APOLIPOPROTEIN B DEFICIENCY.

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INTRODUCTION

Family hypobetalipoproteinemia (HBF) is a rare genetic disorder, in 50% of cases are due to mutations APOB gene, which leads to decreased values of total cholesterol, low density lipoprotein (LDL-cholesterol) and apo-B. It is inherited as an autosomal dominant and heterozygous carriers are usually asymptomatic.

CLINICAL CASE

32 years old male with abdominal pain and diarrhea associated with food, no relevant history and physical examination standard, which hypolipidemia is detected. Once discarded hypolipidemia secondary causes of decreased cholesterol (85mg / dl), triglycerides (20mg / dl), c-LDL (32mg / dl), apo-B (23mg / dl) with slightly elevated levels are confirmed TGP (50U / L) and lack of vitamin E (473) .The family proceeded to study lipid and other affected cases with an autosomal dominant inheritance pattern detected suspect mutation in the gene for apoB. The genetic study of the index case confirmed the presence of a mutation in compound heterozygosity for the ApoB gene. With low-fat dietary treatment the patient's symptoms disappeared are currently taking supplements of vitamin E.

	index case	brother	father	grandfather	NV
Cholesterol	85 mg/dl	139mg/dl	121 mg/dl	128 mg/dl	(80-200mg/dl)
Cholesterol HDL	49mg/dl	67mg/dl	57mg/dl	75mg/dl	(37-100mg/dl)
Cholesterol LDL	32mg/dl	51mg/dl	57mg/dl	41mg/dl	
Triglycerides	20 mg/dl	106 mg/dl	36 mg/dl	61 mg/dl	(50-150mg/dl)
Apolipoprotein A1	133mg/dl				(94-178mg/dl)
Apolipoprotein B	23 mg/dl	35.1 mg/dl	38.8 mg/dl	38.8 mg/dl	(52-109mg/dl)
TGP	50 U/l	13U/l	27 U/l	17 U/l	(5-41U/l)
TGO	34U/l	16U/l	22U/l	18U/l	(5-37U/l)
Vitamin E	473.2	1124.2	913.7	975.1	(500-1800)
Vitamin A	39.4	54.7	49.6	33.6	(30-100)
25 OH vitamin D	86nmol/L	83.9nmol/L	141nmol/L	10.7nmol/L	(75-250nmol/L)

DISCUSSION

The HBF affects lipoproteins containing apo-B, so that heterozygous carriers have decreased levels of apo-B 25-30% of normal, while asymptomatic but most often present as hepatic steatosis and our homozygous paciente.Los compound heterozygotes have a similar clinical picture Abetalipoproteinemia also frequently presenting our case as diarrhea and index deficit soluble vitamins, mainly A, D and E.

CONCLUSIONS

The presence of LDL cholesterol and ApoB below P5 for the population should make us suspect the presence of HBF. The genetic study differentiates heterozygous and homozygous forms allowing family counseling and appropriate treatment.