

Clara Barbot

List of Publications by Year in descending order

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35
papers

3,224
citations

236612

25
h-index

329751

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. <i>Cells</i> , 2022, 11, 981.	1.8	6
2	Novel MAG Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. <i>Journal of Clinical Medicine</i> , 2020, 9, 1212.	1.0	3
3	Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. <i>American Journal of Human Genetics</i> , 2015, 96, 474-479.	2.6	127
4	Hereditary Ataxia and Spastic Paraplegia in Portugal. <i>JAMA Neurology</i> , 2013, 70, 746.	4.5	106
5	Liver Transplantation Prevents Progressive Neurological Impairment in Argininemia. <i>JIMD Reports</i> , 2013, 11, 25-30.	0.7	24
6	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. <i>Nature Genetics</i> , 2012, 44, 193-199.	9.4	157
7	Rett syndrome with and without detected MECP2 mutations: An attempt to redefine phenotypes. <i>Brain and Development</i> , 2011, 33, 69-76.	0.6	24
8	Short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: the clinical relevance of an early diagnosis and report of four new cases. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 835-842.	1.7	28
9	Novel <i>TMEM67</i> mutations and genotype-phenotype correlates in meckelin-related ciliopathies. <i>Human Mutation</i> , 2010, 31, n/a-n/a.	1.1	77
10	Variant Creutzfeldt-Jakob disease: the first confirmed case from Portugal shows early onset, long duration and unusual pathology. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 112-114.	0.9	4
11	Ataxia with oculomotor apraxia type 2: clinical, biological and genotype/phenotype correlation study of a cohort of 90 patients. <i>Brain</i> , 2009, 132, 2688-2698.	3.7	218
12	Expanding <i>CEP290</i> mutational spectrum in ciliopathies. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2173-2180.	0.7	38
13	Outcome of three cases of untreated maternal glutaric aciduria type I. <i>European Journal of Pediatrics</i> , 2008, 167, 569-573.	1.3	29
14	Movement disorders in Rett syndrome: An analysis of 60 patients with detected MECP2 mutation and correlation with mutation type. <i>Movement Disorders</i> , 2008, 23, 1384-1390.	2.2	70
15	<i>RPGRIPL</i> mutations are mainly associated with the cerebellorenal phenotype of Joubert syndrome-related disorders. <i>Clinical Genetics</i> , 2008, 74, 164-170.	1.0	64
16	<i>LAMA2</i> gene analysis in a cohort of 26 congenital muscular dystrophy patients. <i>Clinical Genetics</i> , 2008, 74, 502-512.	1.0	61
17	Infantile neuroaxonal dystrophy: What's most important for the diagnosis?. <i>European Journal of Paediatric Neurology</i> , 2008, 12, 491-500.	0.7	44
18	Spectrum of MMACHC mutations in Italian and Portuguese patients with combined methylmalonic aciduria and homocystinuria, cblC type. <i>Molecular Genetics and Metabolism</i> , 2008, 93, 475-480.	0.5	80

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19	Neurodegeneration associated with genetic defects in phospholipase A ₂ . <i>Neurology</i> , 2008, 71, 1402-1409.	1.5	236
20	Identification of a new mtDNA mutation (14724G>A) associated with mitochondrial leukoencephalopathy. <i>Biochemical and Biophysical Research Communications</i> , 2007, 354, 937-941.	1.0	14
21	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndrome-Related Disorders. <i>American Journal of Human Genetics</i> , 2007, 81, 104-113.	2.6	137
22	Stereotypies in Rett syndrome: Analysis of 83 patients with and without detected MECP2 mutations. <i>Neurology</i> , 2007, 68, 1183-1187.	1.5	78
23	Distinguishing the four genetic causes of jouberts syndrome-related disorders. <i>Annals of Neurology</i> , 2005, 57, 513-519.	2.8	104
24	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. <i>Nature Genetics</i> , 2004, 36, 225-227.	9.4	454
25	Inherited and acquired risk factors and their combined effects in pediatric stroke. <i>Pediatric Neurology</i> , 2003, 28, 134-138.	1.0	64
26	Trinucleotide Repeats in 202 Families With Ataxia. <i>Archives of Neurology</i> , 2002, 59, 623.	4.9	158
27	Homozygosity Mapping of Portuguese and Japanese Forms of Ataxia-Oculomotor Apraxia to 9p13, and Evidence for Genetic Heterogeneity. <i>American Journal of Human Genetics</i> , 2001, 68, 501-508.	2.6	77
28	Clinical and molecular findings in four new patients harbouring the mtDNA 8993T>C mutation. <i>Journal of Inherited Metabolic Disease</i> , 2001, 24, 883-884.	1.7	12
29	Liver transplantation in a case of argininaemia. <i>Journal of Inherited Metabolic Disease</i> , 2001, 24, 885-887.	1.7	16
30	The gene mutated in ataxia-ocular apraxia 1 encodes the new HIT/Zn-finger protein aprataxin. <i>Nature Genetics</i> , 2001, 29, 189-193.	9.4	424
31	Recessive Ataxia With Ocular Apraxia. <i>Archives of Neurology</i> , 2001, 58, 201.	4.9	109
32	High Germinal Instability of the (CTG) _n at the SCA8 Locus of Both Expanded and Normal Alleles. <i>American Journal of Human Genetics</i> , 2000, 66, 830-840.	2.6	79
33	Clinical and molecular studies in three portuguese mtdna t8993g families. <i>Pediatric Neurology</i> , 2000, 22, 29-32.	1.0	5
34	l-2-Hydroxyglutaric aciduria: clinical, biochemical and magnetic resonance imaging in six Portuguese pediatric patients. <i>Brain and Development</i> , 1997, 19, 268-273.	0.6	80
35	Pontocerebellar hypoplasia with microcephaly and dyskinesia: report of two cases. <i>Developmental Medicine and Child Neurology</i> , 1997, 39, 554-557.	1.1	9