

Nicolas J Abreu

List of Publications by Year in descending order

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Version: 2024-02-01

6
papers

43
citations

2258059

3
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2053705

5
g-index

7
all docs

7
docs citations

7
times ranked

47
citing authors

#	ARTICLE	IF	CITATIONS
1	Overview of gene therapy in spinal muscular atrophy and Duchenne muscular dystrophy. <i>Pediatric Pulmonology</i> , 2021, 56, 710-720.	2.0	31
2	Homozygous variants in <i>AMPD2</i> and <i>COL11A1</i> lead to a complex phenotype of pontocerebellar hypoplasia type 9 and Stickler syndrome type 2. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 557-560.	1.2	5
3	Emerging Subspecialties in Neurology: Neurodevelopmental disabilities. <i>Neurology</i> , 2020, 95, 1113-1115.	1.1	4
4	Longitudinal MRI brain volume changes over one year in children with mucopolysaccharidosis types IIIA and IIIB. <i>Molecular Genetics and Metabolism</i> , 2021, 133, 193-200.	1.1	2
5	Cerliponase alfa for CLN2 disease, a promising therapy. <i>Expert Opinion on Orphan Drugs</i> , 2020, 8, 445-454.	0.8	1
6	Novel truncating variant in <i>KMT2E</i> associated with cerebellar hypoplasia and velopharyngeal dysfunction. <i>Clinical Case Reports (discontinued)</i> , 2022, 10, e05277.	0.5	0