

# Inusha Panigrahi

## List of Publications by Year in descending order

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134  
papers

1,232  
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516710

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477307

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138  
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138  
docs citations

138  
times ranked

1971  
citing authors

#	ARTICLE	IF	CITATIONS
1	Characterization of the CYP21A2 Gene Mutations in Children with Classic Congenital Adrenal Hyperplasia. Indian Journal of Pediatrics, 2024, 91, 137-142.	0.8	2
2	Chromosome 1p36 Deletion Syndrome: Four Patients with Variable Presentations. Journal of Pediatric Genetics, 2023, 12, 342-347.	0.7	0
3	Over-Representation of Recessive Osteogenesis Imperfecta in Asian Indian Children. Journal of Pediatric Genetics, 2022, 11, 081-086.	0.7	1
4	Gas Chromatography Mass Spectrometry Aided Diagnosis of Glutathione Synthetase Deficiency. Laboratory Medicine, 2022, 53, e59-e61.	1.2	1
5	Clinical and Molecular Heterogeneity of Silver Russell Syndrome and Therapeutic Challenges: A Systematic Review. Current Pediatric Reviews, 2022, 18, .	0.8	0
6	How Experts Make a Call: Copy Number Variation Analysis in Unusual/Rare Case Scenarios. Neurology India, 2022, 70, 148.	0.4	1
7	Components of IGF-axis in growth disorders: a systematic review and patent landscape report. Endocrine, 2022, , 1.	2.3	3
8	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
9	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
10	<sc>COFS</sc> type 3 in an Indian family with antenatally detected arthrogyposis. American Journal of Medical Genetics, Part A, 2021, 185, 631-635.	1.2	2
11	Bardet-Biedl syndrome presenting with laryngeal web and bifid epiglottis. BMJ Case Reports, 2021, 14, e236325.	0.5	2
12	Recurrent Apnea in an Infant â€” Think Beyond the Usual. Indian Journal of Pediatrics, 2021, 88, 391-391.	0.8	0
13	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
14	Novel variation in <sc><i>ANTXR2</i></sc> gene causing hyaline fibromatosis syndrome: A report from India. Congenital Anomalies (discontinued), 2021, 61, 140-141.	0.6	1
15	Rare chromosomal aberrations detected in children with multiple congenital anomalies. Clinical Dysmorphology, 2021, Publish Ahead of Print, 125-129.	0.3	0
16	Single gene variants causing deafness in Asian Indians. Journal of Genetics, 2021, 100, 1.	0.7	2
17	Chungâ€Jansen Syndrome with obesity. Obesity Research and Clinical Practice, 2021, 15, 303-305.	1.8	5
18	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

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19	Hypertensive Emergency with Medullary and Spinal Hemorrhage in Turner Syndrome. Indian Journal of Pediatrics, 2021, 88, 929-930.	0.8	0
20	Achondroplasiaâ€”First Report from India of a Rare <i>FGFR3</i> Gene Variant. Laboratory Medicine, 2021, 52, 499-502.	1.2	2
21	Overlapping Phenotypes in Osteopetrosis and Pycnodysostosis in Asian-Indians. Case Reports in Genetics, 2021, 2021, 1-6.	0.2	1
22	Single gene variants causing deafness in Asian Indians. Journal of Genetics, 2021, 100, .	0.7	1
23	Congenital Cytomegalovirus Infection Masquerading as Antenatal Ventriculomegaly With Intraventricular Hemorrhage in a Term Neonate. Neurohospitalist, The, 2020, 10, 55-57.	0.8	1
24	Sotos syndrome in two children from India. American Journal of Medical Genetics, Part A, 2020, 182, 2181-2183.	1.2	2
25	Indian child with novel variant in OFD1 gene. American Journal of Medical Genetics, Part A, 2020, 182, 2236-2238.	1.2	0
26	<scp>Wolfâ€™s</scp> Hirschhorn</i> syndrome: A case series from India. American Journal of Medical Genetics, Part A, 2020, 182, 3048-3051.	1.2	3
27	Clinical profile of symptomatic congenital cytomegalovirus infection: cases from a tertiary hospital in north India. Tropical Doctor, 2020, 50, 282-284.	0.5	2
28	Managing syndromic congenital ichthyosis at a tertiary care instituteâ€™”Genotypeâ€™phenotype correlations, and novel treatments. Dermatologic Therapy, 2020, 33, e13816.	1.7	5
29	Prevalence of Filaggrin Gene R501X Mutation in Indian Children with Allergic Diseases. Indian Journal of Pediatrics, 2020, 87, 587-590.	0.8	4
30	Reply to the Letter: â€™Diagnosis of Peroxisomal Disordersâ€™. Journal of Pediatric Neurology, 2020, 18, 054-054.	0.2	0
31	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
32	It is in the face - Have a relook!. Neurology India, 2020, 68, 1244.	0.4	0
33	Peroxisomal Disorders: Experience from a Genetic Center in North India. Journal of Pediatric Neurology, 2019, 17, 065-070.	0.2	1
34	Hepatomegaly with neutropenia: a girl with glycogen storage disease Ib. BMJ Case Reports, 2019, 12, e230660.	0.5	1
35	Niemann-Pick Disease: An Underdiagnosed Lysosomal Storage Disorder. Case Reports in Genetics, 2019, 2019, 1-5.	0.2	6
36	Hunter syndrome with persistent thrombocytopenia. BMJ Case Reports, 2019, 12, e226518.	0.5	4

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37	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
38	Phenotypic heterogeneity of kyphoscoliosis with vertebral and rib defects: a case series. Clinical Dysmorphology, 2019, 28, 101-111.	0.3	2
39	KLHL40 mutation associated with severe nemaline myopathy, fetal akinesia, and cleft palate. Journal of Pediatric Neurosciences, 2019, 14, 222.	0.3	8
40	Novel mutation in a family with WNT1 -related osteoporosis. European Journal of Medical Genetics, 2018, 61, 369-371.	1.3	11
41	Warty Fingers and Toes in a Child With Congenital Lymphedema. JAMA Dermatology, 2018, 154, 849.	4.1	4
42	CRELD1 gene variants and atrioventricular septal defects in Down syndrome. Gene, 2018, 641, 180-185.	2.2	20
43	Genetic Fingerprinting for Human Diseases: Applications and Implications. , 2018, , 141-150.		2
44	Novel mutation in the <i>CHST14</i> gene causing musculocontractural type of Ehlers-Danlos syndrome. BMJ Case Reports, 2018, 2018, bcr-2018-226165.	0.5	7
45	Identification of microdeletion and microduplication syndromes by chromosomal microarray in patients with intellectual disability with dysmorphism. Neurology India, 2018, 66, 1370.	0.4	4
46	Down syndrome with moyamoya disease: A case series. Journal of Pediatric Neurosciences, 2018, 13, 201.	0.3	11
47	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
48	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
49	Fifteen years of research on oral“facial“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	3.2	85
50	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
51	Methylmalonic Acidemia with Novel<i> MUT</i> Gene Mutations. Case Reports in Genetics, 2017, 2017, 1-2.	0.2	4
52	MTHFR promoter hypermethylation may lead to congenital heart defects in Down syndrome. Intractable and Rare Diseases Research, 2017, 6, 295-298.	0.9	15
53	The first case report of a patient with coexisting hemophilia B and Down syndrome. Blood Research, 2017, 52, 75.	1.3	0
54	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. Nature Genetics, 2016, 48, 648-656.	21.4	119

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55	Zoledronate for <i>Osteogenesis imperfecta</i> : evaluation of safety profile in children. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016, 29, 947-952.	0.9	14
56	Primordial dwarfism: overview of clinical and genetic aspects. <i>Molecular Genetics and Genomics</i> , 2016, 291, 1-15.	2.1	69
57	Severe liver dysfunction in an infant with cystic fibrosis masquerading as metabolic liver disease. <i>Indian Journal of Pathology and Microbiology</i> , 2016, 59, 339.	0.2	0
58	Seizure as the Presenting Manifestation in Griscelli Syndrome Type 2. <i>Pediatric Neurology</i> , 2015, 52, 535-538.	2.1	14
59	Zellweger syndrome: prenatal and postnatal growth failure with epiphyseal stippling. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014, 27, 185-8.	0.9	4
60	Thyroid dysfunction in Indian children with down syndrome. <i>Indian Pediatrics</i> , 2014, 51, 751-753.	0.4	6
61	The Effect of Prophylactic Antipyretic Administration on Post-Vaccination Adverse Reactions and Antibody Response in Children: A Systematic Review. <i>PLoS ONE</i> , 2014, 9, e106629.	2.5	48
62	GENETIC HETEROGENEITY OF BETA GLOBIN MUTATIONS AMONG ASIAN-INDIANS AND IMPORTANCE IN GENETIC COUNSELLING AND DIAGNOSIS. <i>Mediterranean Journal of Hematology and Infectious Diseases</i> , 2013, 5, e2013003.	1.3	10
63	Proteus syndrome: Clinical profile of six patients and review of literature. <i>Indian Journal of Human Genetics</i> , 2013, 19, 202.	0.7	13
64	Overlapping phenotypes in OFD type II and OFD type VI. <i>Clinical Dysmorphology</i> , 2013, 22, 109-114.	0.3	5
65	Lissencephaly presenting with congenital hypothyroidism. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2013, 26, 1175-7.	0.9	1
66	Rubinstein-taybi syndrome: Clinical profile of 11 patients and review of literature. <i>Indian Journal of Human Genetics</i> , 2012, 18, 161.	0.7	18
67	Multiplex Quantitative Fluorescent Polymerase Chain Reaction for Detection of Aneuploidies. <i>Genetic Testing and Molecular Biomarkers</i> , 2012, 16, 624-627.	0.7	12
68	Hypercortisolism and hypothyroidism in an infant with Smith-Lemli-Opitz syndrome. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2012, 25, 1001-5.	0.9	1
69	Efficacy of Deferasirox in North Indian $\beta^2$ -Thalassemia Major Patients. <i>Journal of Pediatric Hematology/Oncology</i> , 2012, 34, 51-53.	0.6	4
70	Maternal serum second trimester screening for chromosomal disorders and neural tube defects in a government hospital of North India. <i>Prenatal Diagnosis</i> , 2012, 32, 1192-1196.	2.3	17
71	Macrocephalyâ€“capillary malformation syndrome: Three new cases. <i>Journal of the Neurological Sciences</i> , 2012, 313, 178-181.	0.6	8
72	Sickle Cell Anemiaâ€“Molecular Diagnosis and Prenatal Counseling: SGPGI Experience. <i>Indian Journal of Pediatrics</i> , 2012, 79, 68-74.	0.8	17

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73	STR markers for detecting heterogeneity in Indian population. <i>Molecular Biology Reports</i> , 2012, 39, 461-465.	2.3	5
74	Xmn1 <sup>G</sup> polymorphism and clinical predictors of severity of disease in $\beta^2$ -thalassemia intermedia. <i>Pediatric Blood and Cancer</i> , 2011, 57, 1025-1028.	1.5	15
75	Non-invasive prenatal diagnosis: improved detection rates. <i>Prenatal Diagnosis</i> , 2011, 31, 221-221.	2.3	1
76	Craniosynostosis genetics: The mystery unfolds. <i>Indian Journal of Human Genetics</i> , 2011, 17, 48.	0.7	37
77	A case of short stature with anterior vertebral beaking. <i>Indian Journal of Human Genetics</i> , 2011, 17, 28.	0.7	0
78	Anti-epileptic drug therapy: an overview of foetal effects. <i>Journal of the Indian Medical Association</i> , 2011, 109, 108-10.	0.2	0
79	Long-term response to deferiprone therapy in Asian Indians. <i>Annals of Hematology</i> , 2010, 89, 135-140.	1.8	6
80	Response to zoledronic acid in children with type III osteogenesis imperfecta. <i>Journal of Bone and Mineral Metabolism</i> , 2010, 28, 451-455.	2.7	32
81	Evaluation of the genetic basis of phenotypic heterogeneity in north Indian patients with Thalassemia major. <i>European Journal of Haematology</i> , 2010, 84, 531-537.	2.2	20
82	A Newborn with Acanthosis Nigricans: Can It Be Crouzon Syndrome with Acanthosis Nigricans?. <i>Pediatric Dermatology</i> , 2010, 27, 43-47.	0.9	13
83	Fibular hemimelia with polysyndactyly: a case report. <i>Clinical Dysmorphology</i> , 2010, 19, 88-90.	0.3	1
84	Diagnosis of Down Syndrome and Detection of Origin of Nondisjunction by Short Tandem Repeat Analysis. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, 14, 489-491.	0.7	14
85	Hemifacial microsomia with pulmonary hypoplasia. <i>BMJ Case Reports</i> , 2010, 2010, bcr0420091759-bcr0420091759.	0.5	0
86	Zoledronate in Osteogenesis Imperfecta. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2009, 22, 763.	0.9	5
87	Child with Mongolian spots and dysostosis multiplex. <i>Indian Journal of Human Genetics</i> , 2009, 15, 38.	0.7	1
88	Ascariasis-associated worm encephalopathy in a young child. <i>Tropical Doctor</i> , 2009, 39, 113-114.	0.5	5
89	Effect of wheat grass therapy on transfusion requirement in $\beta^2$ -thalassemia major. <i>Indian Journal of Pediatrics</i> , 2009, 76, 375-376.	0.8	10
90	Fulminant candida infection in an infant with acrodermatitis enteropathica. <i>Indian Journal of Pediatrics</i> , 2009, 76, 941-942.	0.8	2

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91	Pediatric disorders of sex development. Indian Journal of Pediatrics, 2009, 76, 956-958.	0.8	12
92	Seckel syndrome with chromosomal 18 deletion. Indian Journal of Pediatrics, 2009, 76, 1270-1271.	0.8	4
93	A novel beta-globin mutation (HBB:c.107A>G; or codon 35 <sup>2</sup> (A <sup>+</sup> G)) at alpha <sup>+</sup> beta chain interfaces. Annals of Hematology, 2009, 88, 1269-1271.	1.8	4
94	Prenatal diagnosis of Sheldon Hall syndrome. Prenatal Diagnosis, 2009, 29, 897-898.	2.3	0
95	Cutaneous Rosai <sup>+</sup> Dorfman Disease: Presenting as Massive Bilateral Eyelid Swelling. Pediatric Dermatology, 2009, 26, 633-635.	0.9	13
96	Spontaneous Hematomyelia in a Child With Hemophilia A. Journal of Pediatric Hematology/Oncology, 2009, 31, 766-767.	0.6	10
97	Thalassemia Intermedia With Iron Deficiency. Journal of Pediatric Hematology/Oncology, 2009, 31, 989.	0.6	0
98	The expanding spectrum of thalassemia intermedia. Hematology, 2009, 14, 311-314.	1.5	7
99	Acquired syndactyly in epidermolysis bullosa dystrophica. BMJ Case Reports, 2009, 2009, bcr0420091785-bcr0420091785.	0.5	0
100	Neurofibromatosis Type 1 With Intracranial Hemorrhage and Horseshoe Kidney. Pediatric Neurology, 2008, 39, 295-297.	2.1	5
101	Urinary Malondialdehyde Levels in Newborns following Delivery Room Resuscitation. Neonatology, 2008, 94, 96-99.	2.0	17
102	Genetic determinants of phenotype in beta-thalassemia. Hematology, 2008, 13, 247-252.	1.5	25
103	Congenital scoliosis, supernumerary nipples and spina bifida occulta. Clinical Dysmorphology, 2008, 17, 215-218.	0.3	5
104	Hairy cell leukemia: Clinical, pathological and ultrastructural findings in Asian-Indians. Indian Journal of Cancer, 2008, 45, 41.	0.2	13
105	Distal arthrogyposis syndrome. Indian Journal of Human Genetics, 2008, 14, 67.	0.7	4
106	Radial aplasia with oligodactyly. Indian Journal of Human Genetics, 2008, 14, 29.	0.7	1
107	Hydroxyurea in Children with Beta Thalassemia Intermedia.. Blood, 2008, 112, 1879-1879.	1.4	0
108	Chronic lymphocytic leukemia in India-A clinico-hematological profile. Hematology, 2007, 12, 229-233.	1.5	14

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109	Thromboembolic complications in $\beta^2$ -thalassemia: Beyond the horizon. <i>Thrombosis Research</i> , 2007, 120, 783-789.	1.7	35
110	Mutational spectrum of thalassemias in India. <i>Indian Journal of Human Genetics</i> , 2007, 13, 36.	0.7	32
111	HFE mutation H63D predicts risk of iron over load in thalassemia intermedia irrespective of blood transfusions. <i>Indian Journal of Pathology and Microbiology</i> , 2007, 50, 82-5.	0.2	13
112	Cytochemical, immunophenotypic and ultrastructural characterization of acute leukemias: A prospective study of fifty cases. <i>Hematology</i> , 2006, 11, 147-151.	1.5	1
113	Jaundice and alpha gene triplication in beta-thalassemia: Association or causation?. <i>Hematology</i> , 2006, 11, 109-112.	1.5	8
114	Role of MTHFR C677T polymorphism in ischemic stroke. <i>Neurology India</i> , 2006, 54, 48.	0.4	29
115	Imatinib mesylate: A designer drug. <i>Journal of the Association of Physicians of India</i> , The, 2006, 54, 203-6.	0.0	6
116	Common queries in thalassemia care. <i>Indian Pediatrics</i> , 2006, 43, 513-8.	0.4	10
117	Therapy-related acute promyelocytic leukemia after treatment of carcinoma breast—a case report. <i>Indian Journal of Pathology and Microbiology</i> , 2006, 49, 251-4.	0.2	2
118	Molecular characterization of thalassemia intermedia in Indians. <i>Haematologica</i> , 2006, 91, 1279-80.	3.5	8
119	Spectrum of anemia in pregnant Indian women and importance of antenatal screening. <i>Indian Journal of Pathology and Microbiology</i> , 2006, 49, 373-5.	0.2	8
120	Hb Q India: Is it always benign?. <i>American Journal of Hematology</i> , 2005, 78, 245-246.	4.1	12
121	Use of Transmission Electron Microscopy in Diagnosis of Acute Leukemias: A Prospective Study of Fifty Cases.. <i>Blood</i> , 2005, 106, 4509-4509.	1.4	0
122	High frequency of deletional $\beta$ -thalassemia in $\beta$ -thalassemia trait: Implications for genetic counseling. <i>American Journal of Hematology</i> , 2004, 76, 297-299.	4.1	9
123	Knowledge and Attitudes Towards Haemophilia: The Family Side and Role of Haemophilia Societies. <i>Public Health Genomics</i> , 2003, 6, 120-122.	1.0	6
124	Mental retardation, ptosis and polydactyly: a new autosomal recessive syndrome?. <i>Clinical Dysmorphology</i> , 2002, 11, 289-292.	0.3	7
125	Reply to correspondence from Ogino et al.??comment on SMN2 deletion in childhood-onset spinal muscular atrophy?. <i>American Journal of Medical Genetics Part A</i> , 2002, 109, 245-245.	2.4	0
126	Reply to correspondence from Cobben and de Visser- ?SMN2-deletion in childhood-onset spinal muscular atrophy?. <i>American Journal of Medical Genetics Part A</i> , 2002, 109, 247-247.	2.4	0



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127	SMN2-deletion in childhood-onset spinal muscular atrophy. American Journal of Medical Genetics Part A, 2001, 101, 198-202.	2.4	14
128	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
129	Percentile Charts for Body Mass Index of Indian Down Syndrome Children. Journal of Pediatric Genetics, 0, , .	0.7	1
130	Short Stature Syndromes: Case Series from India. Journal of Pediatric Genetics, 0, , .	0.7	0
131	Clinical Profile of Indian Children with Down Syndrome. Journal of Pediatric Genetics, 0, , .	0.7	0
132	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
133	Genetic Defects in Children with Cardiac Anomalies/Malformations: Noonan and CFC Syndromes. Journal of Pediatric Genetics, 0, , .	0.7	0
134	Case Studies of Two Classical Imprinting Growth Disorders: Silverâ€“Russell and Beckwithâ€“Wiedemann Syndromes. Journal of Pediatric Genetics, 0, , .	0.7	0