Ratna Dua Puri

List of Publications by Year
in descending order

41	280	1040056 9	1058476 14
papers	citations	h-index	g-index

42	42	42	504
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genotype-phenotype spectrum of 130 unrelated Indian families with Mucopolysaccharidosis type II. European Journal of Medical Genetics, 2022, 65, 104447.	1.3	3
2	Prenatal phenotypic spectrum of full trisomy 18 in an Indian cohort. American Journal of Medical Genetics, Part A, 2022, , .	1.2	1
3	Indian Undiagnosed Diseases Program (I-UDP) â€" The Unmet Need. Indian Pediatrics, 2022, 59, 198-200.	0.4	1
4	Indian Undiagnosed Diseases Program (I-UDP) - The Unmet Need.. Indian Pediatrics, 2022, , .	0.4	0
5	<scp> COASY </scp> related pontocerebellar hypoplasia type 12: A common Indian mutation with expansion of the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2022, , .	1.2	2
6	Lysosomal storage disorders: Novel and frequent pathogenic variants in a large cohort of Indian patients of Pompe, Fabry, Gaucher and Hurler disease. Clinical Biochemistry, 2021, 89, 14-37.	1.9	6
7	Late onset Pompe Disease in India â€" Beyond the Caucasian phenotype. Neuromuscular Disorders, 2021, 31, 431-441.	0.6	6
8	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
9	Challenges in Chronic Genetic Disorders: Lessons From the COVID-19 Pandemic. Indian Pediatrics, 2021, 58, 391-392.	0.4	4
10	Clinical and genetic spectrum of 104 Indian families with central nervous system white matter abnormalities. Clinical Genetics, 2021, 100, 542-550.	2.0	12
11	Levels of Lyso GL-1 in Gaucher and Lyso GL-3 in Fabry patients from India: Diagnostic aids for these lysosomal storage disorders. Clinica Chimica Acta, 2021, 521, 177-190.	1.1	0
12	Mutation and Phenotypic Spectrum of Patients With RASopathies. Indian Pediatrics, 2021, 58, 30-33.	0.4	1
13	Mutation and Phenotypic Spectrum of Patients With RASopathies. Indian Pediatrics, 2021, 58, 30-33.	0.4	1
14	Filaggrin Null-Mutation in Asthma in an Indian Cohort: One Link in a Polygenic Trait. Indian Journal of Pediatrics, 2020, 87, 583-584.	0.8	0
15	NGS-based expanded carrier screening for genetic disorders in North Indian population reveals unexpected results â€" a pilot study. BMC Medical Genetics, 2020, 21, 216.	2.1	9
16	Expanding the phenotypic and genotypic spectrum of Wiedemannâ€"Steiner syndrome: First patient from India. American Journal of Medical Genetics, Part A, 2020, 182, 953-956.	1.2	9
17	Sialidosis type II: Expansion of phenotypic spectrum and identification of a common mutation in seven patients. Molecular Genetics and Metabolism Reports, 2020, 22, 100561.	1.1	8
18	Mutation and Phenotypic Spectrum of Patients With RASopathies. Indian Pediatrics, 2020, , .	0.4	0

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19	Is Molecular Diagnosis Necessary for Children with Duchenne Muscular Dystrophy?. Indian Pediatrics, 2019, 56, 549-550.	0.4	0
20	Gaucher Disease in Fetus: The Usual and the Unusual Presentations in a Family. Journal of Fetal Medicine, 2019, 6, 147-150.	0.1	1
21	The first case of antenatal presentation in COG8â€congenital disorder of glycosylation with a novel splice site mutation and an extended phenotype. American Journal of Medical Genetics, Part A, 2019, 179, 480-485.	1.2	12
22	Prenatal presentation of a rare genetic disorder: a clinical, autopsy and molecular correlation. Autopsy and Case Reports, 2019, 9, e2019124.	0.6	6
23	Is Molecular Diagnosis Necessary for Children with Duchenne Muscular Dystrophy?. Indian Pediatrics, 2019, 56, 549-550.	0.4	0
24	Expanding the Phenotype of the Founder South Asian Mutation in the Nuclear Encoding Mitochondrial RMND1 Gene. Indian Journal of Pediatrics, 2018, 85, 87-92.	0.8	9
25	ALG9 Associated Gillessen-Kaesbachâ€“Nishimura Syndrome (GIKANIS): An Uncommon Aetiology of Enlarged Foetal Kidneys. Journal of Fetal Medicine, 2018, 05, 237-239.	0.1	1
26	Clinical heterogeneity and molecular profile of triple A syndrome: a study of seven cases. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 799-807.	0.9	13
27	Hotspots in PTPN11 gene among Indian children with Noonan syndrome. Indian Pediatrics, 2017, 54, 638-640.	0.4	5
28	Editorial: New Horizons in Genetic Diagnosis in Pediatric Practice: The Excitement and Challenges!. Indian Journal of Pediatrics, 2016, 83, 1131-1132.	0.8	1
29	Genetic Approach to Diagnosis of Intellectual Disability. Indian Journal of Pediatrics, 2016, 83, 1141-1149.	0.8	8
30	KCNQ1 mutations associated with Jervell and Langeâ€“Nielsen syndrome and autosomal recessive Romanoâ€“Ward syndrome in Indiaâ€”expanding the spectrum of long QT syndrome type 1. American Journal of Medical Genetics, Part A, 2016, 170, 1510-1519.	1.2	11
31	Is the diagnostic yield influenced by the indication for fetal autopsy?. American Journal of Medical Genetics, Part A, 2016, 170, 2119-2126.	1.2	3
32	Phenotype guided characterization and molecular analysis of Indian patients with long QT syndromes. Indian Pacing and Electrophysiology Journal, 2016, 16, 8-18.	0.6	11
33	Next Generation Sequencing in the Clinic. Indian Journal of Pediatrics, 2016, 83, 281-282.	0.8	3
34	Novel and recurrent mutations in WISP3 and an atypical phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 2481-2484.	1.2	21
35	Fetal Dysmorphology. Journal of Fetal Medicine, 2015, 2, 151-159.	0.1	2
36	Molecular Diagnosis of Hereditary Fructose Intolerance: Founder Mutation in a Community from India. JIMD Reports, 2014, 19, 85-93.	1.5	18

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37	Prenatal Diagnosis and Elucidation of a Novel Molecular Mechanism in Carpenter Syndrome. Journal of Fetal Medicine, 2014, 1, 89-93.	0.1	1
38	GALNS mutations in Indian patients with mucopolysaccharidosis IVA. American Journal of Medical Genetics, Part A, 2014, 164, 2793-2801.	1.2	31
39	The Role of Radiographs in Fetal Autopsy. Journal of Fetal Medicine, 2014, 1, 7-9.	0.1	2
40	Do parental perceptions and motivations towards genetic testing and prenatal diagnosis for deafness vary in different cultures?. American Journal of Medical Genetics, Part A, 2013, 161, 76-81.	1.2	18
41	Spectrum of severe skeletal dysplasias in North India. Indian Journal of Pediatrics, 2007, 74, 995-1002.	0.8	15