Matthew T Pastore

List of Publications by Year
in descending order

25 papers	1,430 citations	567281 15 h-index	580821 25 g-index
28 all docs	28 docs citations	28 times ranked	2497 citing authors

#	Article	IF	Citations
1	The prevalence of PTEN mutations in a clinical pediatric cohort with autism spectrum disorders, developmental delay, and macrocephaly. Genetics in Medicine, 2009, 11, 111-117.	2.4	251
2	Confirmation study of PTEN mutations among individuals with autism or developmental delays/mental retardation and macrocephaly. Autism Research, 2010, 3, 137-141.	3.8	218
3	Increasing knowledge ofPTEN germline mutations: Two additional patients with autism and macrocephaly. American Journal of Medical Genetics, Part A, 2007, 143A, 589-593.	1.2	150
4	A transgene carrying an A2G missense mutation in the SMN gene modulates phenotypic severity in mice with severe (type I) spinal muscular atrophy. Journal of Cell Biology, 2003, 160, 41-52.	5.2	140
5	Genetic testing in autism: how much is enough?. Genetics in Medicine, 2007, 9, 268-274.	2.4	97
6	De novo mutations in beta-catenin (CTNNB1) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. Human Genetics, 2015, 134, 97-109.	3.8	93
7	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	6.2	92
8	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
9	Coronary artery disease in a Werner syndromeâ€like form of progeria characterized by low levels of progerin, a splice variant of lamin A. American Journal of Medical Genetics, Part A, 2011, 155, 3002-3006.	1.2	55
10	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	5.1	32
11	Diagnostic Utility of Whole Exome Sequencing in the Neuromuscular Clinic. Neuropediatrics, 2019, 50, 096-102.	0.6	28
12	Variants in MED12L, encoding a subunit of the mediator kinase module, are responsible for intellectual disability associated with transcriptional defect. Genetics in Medicine, 2019, 21, 2713-2722.	2.4	28
13	SRD5A3â€CDG: Expanding the phenotype of a congenital disorder of glycosylation with emphasis on adult onset features. American Journal of Medical Genetics, Part A, 2016, 170, 3165-3171.	1.2	23
14	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	8.2	23
15	Neurodevelopmental disorders among individuals with duplication of 4p13 to 4p12 containing a GABAA receptor subunit gene cluster. European Journal of Human Genetics, 2014, 22, 105-109.	2.8	20
16	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. European Journal of Medical Genetics, 2013, 56, 301-308.	1.3	17
17	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
18	A tale of two deletions: A report of two novel 20p13â€‰â†’â€‰pter deletions. American Journal of Medical Genetics, Part A, 2010, 152A, 1000-1007.	1.2	13

#	Article	IF	Citations
19	MCPH1 deletion in a newborn with severe microcephaly and premature chromosome condensation. European Journal of Medical Genetics, 2013, 56, 609-613.	1.3	11
20	Unexpected detection of dystrophin gene deletions by array comparative genomic hybridization. American Journal of Medical Genetics, Part A, 2010, 152A, 2301-2307.	1.2	10
21	Role of CFTR mutation analysis in the diagnostic algorithm for cystic fibrosis. World Journal of Pediatrics, 2017, 13, 129-135.	1.8	10
22	Characterization of the renal phenotype in RMND1 â€related mitochondrial disease. Molecular Genetics & Genomic Medicine, 2019, 7, e973.	1.2	10
23	Trisomy 16p: A longitudinal profile and photo essay. American Journal of Medical Genetics, Part A, 2006, 140A, 174-179.	1.2	9
24	A Novel Exon Duplication of the Cystic Fibrosis Transmembrane Conductance Regulator in a Patient Presenting With Adult-Onset Recurrent Pancreatitis. Pancreas, 2011, 40, 773-777.	1.1	2
25	Transferring Exome Sequencing Data from Clinical Laboratories to Healthcare Providers: Lessons Learned at a Pediatric Hospital. Frontiers in Genetics, 2018, 9, 54.	2.3	2