
147	Severe neuronopathic autosomal recessive osteopetrosis due to homozygous deletions affecting OSTM1. Bone, 2013, 55, 292-297.	2.9	22
148	Peripheral neuropathy in cystic fibrosis: A prevalence study. Journal of Cystic Fibrosis, 2013, 12, 754-760.	0.7	16
149	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
150	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
151	Prenatal Diagnosis of Fetal Petersâ€™ Plus Syndrome: A Case Report. Case Reports in Genetics, 2013, 2013, 1-3.	0.2	8
152	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
153	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
154	Brachytelephalangic chondrodysplasia punctata. Clinical Dysmorphology, 2012, 21, 113-117.	0.3	3
155	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
156	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
157	Norrie Disease: First Mutation Report and Prenatal Diagnosis in an Indian Family. Indian Journal of Pediatrics, 2012, 79, 1529-1531.	0.8	5
158	Incidence of acute kidney injury in hospitalized children. Indian Pediatrics, 2012, 49, 537-542.	0.4	59
159	Mutation analysis of Indian patients with urea cycle defects. Indian Pediatrics, 2012, 49, 585-586.	0.4	8
160	Novel non-identical MECP2 mutations in Rett syndrome family: A rare presentation. Brain and Development, 2012, 34, 28-31.	1.1	4
161	. The Current Status of Medical Genetics in India. , 2012, , 1161-1163.		1
162	Molecular and structural analysis of metachromatic leukodystrophy patients in Indian population. Journal of the Neurological Sciences, 2011, 301, 38-45.	0.6	21

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163	The mutation spectrum in Indian patients with Gaucher disease. Genome Biology, 2011, 12, .	9.6	6
164	Molecular Genetic Studies in Indian Patients With Megalencephalic Leukoencephalopathy. Pediatric Neurology, 2011, 44, 450-458.	2.1	8
165	Molecular analysis of ABCD1 gene in Indian patients with X-linked Adrenoleukodystrophy. Clinica Chimica Acta, 2011, 412, 2289-2295.	1.1	6
166	Familial Progressive Hypermelanosis in Indian Monozygotic Twins. Pediatric Dermatology, 2011, 28, 62-65.	0.9	2
167	Intranasal versus intravenous lorazepam for control of acute seizures in children: A randomized openâ€label study. Epilepsia, 2011, 52, 788-793.	5.1	72
168	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
169	Hyperekplexia Masquerading as Epilepsy. Indian Journal of Pediatrics, 2011, 78, 757-757.	0.8	3
170	Acute Management of Sick Infants with Suspected Inborn Errors of Metabolism. Indian Journal of Pediatrics, 2011, 78, 854-859.	0.8	6
171	Wiedemannâ€“Rautenstrauch Syndrome: First Indian Case. Indian Journal of Pediatrics, 2011, 78, 1552-1555.	0.8	7
172	De novo deletion in MECP2 in a monozygotic twin pair: a case report. BMC Medical Genetics, 2011, 12, 113.	2.1	6
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