Jayesh Sheth

List of Publications by Year
in descending order

27	339	933447 10	888059 17
papers	citations	h-index	g-index

30	30	30	488
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Burden of Lysosomal Storage Disorders in India: Experience of 387 Affected Children from a Single Diagnostic Facility. JIMD Reports, 2013, 12, 51-63.	1.5	38
2	Identification of Novel Mutations in HEXA Gene in Children Affected with Tay Sachs Disease from India. PLoS ONE, 2012, 7, e39122.	2.5	37
3	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
4	A case of Raine syndrome presenting with facial dysmorphy and review of literature. BMC Medical Genetics, 2018, 19, 76.	2.1	26
5	GM2 gangliosidosis AB variant: novel mutation from India â€“ a case report with a review. BMC Pediatrics, 2016, 16, 88.	1.7	24
6	Novel mutations in the glucocerebrosidase gene of Indian patients with Gaucher disease. Journal of Human Genetics, 2014, 59, 223-228.	2.3	21
7	Pulmonary manifestations in Niemann-Pick type C disease with mutations in NPC2 gene: case report and review of literature. BMC Medical Genetics, 2017, 18, 5.	2.1	18
8	Lysosomal Storage Disorders in Nonimmune Hydrops Fetalis (NIHF): An Indian Experience. JIMD Reports, 2017, 35, 47-52.	1.5	15
9	Lysosomal storage disorders in Indian children with neuroregression attending a genetic center. Indian Pediatrics, 2015, 52, 1029-1033.	0.4	13
10	Prenatal screening of cytogenetic anomalies â€“ a Western Indian experience. BMC Pregnancy and Childbirth, 2015, 15, 90.	2.4	13
11	Biochemical and molecular characterization of adult patients with type I Gaucher disease and carrier frequency analysis of Leu444Pro - a common Gaucher disease mutation in India. BMC Medical Genetics, 2018, 19, 178.	2.1	12
12	Treatment for Lysosomal Storage Disorders. Current Pharmaceutical Design, 2020, 26, 5110-5118.	1.9	12
13	Expanding the spectrum of HEXA mutations in Indian patients with Tayâ€“Sachs disease. Molecular Genetics and Metabolism Reports, 2014, 1, 425-430.	1.1	10
14	Batten disease: biochemical and molecular characterization revealing novel PPT1 and TPP1 gene mutations in Indian patients. BMC Neurology, 2018, 18, 203.	1.8	10
15	Novel LINS1 missense mutation in a family with nonâ€syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2017, 173, 1041-1046.	1.2	9
16	Clinical Characteristics, Molecular Profile, and Outcomes in Indian Patients with Glutaric Aciduria Type 1. Journal of Pediatric Genetics, 2021, 10, 213-221.	0.7	7
17	Prenatal Diagnosis of Lysosomal Storage Disorders by Enzymes Study Using Chorionic Villus and Amniotic Fluid. Journal of Fetal Medicine, 2014, 1, 17-24.	0.1	6
18	Identification of novel variants in a large cohort of children with Tayâ€“Sachs disease: An initiative of a multicentric task force on lysosomal storage disorders by Government of India. Journal of Human Genetics, 2019, 64, 985-994.	2.3	5

#	Article	IF	Citations
19	Rare cause of Hemophagocytic Lymphohistiocytosis due to mutation in PRF1 and SH2D1A genes in two children â€“ a case report with a review. BMC Pediatrics, 2019, 19, 73.	1.7	5
20	Identification of Novel Mutations in FAH Gene and Prenatal Diagnosis of Tyrosinemia in Indian Family. Case Reports in Genetics, 2012, 2012, 1-4.	0.2	4
21	GM2 activator protein deficiency, mimic of Tay-Sachs disease. International Journal of Epilepsy, 2017, 04, 184-187.	0.5	4
22	Identification of deletion-duplication in HEXA gene in five children with Tay-Sachs disease from India. BMC Medical Genetics, 2018, 19, 109.	2.1	4
23	Clinical and molecular characterization of patients with gross hypotonia and impaired lower motor neuron function. Indian Pediatrics, 2013, 50, 591-593.	0.4	3
24	Pure interstitial dup(6)(q22.31q22.31) â€“ a case report. Italian Journal of Pediatrics, 2015, 41, 5.	2.6	3
25	Vitamin K Insufficiency in the Indian Population: Pilot Observational Epidemiology Study. JMIR Public Health and Surveillance, 2022, 8, e31941.	2.6	3
26	Genotype-phenotype spectrum of 130 unrelated Indian families with Mucopolysaccharidosis type II. European Journal of Medical Genetics, 2022, 65, 104447.	1.3	3
27	Novel mutation in the <scp>XPC</scp> gene: a case report of a patient with xeroderma pigmentosum. International Journal of Dermatology, 2015, 54, e487-91.	1.0	1