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Lipid profile in children: The relevance to the precocious diagnosis of lysosomal acid lipase deficiency

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Lysosomal acid lipase deficiency (LAL-D) is an inherited and systemic disorder related to impaired cholesterol ester and triglyceride hydroxylation. This condition is related to the occurrence of *LIPA* gene deleterious mutations and is causative of a storage disease. LAL-D prevalence is very rare, and the clinical onset ranges abroad all ages since infancy to adulthood. The milder clinical picture affects children and adults and is frequently diagnosed based on the liver impairment. The major features are increased transaminase levels and hepatomegaly which variably progresses to liver failure and death. This presentation mimics common disorders including liver steatohepatits or primary hyperlipidemia then it should be underdiagnosed. Serum total (TC) and LDL cholesterol (LDL-C) increases are additional frequent markers while HDL-C is commonly reduced. This latter profile is pro-atherogenic but pathogenetic mechanisms are still discussed and poorly evaluated. Great progress has been reached since the screening test and the enzyme replacement therapy availability. The screening test is not considered to be applied to the general population but a specific clinical-biochemical marker to ascertain subjects to submit to LALD screening test still lacks. A diagnostic algorithm based on a 3 steps levels were applied to primary affected hypercholesterolemic pediatric subjects. 811 outpatients 5-18 yrs old were included in the study. A selection from the database of subjects to submit to the dried blood screening test for LALD detection was: LDL-C levels above160mg/dl, Recessive pedigree and; Transaminase analysis exceeding normal lab reference value. Molecular analysis of *LIPA* gene completed the analysis. Results of this study are represented by 4 new diagnosis 3/4 including asymptomatic children 3-5 yrs old. In conclusion lipid profile is required in pediatrics and attention should be paid also to recessive disorders, when changes are confirmed, to reach a final diagnosis and to improve the patient outcome. Lipids should represent a very precocious diagnostic marker of LAL-D affected children.

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