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RV/D-001 - LYSOSOMAL ACID LIPASE DEFICIENCY (LAL-D): REPORT OF NEW FOUR SPANISH UNRELATED CASES

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Resumen

Objectives: To show Clínical, analytical, genetic, treatment and state of liver data of 4 unrelated cases of CESD.

Material and methods: We report cases of LAL deficiency included in the Registry of Dyslipemias of the Spanish Arteriosclerosis Society. We recorded demographic, anthropometric, familial, Clínical, treatment and state of liver data as well as the activity of LAL and the mutation found in the molecular analyses.

Results: Four patients followed in Lipid Units for hypercholesterolemia were found to have LAL-D. All of them have mild elevation of ALT and AST and three of them have liver steatosis (ultrasonography or hepatic RNM). Two patients underwent a liver biopsy. The degree of fibrosis ranged from F1 to F4. No cirrhosis was present. Only one patient suffered from coronary and valvular heart disease. Baseline LDL cholesterol ranged from 198 to 305 mg/dL and under lipidlowering drugs in three patients dropped to 96-125 mg/dL. LAL activity was almost undetectable in all patients (< 0.02 nmol/punch/hour). Age at diagnosis, genre, mutations found, treatment, and state of liver (steatosis Ultrasound, RMN, fibroscan, yes or not cirrhosis, biopsy realized or no, and level of transaminases) are summarized in the table.

LIVER					
Case	1	2	3	4	
Hepatic steatosis ULTRASOUND	YES	YES		YES	
STEATOSIS RNM	13% Fat	No realized	Without steatosis	No realized	
FIBROSCAN	F3-F4	F2-F3	Not done	F1-2	
CIRRHOSIS	NO	NO	NO	NO	
BIOPSY	No realized	Yes	No	Yes	
ALT (basal)	87/40	59	33	146	
AST (basal)	100/42	82	50	105	
GGT (basal)	88/52	17	31	94	
ALP (basal)	122/117	91		326	

LIPIDS				
Case	1	2	3	4
CoT (basal)	354	301	280	361
LDL (basal)	305	242	198	284
HDL (basal)	27	42	55	34
Tg (basal)	130	83	196	220
CoT (últ. disponible)	147	293	191	176
LDL (UD)	96	235	125	108
HDL (UD)	26	31	33	28
Tg (UD)	127	133	163	200

TREATMENT (actually)					
Case	1	2	3	4	
STATIN	Atorvastatin 80	no	Atorvastatin 20	Atorvastatin 20	
EZETIMIBE	Ezetimibe 10	no			
iPCSK9	Alirocumab 150	no			
OTHERS	Acenocumarol	no	Levotiroxina 100	Levotiroxina 25	

Case	1	2	3	4	
Age (years)	60	16	43	2	
Genre	Female	Female	Female	Male	
Mutations	p.Gln298Gln p.Gln298His	p.Gly87Val+ p.SdelSer275_Gln298	p.del5275_Q298 p.del5275_Q298	p.del5275_Q298 p.T33*	
LAL Activity nmol/punch/hr	< 0.02	< 0.02	< 0.02	< 0.02	
Liponext		negative			
Stroke	NO	NO	NO	NO	
CAD	Yes (CABG, AC)	NO	NO	NO	
Other cardiopaties	Acrtic valve prosthesis	NO	NO	NO	
PAD	NO	NO	NO	NO	
Xanthomas	NO	NO NO		NO	
DLCN score	12	NO	NO	15	

Conclusions: Our data confirms the notion that LAL-D is found in cohorts of patients with Polygenic or Familial Hypercholesterolemia and should be taken into account in the differential diagnosis.