

Amber Begtrup

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/11573555/publications.pdf>

Version: 2024-02-01

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