Clara Barbot

List of Publications by Year in descending order

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35 papers	3,224 citations	236925 25 h-index	330143 37 g-index
38 all docs	38 docs citations	38 times ranked	4028 citing authors

#	Article	IF	Citations
1	Molecular Characterization of Portuguese Patients with Hereditary Cerebellar Ataxia. Cells, 2022, 11, 981.	4.1	6
2	Novel MAG Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. Journal of Clinical Medicine, 2020, 9, 1212.	2.4	3
3	Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. American Journal of Human Genetics, 2015, 96, 474-479.	6.2	127
4	Hereditary Ataxia and Spastic Paraplegia in Portugal. JAMA Neurology, 2013, 70, 746.	9.0	106
5	Liver Transplantation Prevents Progressive Neurological Impairment in Argininemia. JIMD Reports, 2013, 11, 25-30.	1.5	24
6	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	21.4	157
7	Rett syndrome with and without detected MECP2 mutations: An attempt to redefine phenotypes. Brain and Development, 2011, 33, 69-76.	1.1	24
8	Shortâ€chain 3â€hydroxyacylâ€CoA dehydrogenase deficiency: the clinical relevance of an early diagnosis and report of four new cases. Journal of Inherited Metabolic Disease, 2011, 34, 835-842.	3.6	28
9	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
10	Variant Creutzfeldt-Jakob disease: the first confirmed case from Portugal shows early onset, long duration and unusual pathology. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 112-114.	1.9	4
11	Ataxia with oculomotor apraxia type 2: clinical, biological and genotype/phenotype correlation study of a cohort of 90 patients. Brain, 2009, 132, 2688-2698.	7.6	218
12	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
13	Outcome of three cases of untreated maternal glutaric aciduria type I. European Journal of Pediatrics, 2008, 167, 569-573.	2.7	29
14	Movement disorders in Rett syndrome: An analysis of 60 patients with detected MECP2 mutation and correlation with mutation type. Movement Disorders, 2008, 23, 1384-1390.	3.9	70
15	RPGRIP1L mutations are mainly associated with the cerebelloâ€renal phenotype of Joubert syndromeâ€related disorders. Clinical Genetics, 2008, 74, 164-170.	2.0	64
16	LAMA2 gene analysis in a cohort of 26 congenital muscular dystrophy patients. Clinical Genetics, 2008, 74, 502-512.	2.0	61
17	Infantile neuroaxonal dystrophy: What's most important for the diagnosis?. European Journal of Paediatric Neurology, 2008, 12, 491-500.	1.6	44
18	Spectrum of MMACHC mutations in Italian and Portuguese patients with combined methylmalonic aciduria and homocystinuria, cblC type. Molecular Genetics and Metabolism, 2008, 93, 475-480.	1.1	80

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19	Neurodegeneration associated with genetic defects in phospholipase A ₂. Neurology, 2008, 71, 1402-1409.	1.1	236
20	Identification of a new mtDNA mutation (14724G>A) associated with mitochondrial leukoencephalopathy. Biochemical and Biophysical Research Communications, 2007, 354, 937-941.	2.1	14
21	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€"Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	6.2	137
22	Stereotypies in Rett syndrome: Analysis of 83 patients with and without detected MECP2 mutations. Neurology, 2007, 68, 1183-1187.	1.1	78
23	Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519.	5.3	104
24	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nature Genetics, 2004, 36, 225-227.	21.4	454
25	Inherited and acquired risk factors and their combined effects in pediatric stroke. Pediatric Neurology, 2003, 28, 134-138.	2.1	64
26	Trinucleotide Repeats in 202 Families With Ataxia. Archives of Neurology, 2002, 59, 623.	4.5	158
27	Homozygosity Mapping of Portuguese and Japanese Forms of Ataxia-Oculomotor Apraxia to 9p13, and Evidence for Genetic Heterogeneity. American Journal of Human Genetics, 2001, 68, 501-508.	6.2	77
28	Clinical and molecular findings in four new patients harbouring the mtDNA 8993T'C mutation. Journal of Inherited Metabolic Disease, 2001, 24, 883-884.	3.6	12
29	Liver transplantation in a case of argininaemia. Journal of Inherited Metabolic Disease, 2001, 24, 885-887.	3.6	16
30	The gene mutated in ataxia-ocular apraxia 1 encodes the new HIT/Zn-finger protein aprataxin. Nature Genetics, 2001, 29, 189-193.	21.4	424
31	Recessive Ataxia With Ocular Apraxia. Archives of Neurology, 2001, 58, 201.	4.5	109
32	High Germinal Instability of the (CTG)n at the SCA8 Locus of Both Expanded and Normal Alleles. American Journal of Human Genetics, 2000, 66, 830-840.	6.2	79
33	Clinical and molecular studies in three portuguese mtdna t8993g families. Pediatric Neurology, 2000, 22, 29-32.	2.1	5
34	l-2-Hydroxyglutaric aciduria: clinical, biochemical and magnetic resonance imaging in six Portuguese pediatric patients. Brain and Development, 1997, 19, 268-273.	1.1	80
35	Pontocerebellar hypoplasia with microcephaly and dyskinesia: report of two cases. Developmental Medicine and Child Neurology, 1997, 39, 554-557.	2.1	9