Amber Begtrup

List of Publications by Year
in descending order

3 papers	109 citations	2258059 3 h-index	2550090 3 g-index
3 all docs	3 docs citations	3 times ranked	398 citing authors

#	Article	IF	Citations
1	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. American Journal of Human Genetics, 2018, 102, 995-1007.	6.2	49
2	Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. American Journal of Human Genetics, 2016, 99, 1368-1376.	6.2	46
3	2.5 yearsâ€™ experience of GeneMatcher data-sharing: a powerful tool for identifying new genes responsible for rare diseases. Genetics in Medicine, 2019, 21, 1657-1661.	2.4	14