LYMPOSOMAL ACID LIPASE DEFICIT IN PATIENTS WITH HYPERCHOLESTEROLEMIA

Osakidetza
ARABA ERAKUNDE SANITARIO INTEGRATUA
ORGANIZACIÓN SANITARIA INTEGRADA ARABA
bioaraba
health research institute

I.Díez López, A.Sarasua Miranda & I. Lorente Blazquez

eman ta zabal zazu
Universidad del País Vasco
Euskal Herriko Unibertsitatea

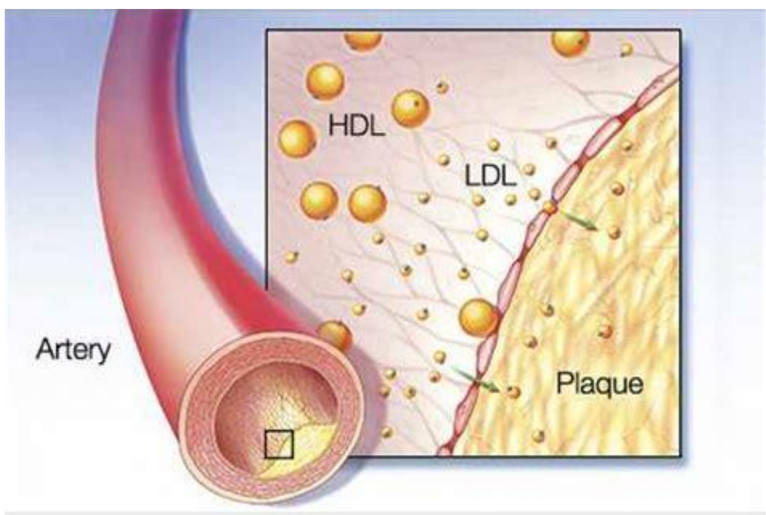
INTRODUCTION:

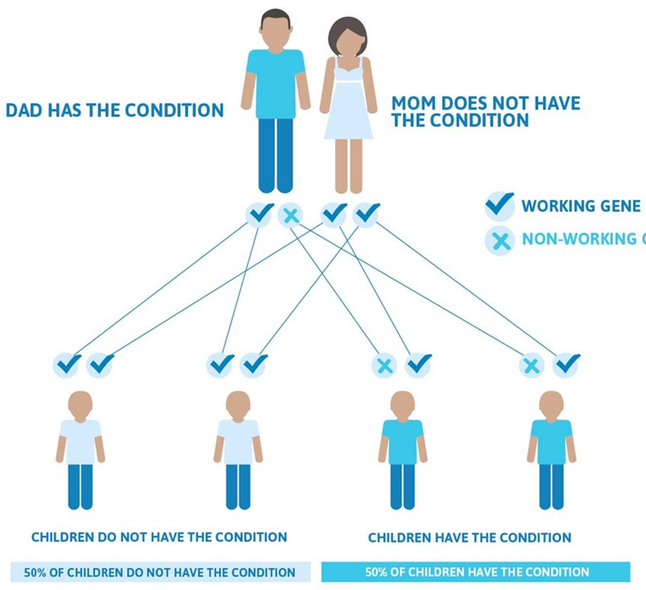
The deficit of lysosomal acid lipase (LAL) is an infrequent (1: 40,000-300,000 prevalence), autosomal recessive, monogenic pathology. It can aggressively (Wolman's disease): malabsorption and severe dyslipidemia with survival less than one year of life. The cholesterol ester storage disease (CESD) presents with dyslipidemia, liver disease and early cardiovascular disease.

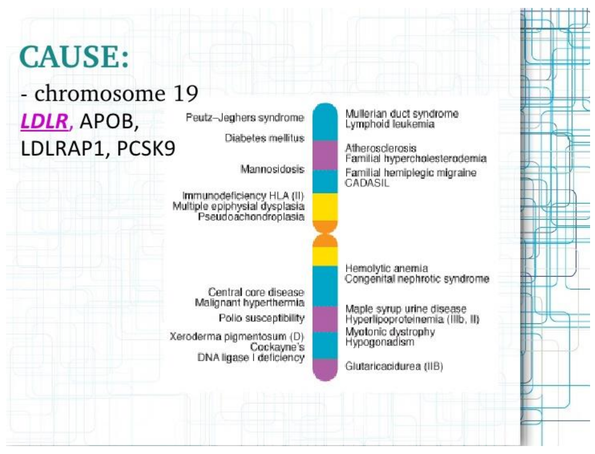
METHODS & MATERIAL:

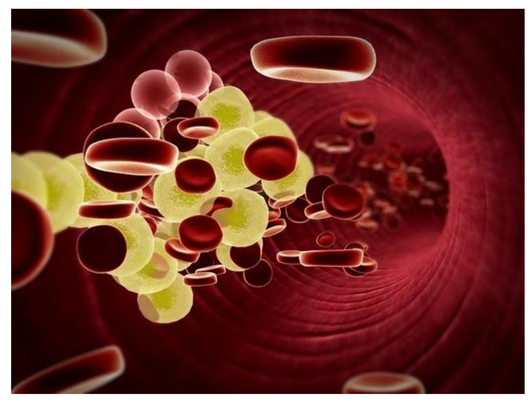
Descriptive study of the prevalence of LAL deficiency and carriers in a subsample of patients with hypercholesterolemia. Comparison with data already published

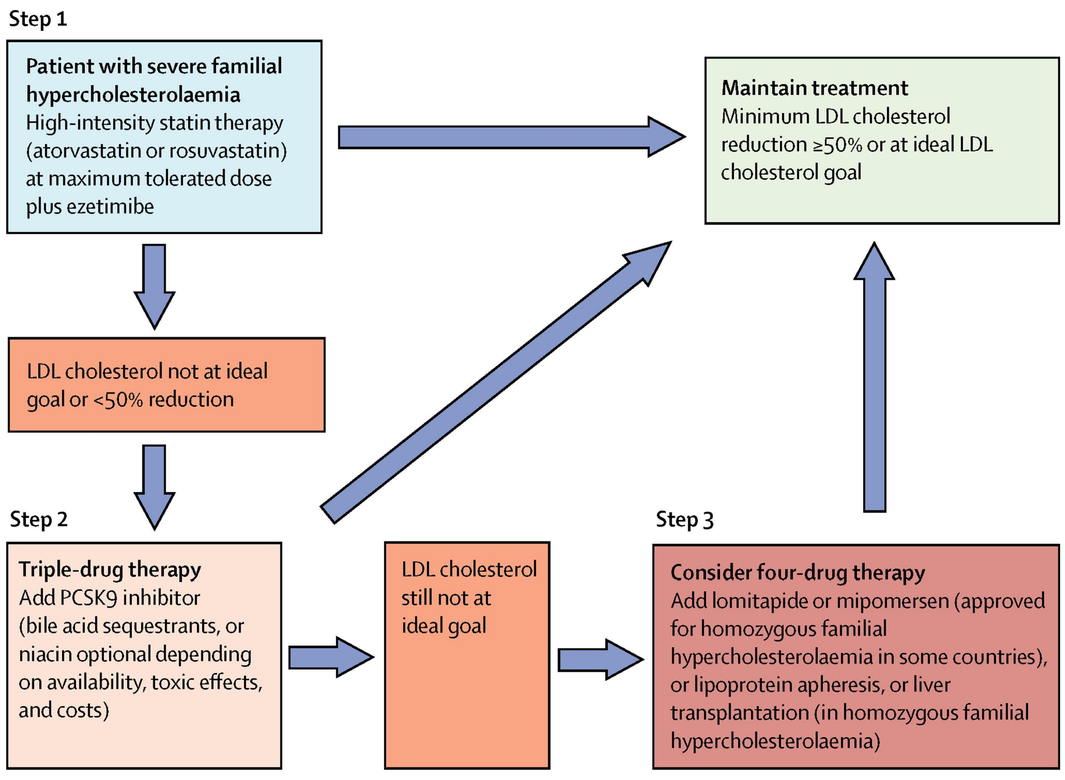
Of 42 patients monitored in the clinic for suspected familial hypercholesterolemia but a genetic study for negative HFC, 12 patients with persistent dyslipidemia were selected despite strict dietary measures. A sample of dried blood was collected in which the enzyme activity was analyzed, with prior informed consent. Reference values were considered for LAL 0.61 -2.79 nmol / punch / h. For LAL activity values with values close to the minimum of the range in the reference population, the genetic variant c.894G> A (p.delS275_Q298) ["Exon 8 Splice Junction Mutation", E8SJM] was studied,
We analyzed: age, sex, time since diagnosis, BMI, nutritional status, total cholesterol, HDL-cholesterol, LDL-cholesterol, triglycerides, treatment with statins / resins; family history of obesity, dyslipidemia and cardiovascular disease
early. Data processing with SPSS-19.0











RESULTS:

We studied 10 patients, 60% males, average age at diagnosis 8 ± 2.5 years, mean time from diagnosis 4.5 ± 1.2 years. Mean BMI 20.2 ± 3.1Kg / m2, overweight 20%, obesity 10%. Average values of: total cholesterol 225 ± 29mg / dl, HDL-cholesterol 50 ± 18 mg / dl, LDL-cholesterol 161 ± 27mg / dl, triglycerides 101 ± 72 mg / dl. Hepatic echo 2/10 mild steatosis Statin treatment: 20%, 30% ezetrol, 50% resins. Family history of: obesity 2/10, dyslipidemia 7/10 and early cardiovascular disease in the father of 1 patient Mean values of: LAL 1.32 ± 0.58 nmol / punch / h and enzymatic activity 98.1 ± 52%. Values close to the minimum range in 2 patients, both with normal E8SJM and 1 below with genetic heterozygosis mutation

CONCLUSIONS:

LAL deficiency is an infrequent entity, detecting a carrier (10%)
LAL deficit screening may be beneficial in patients with diliphemia not affiliated
The data coincide with other nearby Spanish region (Navarra)