Melissa A Parisi

List of Publications by Year
in descending order

19 papers	1,452 citations	516710 16 h-index	888059 17 g-index
19 all docs	19 docs citations	19 times ranked	1651 citing authors

#	Article	IF	Citations
1	The NPHP1 Gene Deletion Associated with Juvenile Nephronophthisis Is Present in a Subset of Individuals with Joubert Syndrome. American Journal of Human Genetics, 2004, 75, 82-91.	6.2	228
2	Molar tooth sign of the midbrain-hindbrain junction: Occurrence in multiple distinct syndromes. American Journal of Medical Genetics Part A, 2004, 125A, 125-134.	2.4	213
3	Human malformations of the midbrain and hindbrain: review and proposed classification scheme. Molecular Genetics and Metabolism, 2003, 80, 36-53.	1.1	205
4	Clinical and molecular features of Joubert syndrome and related disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2009, 151C, 326-340.	1.6	202
5	Molecular genetic findings and clinical correlations in 100 patients with Joubert syndrome and related disorders prospectively evaluated at a single center. Genetics in Medicine, 2017, 19, 875-882.	2.4	100
6	The molecular genetics of Joubert syndrome and related ciliopathies: The challenges of genetic and phenotypic heterogeneity. Translational Science of Rare Diseases, 2019, 4, 25-49.	1.5	79
7	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
8	Prospective Evaluation of Kidney Disease in Joubert Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2017, 12, 1962-1973.	4.5	56
9	Joubert syndrome: neuroimaging findings in 110 patients in correlation with cognitive function and genetic cause. Journal of Medical Genetics, 2017, 54, 521-529.	3.2	53
10	We don't know what we don't study: The case for research on medication effects in pregnancy. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 247-250.	1.6	43
11	Joubert Syndrome: Ophthalmological Findings in Correlation with Genotype and Hepatorenal Disease in 99 Patients Prospectively Evaluated at a Single Center. Ophthalmology, 2018, 125, 1937-1952.	5.2	43
12	Eye Movement Abnormalities in Joubert Syndrome. , 2009, 50, 4669.		38
13	Congenital diaphragmatic hernia and microtia in a newborn with mycophenolate mofetil (MMF) exposure: Phenocopy for Fryns syndrome or broad spectrum of teratogenic effects?. American Journal of Medical Genetics, Part A, 2009, 149A, 1237-1240.	1.2	32
14	Neuropsychological phenotypes of 76 individuals with Joubert syndrome evaluated at a single center. American Journal of Medical Genetics, Part A, 2017, 173, 1796-1812.	1.2	26
15	Compound heterozygous alterations in intraflagellar transport protein CLUAP1 in a child with a novel Joubert and oralâ€“facialâ€“digital overlap syndrome. Journal of Physical Education and Sports Management, 2017, 3, a001321.	1.2	23
16	Cerebral and cerebellar motor activation abnormalities in a subject with Joubert syndrome: functional magnetic resonance imaging (MRI) study. Journal of Child Neurology, 2004, 19, 214-8.	1.4	22
17	Characteristics of Liver Disease in 100 Individuals With Joubert Syndrome Prospectively Evaluated at a Single Center. Journal of Pediatric Gastroenterology and Nutrition, 2018, 66, 428-435.	1.8	21
18	Systematic analysis of physical examination characteristics of 94 individuals with Joubert syndrome: Keys to suspecting the diagnosis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, , .	1.6	1

#	Article	IF	Citations
19	Growth in Joubert syndrome: Growth curves and physical measurements with correlation to genotype and hepatorenal disease in 170 individuals. American Journal of Medical Genetics, Part A, 2022, 188, 847-857.	1.2	1