

عنوان مقاله:

CASCADE screening and registry of familial hypercholesterolemia in Iran: Rationale and design

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خلاصه مقاله:

BACKGROUND: Familial hypercholesterolemia (FH) is one of the most common genetic disorders, which leads to premature coronary artery disease (CAD). It has been suggested that heterozygous FH affects around ۱:۲۵۰ to ۱:۵۰۰ in the general population or even more than this, and homozygous FH affects ۱:۱۰۰۰۰۰ of the population. If patients with FH are not diagnosed and treated early in life, many of them will develop premature CAD event. As most of the patients with FH are undiagnosed, it is recommended that the general population be screened for high risks of the events since early treatments can reduce the risk of premature CADs. The clinical diagnostic criteria for FH consist of increased plasma low-density lipoprotein cholesterol (LDL-C), clinical features and family history of CAD. However, deoxyribonucleic acid (DNA)-based detection of FH mutation has high diagnostic values. As there was no screening for FH in Iran up until now, we have started screening and registering patients with FH using the CASCADE method. **METHODS:** We detected FH subjects in the general population by screening laboratories according to their

high LDL-C levels (more than ۱۹۰ mg/dl or ۱۵۰ mg/dl if receiving treatments), while our second approach was hospital-based in which one screens hospitalized patients with premature CAD events. RESULTS: We intended to screen families of indexed patients to provide standard care and therapy in order to optimize their LDL-C. CONCLUSION: This article provides detailed information on the rationale and design of this screening and registry in Iran

کلمات کلیدی:

Screening, Registries, Familial Hypercholesterolemia, Iran

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