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List of Publications by Year
in descending order

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22 papers	1,002 citations	516710 16 h-index	642732 23 g-index
24 all docs	24 docs citations	24 times ranked	1600 citing authors

#	Article	IF	Citations
1	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409.	3.2	13
2	Clinical variability at the mild end of BRAT1 â€related spectrum: Evidence from two families with genotypeâ€“phenotype discordance. Human Mutation, 2022, 43, 67-73.	2.5	9
3	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202.	0.7	15
4	A novel IRF2BPL truncating variant is associated with endolysosomal storage. Molecular Biology Reports, 2020, 47, 711-714.	2.3	16
5	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.1	26
6	Agenesis of the putamen and globus pallidus caused by recessive mutations in the homeobox gene GSX2. Brain, 2019, 142, 2965-2978.	7.6	12
7	Novel mutations in the ciliopathy-associated gene CPLANE1 (C5orf42) cause OFD syndrome type VI rather than Joubert syndrome. European Journal of Medical Genetics, 2018, 61, 585-595.	1.3	22
8	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. American Journal of Human Genetics, 2017, 101, 552-563.	6.2	45
9	A novel PMCA3 mutation in an ataxic patient with hypomorphic phosphomannomutase 2 (PMM2) heterozygote mutations: Biochemical characterization of the pump defect. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 3303-3312.	3.8	17
10	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	4.5	36
11	Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Porettiâ€“Boltshauser syndrome). European Journal of Human Genetics, 2016, 24, 1262-1267.	2.8	43
12	Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615.	3.2	55
13	Very mild features of dysequilibrium syndrome associated with a novel VLDLR missense mutation. Neurogenetics, 2016, 17, 191-195.	1.4	9
14	A rare case of cerebellar agenesis: a probabilistic Constrained Spherical Deconvolution tractographic study. Brain Imaging and Behavior, 2016, 10, 158-167.	2.1	13
15	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126.	3.8	30
16	Infantile and childhood onset <scp>PLA</scp>2G6â€associated neurodegeneration in a large North African cohort. European Journal of Neurology, 2015, 22, 178-186.	3.3	25
17	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	2.5	113
18	Mutations in B9D1 and MKS1 cause mild Joubert syndrome: expanding the genetic overlap with the lethal ciliopathy Meckel syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 72.	2.7	63

#	Article	IF	Citations
19	Dandy-Walker malformation and Wisconsin syndrome: novel cases add further insight into the genotype-phenotype correlations of 3q23q25 deletions. Orphanet Journal of Rare Diseases, 2013, 8, 75.	2.7	19
20	Joubert syndrome: congenital cerebellar ataxia with the molar tooth. Lancet Neurology, The, 2013, 12, 894-905.	10.2	307
21	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
22	Pontine tegmental cap dysplasia: developmental and cognitive outcome in three adolescent patients. Orphanet Journal of Rare Diseases, 2011, 6, 36.	2.7	44