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Trabalhos Científicos

Título: Elevated Transaminases And Dyslipidemia Are Common In Children And Adults With Lysosomal Acid Lipase Deficiency

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Resumo: LAL Deficiency (LALD) is an underappreciated cause of cirrhosis, dyslipidemia and premature atherosclerotic disease in children and adults. Diagnosis requires a high index of suspicion as disease manifestations resemble those seen in some common disorders and delayed diagnosis is common. In order to better characterize the presentation and progression of LALD, we conducted a multinational observational study in children and adults with LALD. 48 patients with a confirmed diagnosis of LALD were enrolled in the study. The median age (range) of first symptoms was 5.8 (0.0 to 42.0) years and the median age at diagnosis was 9.5 (1.2 to 46.1) years. The median age at which dyslipidemia or elevated transaminases was reported was 8.4 and 8.2 years, respectively. 44 of the 48 patients had an ALT above the upper limit of normal (median 80.5 U/L) at the first recorded assessment (on study), and almost all remained abnormal during their follow-up intervals (3 to 24 months). The median highest reported total cholesterol, LDL and triglycerides were 316 mg/dL, 239 mg/dL, and 219 mg/dL respectively and the median lowest HDL was 26.5 mg/dL. Both dietary interventions and lipid lowering medications seemed to have a limited effect on controlling the liver disease, although improvements in serum lipids were observed in some patients. Six of the 48 patients underwent liver transplant: 4 pts < 18 yrs of age at the time of transplant, while 2 pts > 40 yrs of age and both developed end-stage liver disease rather precipitously. In conclusion, this study confirms that signs and symptoms of LALD usually develop in childhood, although the diagnosis is often delayed because of non-specific presentation. In some patients, disease appears to be progressive despite symptomatic management with restriction of dietary fat or lipid lowering therapy. End-stage hepatic complications may develop rapidly and without apparent warning.