Código: 13773
Chave: 0053EF2424
Área Científica: Doenças Hereditárias do Metabolismo
Tipo: Caso Clínico

Título: A MULTIPLEX BIOMARKER PANEL: A POWERFUL TOOL FOR LSDS DIAGNOSIS

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Palavras-chave: Doenças Lisossomais de Sobrecarga, Niemann-PicK, Biomarcadores, SMPD1, LisoEsfingomielina

Introdução / Descrição do Caso

Lysosomal Storage Disorders (DLSs) are a set of rare, chronic and multisystemic pathologies with a variable mode of presentation and severity. Acid sphingomyelinase deficiency (ASMD), historically known as Niemann–Pick disease (NPD) types A, A/B, and B, is a rare, progressive, potentially fatal lysosomal storage disease caused by pathogenic variants in *SMPD1* gene. The disease manifestations frequently involve hepatosplenomegaly with progressive organ dysfunction, interstitial lung disease, and bleeding. The cellular damage caused by sphingomyelin accumulation can be irreversible and can lead to life-threatening complications with reduced life expectancy. ASMD can be underestimate and the diagnostic odyssey arise from an overlap in symptomology with other diseases, including primary hepatic disease, Gaucher disease, NPC, and lysosomal acid lipase deficiency.

Comentários / Conclusões

This patient had hepatosplenomegaly, elevated transaminases in which the primary clinical suspicion was an acid lipase deficiency. The enzyme activity of lysosomal acid lipase was determined, and it was normal. By the analysis of our multiplex biomarker panel, we were able to do a differential diagnosis. The biomarkers Lysosphingomyelin and Lysosm509 were approximately 500x higher than normal, suggestive of NPD. The diagnosis of ASMD was confirmed by reduced ASM enzyme activity measured in peripheral blood leukocytes and the presence of a pathogenic variant in both alleles in the *SMPD1* gene.The multiplex biomarker panel, with different lysolipids, allows a simultaneously diagnosis of different LSDs, in a timely manner, leading to an early intervention, before the appearance of more deleterious symptoms.