CASCADE SCREENING PROGRAM FOR DYSBETALIPOPROTEINEMIA THROUGH LABORATORY TEST USING VLDL-c/TG RATIO

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1- INTRODUCTION:

Hyperlipidemia: **type III**, characteristic electrophoretic pattern: **WIDE BETA DISEASE**. (broad band between beta and pre-beta lipoproteins) Prevalence: **1 / 10,000**. Autosomal. It presents a high atherogenic risk, up to **50% of affected individuals can develop premature and severe atherosclerosis**, with a similar affection of the coronary and peripheral vascular disease. More common in men than women. Detection over 40 years. Triggers: DM2, obesity, hypothyroidism, estrogen deficiency,... Environmental: alcoholism or drugs. Characterized by a defect in **ApoE** that produces an **accumulation of chylomicrons and VLDL remnants**. Less than <10% E2/E2 develop disease.

2- OBJECTIVES:

Primary objective:

- Screening for dysbetalipoproteinemia using the VLDL-c/TG ratio.

Secondary objectives:

- Cascade family study
- Compare VLDL-c/TG ratio with other ratios and algorithms

3- METHODS:

Diagnosis procedure:

Diagnosis VLDL-c/TG ratio:

Genetic diagnosis:

4- RESULTS:

Dysbetalipoproteinemia screening:

Period of time: 2 years [2016-2018]

Cascade screening :

Genotype	Index / Familial	Treatment Yes/No	Gender	Age	CVD	HT	DM	VLDL-c/TG (>0,3mg/dl)	TG>200 + CT/ApoB >2,4 + TG/ApoB < 8,85 (mg/dl) (2)	NonHDLc/ApoB (>4,91mg/dl) (3)	VLDL-c	LDL-c	HDL-c	CT	ApoB	Lp(a)	Glucose	
E2/E3	Index	No	F	91	Ictus	Yes	Yes	0,340	Yes	3,941	128	342	381	47	516	119	8	166
E2/E2	Familial	Yes	F	24	No	No	No	0,278	Yes	2,660	68	81	245	43	192	56	7	--
E2/E3	Familial	No	F	70	Yes	Yes	No	0,198	Not applicable	1,821	35	125	177	55	228	95	13	--
E2/E3	Familial	Yes	F	68	No	Yes	No	0,203	Not applicable	1,430	29	97	143	62	175	79	13	--
E2/E3	Familial	Yes	M	61	Yes	No	No	0,256	NO	1,669	97	135	379	36	268	139	13	--
E2/E3	Familial	No	M	66	No	Yes	No	0,202	--	--	25	94	125	31	150	--	--	87
E2/E3	Familial	No	F	63	No	Yes	No	0,200	NO	1,580	39	119	220	45	203	100	16	--
E2/E3	Familial	No	F	54	No	No	No	0,203	Not applicable	1,561	24	101	118	42	181	89	12	--
E2/E3	Familial	No	F	49	No	No	No	0,201	Not applicabe	1,438	29	93	144	51	156	73	11	--
E2/E3	Familial	No	M	52	No	Yes	No	0,019	NO	1,471	5	125	267	40	196	106	--	94
E3/E3	Familial	No	F	19	No	No	No	0,202	Not applicable	1,549	21	61	104	48	127	51	5	89

Dysbetalipoproteinemia ratios:

Ratio INDEX CASES	% Positive Ratio	Ratio FAMILY CASES	% Positive Ratio	% Negative Ratio (VLDLc/TG [0,25-0,3 mg/dl]) + E2/E2 or E2/E3	% Negative Ratio (VLDLc/TG<0,25mg/dl) + E2/E2 or E2/E3 or E3/E3
VLDLc/TG ≥ 0,3 mg/dl	100%	VLDLc/TG >or< 0,3 mg/dl	0%	6,06%	93,94%
TG > 200 mg/dl	14,8%	TG > 200 mg/dl	6,06%	3,03%	90,9%
CT/ApoB > 2,4 mg/dl		CT/ApoB > 2,4 mg/dl			
TG/ApoB > 8,85 mg/dl		TG/ApoB > 8,85 mg/dl			
NonHDLc/ApoB > 4,91 mg/dl	7,4%	NonHDLc/ApoB > 4,91 mg/dl	0%	3,03%	96,9

5- CONCLUSIONS:

- The use of this ratio (VLDL-c/TG>0,3mg/dl) is an extra diagnostic tool available and necessary to identify new cases for dysbetalipoproteinemia.
- The positive ratio (VLDL-c/TG >0,3 mg/dl) could identify individuals on a high atherogenic state but with normal alleles for dysbetalipoproteinemia.
- Initiate cascades on the families, it could be a good strategy to identify new cases that are negative for the ratios.
- Comparing the ratios, VLDL-c/TG > 0,3 mg/dl has shown more efficacy for the index cases. For the cascade families the algorithm worked better than the other ratios.