Carlos Ignacio Ortez Gonzlez

List of Publications by Citations

16 papers	372 citations	7 h-index	19 g-index
19 ext. papers	473 ext. citations	4.8 avg, IF	1.83 L-index

#	Paper	IF	Citations
16	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. *Journal of Inherited Metabolic Disease*, **2015**, 38, 1041-57	5.4	143
15	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. *Journal of Inherited Metabolic Disease*, **2015**, 38, 1059-74	5.4	135
14	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. *Genetics in Medicine*, **2020**, 22, 1478-1488	8.1	25
13	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. *Brain*, **2020**, 143, 2696-2708	11.2	15
12	Growth Differentiation Factor 15 is a potential biomarker of therapeutic response for TK2 deficient myopathy. *Scientific Reports*, **2020**, 10, 10111	4.9	12
11	Longitudinal Study of Three microRNAs in Duchenne Muscular Dystrophy and Becker Muscular Dystrophy. *Frontiers in Neurology*, **2020**, 11, 304	4.1	7
10	Targeted Next-Generation Sequencing in a Large Cohort of Genetically Undiagnosed Patients with Neuromuscular Disorders in Spain. *Genes*, **2020**, 11,	4.2	6
9	Neuromuscular Manifestations in Mitochondrial Diseases in Children. *Seminars in Pediatric Neurology*, **2016**, 23, 290-305	2.9	4
8	The Phenotype and Genotype of Congenital Myopathies Based on a Large Pediatric Cohort. *Pediatric Neurology*, **2021**, 115, 50-65	2.9	3
7	Translational Diagnostics: An In-House Pipeline to Validate Genetic Variants in Children with Undiagnosed and Rare Diseases. *Journal of Molecular Diagnostics*, **2021**, 23, 71-90	5.1	3
6	S□drome de Guillain-Barr□en la infancia. *Anales De Pediatria Continuada*, **2013**, 11, 98-103		2
5	Early and long-term effect of the treatment with pyridostigmine in patients with GMPPB-related congenital myasthenic syndrome. *Neuromuscular Disorders*, **2020**, 30, 719-726	2.9	2
4	Association of Initial Maximal Motor Ability With Long-term Functional Outcome in Patients With COL6-Related Dystrophies. *Neurology*, **2021**, 96, e1413-e1424	6.5	2
3	Pediatric SMA patients with complex spinal anatomy: Implementation and evaluation of a decision-tree algorithm for administration of nusinersen. *European Journal of Paediatric Neurology*, **2021**, 31, 92-101	3.8	1
2	The Increasing Impact of Translational Research in the Molecular Diagnostics of Neuromuscular Diseases. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
1	Platelet Derived Growth Factor-AA Correlates With Muscle Function Tests and Quantitative Muscle Magnetic Resonance in Dystrophinopathies. *Frontiers in Neurology*, **2021**, 12, 659922	4.1	1