This investigation had two objectives:

1. To determine the prevalence of LAL-D in an adult lipid clinic population.
2. To determine if there were any correlations between:
   - LAL activity
   - Patient demographics,
   - Lipid level
   - Magnitude of LFT abnormalities
   - The presence of cardiovascular disease (CVD).

Methods

- 4,560 patients entered into the EMR of the Florida Lipid Institute (FLI) were screened based on diagnosis of dyslipidemia (ICD-9 codes 272.0, 272.1, 272.2 or 272.5) AND the presence of LFT abnormalities (ICD-9 573.9).
- Charts of patients meeting the above criteria were reviewed and those patients with a diagnosis of pre-existing liver disease (i.e. viral hepatitis, biliary sclerosis, autoimmune hepatitis, sclerosing cholangitis) were excluded.
- The remaining patients were invited to have a free LAL assay. Blood samples were collected at the FLI and were analyzed by the Seattle Children’s Hospital clinical laboratory.
- LAL activity, patient demographics, lipid levels, LFTs and the presence of CVD were collected and analyzed.

Results

- Of the 4,560 patients in the clinic data base 97 met the ICD-9 inclusion criteria and 42 agreed to have the LAL assay performed.
- With regard to the first objective, none of the patients had LAL-D.
- With respect to the second objective, patients were divided into quartiles based on LAL activity (all within the normal range of 40 to 800 pmole/hr/spt).
- There were no significant differences among the quartiles with respect to age, gender, total cholesterol, LDL-C, HDL-C, nonHDL-C, AST or ALT levels.
- However, there was a significant difference among the quartiles (Q) with respect to the prevalence of CVD.

Discussion

1. LAL-D has a low prevalence in an adult lipid clinic population.
2. Low levels of LAL activity (within the normal range) may represent an independent risk factor for CVD.

Background

- Lysosomal acid lipase (LAL) is an enzyme found in hepatocytes and other cells that hydrolyzes cholesterol esters and triglycerides from lipoproteins taken up by receptor mediated endocytosis.
- Lysosomal acid lipase deficiency (LAL-D) is a rare autosomal recessive mutation in the LIPA gene that can present at different ages with varying rates of cardiovascular disease (CVD) progression.
- In adults, it manifests with a combination of dyslipidemia and liver function (LFT) abnormalities and is often misdiagnosed because of its commonality with other lipid and liver disorders.

Results

- The prevalence of CVD in relation to LAL enzyme activity is shown in the graph below.

References


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