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REVIEW ARTICLE

Mexican consensus on lysosomal acid lipase deficiency diagnosis☆

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KEYWORDS
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Abstract
Introduction: Lysosomal acid lipase deficiency (LAL-D) causes progressive cholesteryl ester and triglyceride accumulation in the lysosomes of hepatocytes and monocyte-macrophage system cells, resulting in a systemic disease with various manifestations that may go unnoticed. It is indispensable to recognize the deficiency, which can present in patients at any age, so that specific treatment can be given. The aim of the present review was to offer a guide for physicians in understanding the fundamental diagnostic aspects of LAL-D, to successfully aid in its identification.

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Methods: The review was designed by a group of Mexican experts and is presented as an orienting algorithm for the pediatrician, internist, gastroenterologist, endocrinologist, geneticist, pathologist, radiologist, and other specialists that could come across this disease in their patients. An up-to-date review of the literature in relation to the clinical manifestations of LAL-D and its diagnosis was performed. The statements were formulated based on said review and were then voted upon. The structured quantitative method employed for reaching consensus was the nominal group technique.
Results: A practical algorithm of the diagnostic process in LAL-D patients was proposed, based on clinical and laboratory data indicative of the disease and in accordance with the consensus established for each recommendation.
Conclusion: The algorithm provides a sequence of clinical actions from different studies for optimizing the diagnostic process of patients suspected of having LAL-D.

PALABRAS CLAVE
Lipasa ácida lisosomal;
Deficiencia de lipasa ácida lisosomal;
Hepatomegalia;
Dislipidemia;
Enfermedad por depósito de ésteres de colesterol

Consenso mexicano sobre el diagnóstico de la deficiencia de lipasa ácida lisosomal

Resumen
Introducción: La deficiencia de lipasa ácida lisosomal (DLAL) ocasiona el almacenamiento de ésteres de colesterol y triglicéridos en los lisosomas de los hepatocitos y células del sistema monocito-macrófago y, como consecuencia, produce una enfermedad sistémica con manifestaciones variadas que puede pasar inadvertida; es indispensable reconocerla ya que puede diagnosticarse a cualquier edad y recibir tratamiento específico. El objetivo de este documento es ofrecer una guía que permita al médico conocer los aspectos fundamentales relacionados con el diagnóstico de la DLAL para garantizar su identificación. Este documento fue diseñado por un grupo de expertos y se presenta como un algoritmo para orientar al médico pediatra, internista, gastroenterólogo, endocrinólogo, genetista, patólogo, imagenólogo y otros especialistas que pudieran enfrentar a esta entidad.
Métodos: Se realizó una revisión actualizada de la literatura con respecto a las manifestaciones clínicas y el diagnóstico de la DLAL por parte de los expertos mexicanos. Se plantearon las declaraciones con base en esta revisión y se sometieron a votación. Se utilizó el método cuantitativo estructurado de técnica de grupo nominal para alcanzar un consenso.
Resultado: Se propone un algoritmo práctico del proceso diagnóstico de pacientes con DLAL, con base en datos clínicos y de laboratorio indicativos de la enfermedad, acorde con el consenso estabilizador para cada recomendación.
Conclusión: Este algoritmo proporciona una secuencia de acciones clínicas, basado en las manifestaciones clínicas obtenidas de los diferentes estudios, con el propósito de optimizar el proceso diagnóstico de los pacientes con sospecha de DLAL.

Introduction and aims

Lysosomal acid lipase deficiency (LAL-D) is a recessive autosomal disease characterized by the progressive accumulation of cholesteryl esters and triglycerides in the lysosomes of hepatocytes and the monocyte-macrophage system. It is not exclusive to children and has been diagnosed in persons of all ages. It is a poorly-recognized cause of dyslipidemia, associated with the development of atherosclerosis, cardiovascular disease, and progressive liver disease. It typically presents as hepatomegaly, elevated aminotransferases, and diffuse microvesicular steatosis in liver biopsy.[1,2]

LAL-D is a disease that can go unnoticed if not suspected, or confused with other entities, such as nonalcoholic fatty liver disease (NAFLD), non-alcoholic steatohepatitis, heterozygote familial hypercholesterolemia, or familial combined hyperlipidemia, among others.[2]

The aim of the present work was to propose a quick guide for the suspicion and identification of LAL-D through a proposed diagnostic algorithm.