USMF „NICOLAE TESTEMIȚANU”
CONFERINȚA ȘTIINȚIFICĂ ANUALĂ
CERCETAREA ÎN BIOMEDICINĂ ȘI SĂNĂTATE: CALITATE, EXCELENȚĂ ȘI PERFORMANȚĂ
20 22 octombrie 2021

DIAGNOSIS OF FAMILY HYPERCHOLESTEROLEMIA- LDL receptor deficiency

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Introduction

Family hypercholesterolemia (FH) is an autosomal dominant genetic desease. Despite the scientific progress made in recent years, FH tends to remain a challenge in terms of obtaining a clear diagnosis, a complete and lasting response to treatment.

Purpose

We studied the role of the gene involved and the consequences of its mutations.

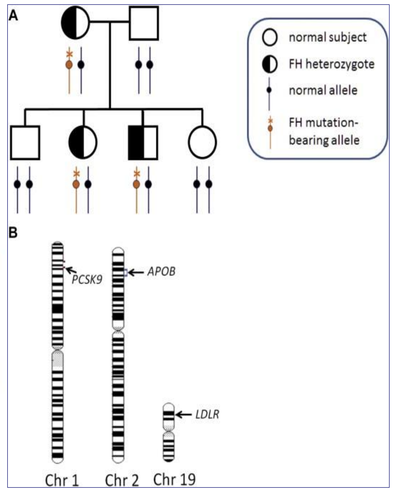

Figure 1 Genetics of FH. (A) Familial inheritance of heterozygous FH (HeFH). (B) Main genes causing FH.

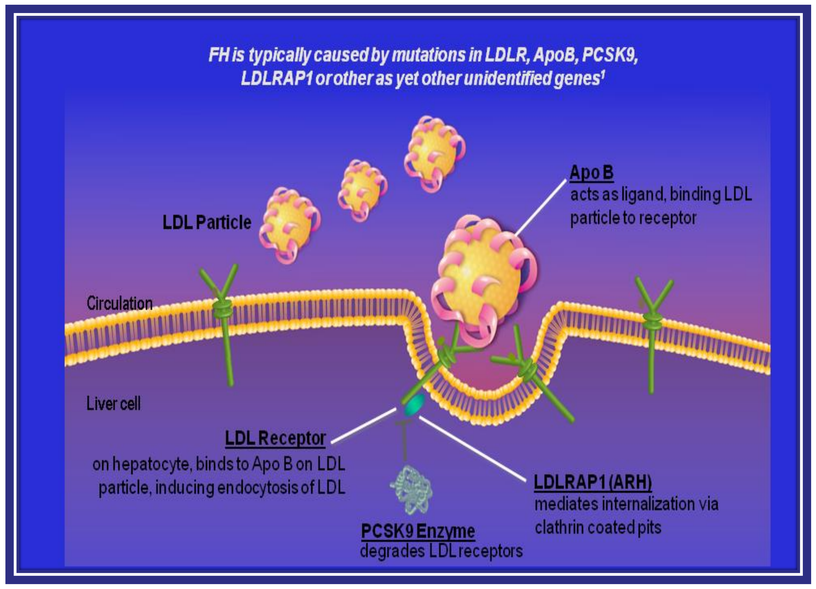

Figure 2 The most common genetic mutations associated with familial hypercholesterolemia

Keywords

family hypercholesterolemia, LDL-cholesterol, LDL-receptor.

Material and methods

The information was analyzed using the PubMed, Medscape and MEDLINE search engines.

Results

One of the causative mutations is located in the LDL-cholesterol receptor gene. Affected subjects, have high values of total serum cholesterol (> 7.8 mmol / L) and LDL-cholesterol (> 4.94 mmol /L).

FH is present from childhood, being asymptomatic, but if left untreated, 50% of men will suffer a heart attack by the age of 50 andwomen by the age of 60.

The gold standard for FH patients would be Real time PCR genotyping, using TaqMan probes or new-generation sequencing. Diagnosis of early mutation is paramount because FH is associated with an increased risk for premature coronary heart disease.

Conclusions

Although difficult, the molecular diagnosis of FH has a positive impact leading to an increase in the proportion of patients who start or intensify cholesterol lowering therapy, thus preventing and slowing the progression of atherosclerosis.