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Familiar hyper-colesterolemia and cardiovascular risk

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Familiar hypercholesterolemia was describe for first time by novel prize, studies in patients by Brown and Goldstein, both considered the fathers of this condition, who also investigated the regulation of the cholesterol synthetic Pathway. At the beginning, this condition was atribuible to defective disorder related to de HMG-CoA reductase. But in 1973 it was determine that it was genetical abnormality in the LDL particle receptor. They presume that a mutation on the LDL particle receptor gene cause a malfunction in the live capacity for capture and degrade LDL particles , leaving to the consequence of a sustained elevation of LDL level on the blood. FH is classified as two simplified phenotypes of disease according to the severity of the metabolic derangement, familiar homozygous hypercholesterolemia (HoFH), with a prevalence according to some studies of 1/1000 000 and familiar heterozygous hyper-colesterolemia (HeHF) founded in 1/500. Those studies where perform in populations with premature atehroscletorotic cardiovascular disease, associate with markedly elevated risk of death. The affected genes associated are the low-density lipoprotein (LDL) receptor, apolipoprotein (APO) B, pro-protein convertase subtilisin/kexin 9 and the Autosomal Recessive Hyper-cholesterolaemia (ARH) adaptor protein. All of these disorders have in common defective clearance of LDL within a complex system of lipid and lipoprotein metabolism and regulation. Estimating the risk the absolute cardiovascular risk in familiar hypercholesterolemia can be complicated. The CVD risk due to FH is high and represents unmet medical need for patients with FH. CHD is a cause of mortality and morbidity in individuals diagnosed with FH. Registries showed that the cumulative risk of a fatal or non-fatal coronary event in the patients with FH by the age of 60 years without effective treatment is at least 50% in men and 30% in women with a marked increase in postmenopausal women. The coronary mortality it's higher in people with a clinical diagnosis of definite FH when it is compared with the general population. The first major cardiovascular event occurs during asolescene with angina and myocardial infarction in early childhood is very often, especially in the LDLR - negative individuals. New Imaging techniques play a major role in HoFH cases. HoFH is typically respond low to existing lipid-lowering medications and this is the reason why lipoprotein apheresis is recommended in patients with HoFH as soon as possible, ideally by age 5 and before 8years.

Biography

Maxima Mendez is working as a Cardiologist Lipid master in CEDIMAT cardiovascular center in Santo Domingo RD. She has graduated from Autonoma University in Santo Domingo RD in the year 2002. She also studied ICCU Training and Cardiovascular surgical training from National Institute of Cardiology Dr. Ignacio Chavez in Mexico, DF in the year 2010. She became a Lipid master in 2015 from National Lipid Association, USA. Her main area of interest is in clinical cardiology and lipid mastering and she has participated in multiple International Cardiology Conference.

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