

MarÃ-a-JesÃºs Sobrido

List of Publications by Year in descending order

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8
papers

788
citations

1478505
6
h-index

1588992
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g-index

8
all docs

8
docs citations

8
times ranked

1236
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in SLC20A2 link familial idiopathic basal ganglia calcification with phosphate homeostasis. Nature Genetics, 2012, 44, 254-256.	21.4	356
2	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. Nature Genetics, 2015, 47, 579-581.	21.4	237
3	Mutations in SLC20A2 are a major cause of familial idiopathic basal ganglia calcification. Neurogenetics, 2013, 14, 11-22.	1.4	131
4	Primary familial brain calcifications. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 307-317.	1.8	21
5	PET and MRI detection of early and progressive neurodegeneration in spinocerebellar ataxia type 36. Movement Disorders, 2017, 32, 264-273.	3.9	16
6	Recommendations for patient screening in ultra-rare inherited metabolic diseases: what have we learned from Niemann-Pick disease type C?. Orphanet Journal of Rare Diseases, 2019, 14, 20.	2.7	15
7	Phenotypical features of a new dominant GDAP1 pathogenic variant (p.R226del) in axonal Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2017, 27, 667-672.	0.6	6
8	Cerebellar Cognitive Affective Syndrome in Costa da Morte Ataxia (SCA36). Cerebellum, 2020, 19, 501-509.	2.5	6