

# Nicolas J Abreu

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6361851/publications.pdf>

Version: 2024-02-01

6  
papers

43  
citations

2258059

3  
h-index

2053705

5  
g-index

7  
all docs

7  
docs citations

7  
times ranked

47  
citing authors

#	ARTICLE	IF	CITATIONS
1	Novel truncating variant in KMT2E associated with cerebellar hypoplasia and velopharyngeal dysfunction. <i>Clinical Case Reports (discontinued)</i> , 2022, 10, e05277.	0.5	0
2	Overview of gene therapy in spinal muscular atrophy and Duchenne muscular dystrophy. <i>Pediatric Pulmonology</i> , 2021, 56, 710-720.	2.0	31
3	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
4	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
5	Emerging Subspecialties in Neurology: Neurodevelopmental disabilities. <i>Neurology</i> , 2020, 95, 1113-1115.	1.1	4
6	Cerliponase alfa for CLN2 disease, a promising therapy. <i>Expert Opinion on Orphan Drugs</i> , 2020, 8, 445-454.	0.8	1