Inusha Panigrahi

List of Publications by Year
in descending order

134	1,232	516710 16	477307 29
papers	citations	h-index	g-index

138	138	138	1971
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. Nature Genetics, 2016, 48, 648-656.	21.4	119
2	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	3.2	85
3	Primordial dwarfism: overview of clinical and genetic aspects. Molecular Genetics and Genomics, 2016, 291, 1-15.	2.1	69
4	The Effect of Prophylactic Antipyretic Administration on Post-Vaccination Adverse Reactions and Antibody Response in Children: A Systematic Review. PLoS ONE, 2014, 9, e106629.	2.5	48
5	Craniosynostosis genetics: The mystery unfolds. Indian Journal of Human Genetics, 2011, 17, 48.	0.7	37
6	Thromboembolic complications in Î²-thalassemia: Beyond the horizon. Thrombosis Research, 2007, 120, 783-789.	1.7	35
7	Response to zolendronic acid in children with type III osteogenesis imperfecta. Journal of Bone and Mineral Metabolism, 2010, 28, 451-455.	2.7	32
8	Mutational spectrum of thalassemias in India. Indian Journal of Human Genetics, 2007, 13, 36.	0.7	32
9	Role of MTHFRC677Tpolymorphism in ischemic stroke. Neurology India, 2006, 54, 48.	0.4	29
10	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
11	Genetic determinants of phenotype in beta-thalassemia. Hematology, 2008, 13, 247-252.	1.5	25
12	Evaluation of the genetic basis of phenotypic heterogeneity in north Indian patients with Thalassemia major. European Journal of Haematology, 2010, 84, 531-537.	2.2	20
13	CRELD1 gene variants and atrioventricular septal defects in Down syndrome. Gene, 2018, 641, 180-185.	2.2	20
14	Rubinstein-taybi syndrome: Clinical profile of 11 patients and review of literature. Indian Journal of Human Genetics, 2012, 18, 161.	0.7	18
15	Concentrations of leptin, adiponectin and other metabolic parameters in non-obese children with Down syndrome. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 831-837.	0.9	18
16	Urinary Malondialdehyde Levels in Newborns following Delivery Room Resuscitation. Neonatology, 2008, 94, 96-99.	2.0	17
17	Maternal serum second trimester screening for chromosomal disorders and neural tube defects in a government hospital of North India. Prenatal Diagnosis, 2012, 32, 1192-1196.	2.3	17
18	Sickle Cell Anemiaâ€”Molecular Diagnosis and Prenatal Counseling: SGPGI Experience. Indian Journal of Pediatrics, 2012, 79, 68-74.	0.8	17

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19	Xmn1â€‹^GÎ³ polymorphism and clinical predictors of severity of disease in Î²â€‹thalassemia intermedia. Pediatric Blood and Cancer, 2011, 57, 1025-1028.	1.5	15
20	MTHFR promoter hypermethylation may lead to congenital heart defects in Down syndrome. Intractable and Rare Diseases Research, 2017, 6, 295-298.	0.9	15
21	SMN2-deletion in childhood-onset spinal muscular atrophy. American Journal of Medical Genetics Part A, 2001, 101, 198-202.	2.4	14
22	Chronic lymphocytic leukemia in India-A clinico-hematological profile. Hematology, 2007, 12, 229-233.	1.5	14
23	Diagnosis of Down Syndrome and Detection of Origin of Nondisjunction by Short Tandem Repeat Analysis. Genetic Testing and Molecular Biomarkers, 2010, 14, 489-491.	0.7	14
24	Seizure as the Presenting Manifestation in Griscelli Syndrome Type 2. Pediatric Neurology, 2015, 52, 535-538.	2.1	14
25	Zoledronate for Osteogenesis imperfecta: evaluation of safety profile in children. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 947-952.	0.9	14
26	Cutaneous Rosaiâ€Dorfman Disease: Presenting as Massive Bilateral Eyelid Swelling. Pediatric Dermatology, 2009, 26, 633-635.	0.9	13
27	A Newborn with Acanthosis Nigricans: Can It Be Crouzon Syndrome with Acanthosis Nigricans?. Pediatric Dermatology, 2010, 27, 43-47.	0.9	13
28	Proteus syndrome: Clinical profile of six patients and review of literature. Indian Journal of Human Genetics, 2013, 19, 202.	0.7	13
29	Hairy cell leukemia: Clinical, pathological and ultrastructural findings in Asian-Indians. Indian Journal of Cancer, 2008, 45, 41.	0.2	13
30	HFE mutation H63D predicts risk of iron over load in thalassemia intermedia irrespective of blood transfusions. Indian Journal of Pathology and Microbiology, 2007, 50, 82-5.	0.2	13
31	Hb Q India: Is it always benign?. American Journal of Hematology, 2005, 78, 245-246.	4.1	12
32	Pediatric disorders of sex development. Indian Journal of Pediatrics, 2009, 76, 956-958.	0.8	12
33	Multiplex Quantitative Fluorescent Polymerase Chain Reaction for Detection of Aneuploidies. Genetic Testing and Molecular Biomarkers, 2012, 16, 624-627.	0.7	12
34	Novel mutation in a family with WNT1 -related osteoporosis. European Journal of Medical Genetics, 2018, 61, 369-371.	1.3	11
35	Down syndrome with moyamoya disease: A case series. Journal of Pediatric Neurosciences, 2018, 13, 201.	0.3	11
36	Severe Early Onset Obesity due to a Novel Missense Mutation in Exon 3 of the Leptin Gene in an Infant from Northwest India. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018, 10, 274-278.	0.9	11

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37	Effect of wheat grass therapy on transfusion requirement in Î²-thalassemia major. Indian Journal of Pediatrics, 2009, 76, 375-376.	0.8	10
38	Spontaneous Hematomyelia in a Child With Hemophilia A. Journal of Pediatric Hematology/Oncology, 2009, 31, 766-767.	0.6	10
39	GENETIC HETEROGENEITY OF BETA GLOBIN MUTATIONS AMONG ASIAN-INDIANS AND IMPORTANCE IN GENETIC COUNSELLING AND DIAGNOSIS. Mediterranean Journal of Hematology and Infectious Diseases, 2013, 5, e2013003.	1.3	10
40	Common queries in thalassemia care. Indian Pediatrics, 2006, 43, 513-8.	0.4	10
41	High frequency of deletional ?-thalassemia in ?-thalassemia trait: Implications for genetic counseling. American Journal of Hematology, 2004, 76, 297-299.	4.1	9
42	Jaundice and alpha gene triplication in beta-thalassemia: Association or causation?. Hematology, 2006, 11, 109-112.	1.5	8
43	Macrocephalyâ€“capillary malformation syndrome: Three new cases. Journal of the Neurological Sciences, 2012, 313, 178-181.	0.6	8
44	KLHL40 mutation associated with severe nemaline myopathy, fetal akinesia, and cleft palate. Journal of Pediatric Neurosciences, 2019, 14, 222.	0.3	8
45	Molecular characterization of thalassemia intermedia in Indians. Haematologica, 2006, 91, 1279-80.	3.5	8
46	Spectrum of anemia in pregnant Indian women and importance of antenatal screening. Indian Journal of Pathology and Microbiology, 2006, 49, 373-5.	0.2	8
47	Mental retardation, ptosis and polydactyly: a new autosomal recessive syndrome?. Clinical Dysmorphology, 2002, 11, 289-292.	0.3	7
48	The expanding spectrum of thalassemia intermedia. Hematology, 2009, 14, 311-314.	1.5	7
49	Novel mutation in the CHST14 gene causing musculocontractural type of Ehlers-Danlos syndrome. BMJ Case Reports, 2018, 2018, bcr-2018-226165.	0.5	7
50	Knowledge and Attitudes Towards Haemophilia: The Family Side and Role of Haemophilia Societies. Public Health Genomics, 2003, 6, 120-122.	1.0	6
51	Long-term response to deferiprone therapy in Asian Indians. Annals of Hematology, 2010, 89, 135-140.	1.8	6
52	Thyroid dysfunction in Indian children with down syndrome. Indian Pediatrics, 2014, 51, 751-753.	0.4	6
53	Niemann-Pick Disease: An Underdiagnosed Lysosomal Storage Disorder. Case Reports in Genetics, 2019, 2019, 1-5.	0.2	6
54	Imatinib mesylate: A designer drug. Journal of the Association of Physicians of India, The, 2006, 54, 203-6.	0.0	6

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55	Neurofibromatosis Type 1 With Intracranial Hemorrhage and Horseshoe Kidney. Pediatric Neurology, 2008, 39, 295-297.	2.1	5
56	Congenital scoliosis, supernumerary nipples and spina bifida occulta. Clinical Dysmorphology, 2008, 17, 215-218.	0.3	5
57	Zolendronate in Osteogenesis Imperfecta. Journal of Pediatric Endocrinology and Metabolism, 2009, 22, 763.	0.9	5
58	Ascariasis-associated worm encephalopathy in a young child. Tropical Doctor, 2009, 39, 113-114.	0.5	5
59	STR markers for detecting heterogeneity in Indian population. Molecular Biology Reports, 2012, 39, 461-465.	2.3	5
60	Overlapping phenotypes in OFD type II and OFD type VI. Clinical Dysmorphology, 2013, 22, 109-114.	0.3	5
61	Managing syndromic congenital ichthyosis at a tertiary care instituteâ€"Genotypeâ€phenotype correlations, and novel treatments. Dermatologic Therapy, 2020, 33, e13816.	1.7	5
62	Chungâ€"Jansen Syndrome with obesity. Obesity Research and Clinical Practice, 2021, 15, 303-305.	1.8	5
63	Seckel syndrome with chromosomal 18 deletion. Indian Journal of Pediatrics, 2009, 76, 1270-1271.	0.8	4
64	A novel beta-globin mutation (HBB:c.107A>G; or codon 35ÂÎ² (Aâ†'G)) at alphaâ€"beta chain interfaces. Annals of Hematology, 2009, 88, 1269-1271.	1.8	4
65	Efficacy of Deferasirox in North Indian Î²-Thalassemia Major Patients. Journal of Pediatric Hematology/Oncology, 2012, 34, 51-53.	0.6	4
66	Zellweger syndrome: prenatal and postnatal growth failure with epiphyseal stippling. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 185-8.	0.9	4
67	Methylmalonic Acidemia with Novel MUT Gene Mutations. Case Reports in Genetics, 2017, 2017, 1-2.	0.2	4
68	Warty Fingers and Toes in a Child With Congenital Lymphedema. JAMA Dermatology, 2018, 154, 849.	4.1	4
69	Hunter syndrome with persistent thrombocytopenia. BMJ Case Reports, 2019, 12, e226518.	0.5	4
70	Prevalence of Filaggrin Gene R501X Mutation in Indian Children with Allergic Diseases. Indian Journal of Pediatrics, 2020, 87, 587-590.	0.8	4
71	Identification of microdeletion and microduplication syndromes by chromosomal microarray in patients with intellectual disability with dysmorphism. Neurology India, 2018, 66, 1370.	0.4	4
72	Distal arthrogryposis syndrome. Indian Journal of Human Genetics, 2008, 14, 67.	0.7	4

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73	<scp>Wolfâ€“Hirschhorn</scp> syndrome: A case series from India. American Journal of Medical Genetics, Part A, 2020, 182, 3048-3051.	1.2	3
74	Association of VEGF and p53 Polymorphisms and Spiral Artery Remodeling in Recurrent Pregnancy Loss: A Systematic Review and Meta-Analysis. Thrombosis and Haemostasis, 2021, , .	3.4	3
75	Components of IGF-axis in growth disorders: a systematic review and patent landscape report. Endocrine, 2022, , 1.	2.3	3
76	Fulminant candida infection in an infant with acrodermatitis enteropathica. Indian Journal of Pediatrics, 2009, 76, 941-942.	0.8	2
77	MTRR gene variants may predispose to the risk of Congenital Heart Disease in Down syndrome patients of Indian origin. Egyptian Journal of Medical Human Genetics, 2017, 18, 61-66.	1.0	2
78	Phenotypic heterogeneity of kyphoscoliosis with vertebral and rib defects: a case series. Clinical Dysmorphology, 2019, 28, 101-111.	0.3	2
79	Sotos syndrome in two children from India. American Journal of Medical Genetics, Part A, 2020, 182, 2181-2183.	1.2	2
80	Clinical profile of symptomatic congenital cytomegalovirus infection: cases from a tertiary hospital in north India. Tropical Doctor, 2020, 50, 282-284.	0.5	2
81	Simple virilising congenital adrenal hyperplasia in monozygotic twins: A rare report and review of previous cases. Pediatric Endocrinology, Diabetes and Metabolism, 2020, 26, 58-62.	0.7	2
82	<scp>COFS</scp> type 3 in an Indian family with antenatally detected arthrogryposis. American Journal of Medical Genetics, Part A, 2021, 185, 631-635.	1.2	2
83	Bardet-Biedl syndrome presenting with laryngeal web and bifid epiglottis. BMJ Case Reports, 2021, 14, e236325.	0.5	2
84	Single gene variants causing deafness in Asian Indians. Journal of Genetics, 2021, 100, 1.	0.7	2
85	Achondroplasiaâ€"First Report from India of a Rare FGFR3 Gene Variant. Laboratory Medicine, 2021, 52, 499-502.	1.2	2
86	Genetic Fingerprinting for Human Diseases: Applications and Implications. , 2018, , 141-150.		2
87	Characterization of the CYP21A2 Gene Mutations in Children with Classic Congenital Adrenal Hyperplasia. Indian Journal of Pediatrics, 2024, 91, 137-142.	0.8	2
88	Therapy-related acute promyelocytic leukemia after treatment of carcinoma breast--a case report. Indian Journal of Pathology and Microbiology, 2006, 49, 251-4.	0.2	2
89	Cytochemical, immunophenotypic and ultrastructural characterization of acute leukemias: A prospective study of fifty cases. Hematology, 2006, 11, 147-151.	1.5	1
90	Child with Mongolian spots and dysostosis multiplex. Indian Journal of Human Genetics, 2009, 15, 38.	0.7	1

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91	Fibular hemimelia with polysyndactyly: a case report. Clinical Dysmorphology, 2010, 19, 88-90.	0.3	1
92	Nonâ€invasive prenatal diagnosis: improved detection rates. Prenatal Diagnosis, 2011, 31, 221-221.	2.3	1
93	Hypercortisolism and hypothyroidism in an infant with Smith-Lemli-Opitz syndrome. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 1001-5.	0.9	1
94	Lissencephaly presenting with congenital hypothyroidism. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 1175-7.	0.9	1
95	Peroxisomal Disorders: Experience from a Genetic Center in North India. Journal of Pediatric Neurology, 2019, 17, 065-070.	0.2	1
96	Hepatomegaly with neutropenia: a girl with glycogen storage disease Ib. BMJ Case Reports, 2019, 12, e230660.	0.5	1
97	Congenital Cytomegalovirus Infection Masquerading as Antenatal Ventriculomegaly With Intraventricular Hemorrhage in a Term Neonate. Neurohospitalist, The, 2020, 10, 55-57.	0.8	1
98	Over-Representation of Recessive Osteogenesis Imperfecta in Asian Indian Children. Journal of Pediatric Genetics, 2022, 11, 081-086.	0.7	1
99	Ten-year use of recombinant parathyroid hormone for the treatment of hypoparathyroidism in a boy with partial Jacobsen syndrome. Pediatric Endocrinology, Diabetes and Metabolism, 2021, 27, 57-61.	0.7	1
100	Gas Chromatography Mass Spectrometry Aided Diagnosis of Glutathione Synthetase Deficiency. Laboratory Medicine, 2022, 53, e59-e61.	1.2	1
101	Clinical and Radiological Characterisation of Patients with Mucopolysaccharidosis in a Genetic Clinic. Journal of Inborn Errors of Metabolism and Screening, 0, 9, .	0.3	1
102	Percentile Charts for Body Mass Index of Indian Down Syndrome Children. Journal of Pediatric Genetics, 0, , .	0.7	1
103	Novel variation in <scp>ANTXR2</scp> gene causing hyaline fibromatosis syndrome: A report from India. Congenital Anomalies (discontinued), 2021, 61, 140-141.	0.6	1
104	Radial aplasia with oligodactyly. Indian Journal of Human Genetics, 2008, 14, 29.	0.7	1
105	Overlapping Phenotypes in Osteopetrosis and Pycnodysostosis in Asian-Indians. Case Reports in Genetics, 2021, 2021, 1-6.	0.2	1
106	Single gene variants causing deafness in Asian Indians. Journal of Genetics, 2021, 100, .	0.7	1
107	How Experts Make a Call: Copy Number Variation Analysis in Unusual/Rare Case Scenarios. Neurology India, 2022, 70, 148.	0.4	1
108	Reply to correspondence from Ogino et al.??comment on SMN2 deletion in childhood-onset spinal muscular atrophy?. American Journal of Medical Genetics Part A, 2002, 109, 245-245.	2.4	0

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109	Reply to correspondence from Cobben and de Visser- ?SMN2-deletion in childhood-onset spinal muscular atrophy?. American Journal of Medical Genetics Part A, 2002, 109, 247-247.	2.4	0
110	Prenatal diagnosis of Sheldon Hall syndrome. Prenatal Diagnosis, 2009, 29, 897-898.	2.3	0
111	Thalassemia Intermedia With Iron Deficiency. Journal of Pediatric Hematology/Oncology, 2009, 31, 989.	0.6	0
112	The first case report of a patient with coexisting hemophilia B and Down syndrome. Blood Research, 2017, 52, 75.	1.3	0
113	Indian child with novel variant in OFD1 gene. American Journal of Medical Genetics, Part A, 2020, 182, 2236-2238.	1.2	0
114	Reply to the Letter: â€œDiagnosis of Peroxisomal Disordersâ€• Journal of Pediatric Neurology, 2020, 18, 054-054.	0.2	0
115	Study of Prothrombotic Gene Variations Associated with the Risk of Development of Thrombosis in Patients with Down Syndrome. Indian Journal of Hematology and Blood Transfusion, 2021, 37, 507-508.	0.6	0
116	Recurrent Apnea in an Infant â€" Think Beyond the Usual. Indian Journal of Pediatrics, 2021, 88, 391-391.	0.8	0
117	Xâ€linked frontometaphyseal dysplasia with severe scoliosis and spinal cord compromise in an Indian boy. American Journal of Medical Genetics, Part A, 2021, 185, 1550-1553.	1.2	0
118	Short Stature Syndromes: Case Series from India. Journal of Pediatric Genetics, 0, , .	0.7	0
119	Rare chromosomal aberrations detected in children with multiple congenital anomalies. Clinical Dysmorphology, 2021, Publish Ahead of Print, 125-129.	0.3	0
120	Chromosome 1p36 Deletion Syndrome: Four Patients with Variable Presentations. Journal of Pediatric Genetics, 2023, 12, 342-347.	0.7	0
121	Clinical Profile of Indian Children with Down Syndrome. Journal of Pediatric Genetics, 0, , .	0.7	0
122	Hypertensive Emergency with Medullary and Spinal Hemorrhage in Turner Syndrome. Indian Journal of Pediatrics, 2021, 88, 929-930.	0.8	0
123	Use of Transmission Electron Microscopy in Diagnosis of Acute Leukemias: A Prospective Study of Fifty Cases.. Blood, 2005, 106, 4509-4509.	1.4	0
124	Hydroxyurea in Children with Beta Thalassemia Intermedia.. Blood, 2008, 112, 1879-1879.	1.4	0
125	Acquired syndactyly in epidermolysis bullosa dystrophica. BMJ Case Reports, 2009, 2009, bcr0420091785-bcr0420091785.	0.5	0
126	Hemifacial microsomia with pulmonary hypoplasia. BMJ Case Reports, 2010, 2010, bcr0420091759-bcr0420091759.	0.5	0

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127	A case of short stature with anterior vertebral beaking. Indian Journal of Human Genetics, 2011, 17, 28.	0.7	0
128	Severe liver dysfunction in an infant with cystic fibrosis masquerading as metabolic liver disease. Indian Journal of Pathology and Microbiology, 2016, 59, 339.	0.2	0
129	Growth Pattern and Use of Inter-pupillary Distance in the Detection of Ocular Hypertelorism and Hypotelorism in Indian Down Syndrome Children. Journal of Pediatric Genetics, 0, , .	0.7	0
130	Genetic Defects in Children with Cardiac Anomalies/Malformations: Noonan and CFC Syndromes. Journal of Pediatric Genetics, 0, , .	0.7	0
131	Case Studies of Two Classical Imprinting Growth Disorders: Silverâ€“Russell and Beckwithâ€“Wiedemann Syndromes. Journal of Pediatric Genetics, 0, , .	0.7	0
132	Anti-epileptic drug therapy: an overview of foetal effects. Journal of the Indian Medical Association, 2011, 109, 108-10.	0.2	0
133	Clinical and Molecular Heterogeneity of Silver Russell Syndrome and Therapeutic Challenges: A Systematic Review. Current Pediatric Reviews, 2022, 18, .	0.8	0
134	It is in the face - Have a relook!. Neurology India, 2020, 68, 1244.	0.4	0