Jorge Javier Cebolla

List of Publications by Year
in descending order

16 papers	163 citations	1163117 8 h-index	1125743 13 g-index
25 all docs	25 docs citations	25 times ranked	261 citing authors

#	Article	IF	Citations
1	The Influence of Genetic Variability and Proinflammatory Status on the Development of Bone Disease in Patients with Gaucher Disease. PLoS ONE, 2015, 10, e0126153.	2.5	22
2	Assessment of plasma chitotriosidase activity, CCL18/PARC concentration and NP-C suspicion index in the diagnosis of Niemann-Pick disease type C: a prospective observational study. Journal of Translational Medicine, 2017, 15, 43.	4.4	19
3	ActualizaciÃ³n en deficiencia de lipasa Ã¡cida lisosomal: diagnÃ³stico, tratamiento y seguimiento de los pacientes. Medicina ClÃnica, 2017, 148, 429.e1-429.e10.	0.6	18
4	Increased glycolipid storage produced by the inheritance of a complex intronic haplotype in the Î±-galactosidase A (GLA) gene. BMC Genetics, 2015, 16, 109.	2.7	17
5	Identification of risk features for complication in Gaucherâ€™s disease patients: a machine learning analysis of the Spanish registry of Gaucher disease. Orphanet Journal of Rare Diseases, 2020, 15, 256.	2.7	15
6	Biomarker combination is necessary for the assessment of Gaucher disease?. Annals of Translational Medicine, 2018, 6, S81-S81.	1.7	14
7	LC-MS/MS analysis of plasma glucosylsphingosine as a biomarker for diagnosis and follow-up monitoring in Gaucher disease in the Spanish population. Clinical Chemistry and Laboratory Medicine, 2020, 58, 798-809.	2.3	12
8	Inhibition of Intermediate-Conductance Calcium-Activated K Channel (KCa3.1) and Fibroblast Mitogenesis by Î±-Linolenic Acid and Alterations of Channel Expression in the Lysosomal Storage Disorders, Fabry Disease, and Niemann Pick C. Frontiers in Physiology, 2017, 8, 39.	2.8	11
9	The erythrocyte osmotic resistance test as screening tool for cholesterol-related lysosomal storage diseases. Clinica Chimica Acta, 2018, 480, 161-165.	1.1	7
10	Update on lysosomal acid lipase deficiency: Diagnosis, treatment and patient management. Medicina ClÃnica (English Edition), 2017, 148, 429.e1-429.e10.	0.2	5
11	New variants in Spanish Niemannâ€“Pick type c disease patients. Molecular Biology Reports, 2020, 47, 2085-2095.	2.3	4
12	Mutations identified in a cohort of Mexican patients with lysosomal acid lipase deficiency. Annals of Hepatology, 2019, 18, 646-650.	1.5	3
13	Evaluation of two approaches to lysosomal acid lipase deficiency patient identification: An observational retrospective study. Atherosclerosis, 2019, 285, 49-54.	0.8	3
14	Serum protein profile analysis in lysosomal storage disorders patients. Clinica Chimica Acta, 2020, 510, 430-436.	1.1	3
15	Muscle-tendon weakness contributes to chronic fatigue syndrome in Gaucherâ€™s disease. Journal of Orthopaedic Surgery and Research, 2019, 14, 383.	2.3	2
16	Web-Based Bioinformatics Predictors: Recommendations to Assess Lysosomal Cholesterol Trafficking Diseases-Related Genes. Methods of Information in Medicine, 2019, 58, 050-059.	1.2	1