Familial Hypercholesterolemia in the Faroe Islands

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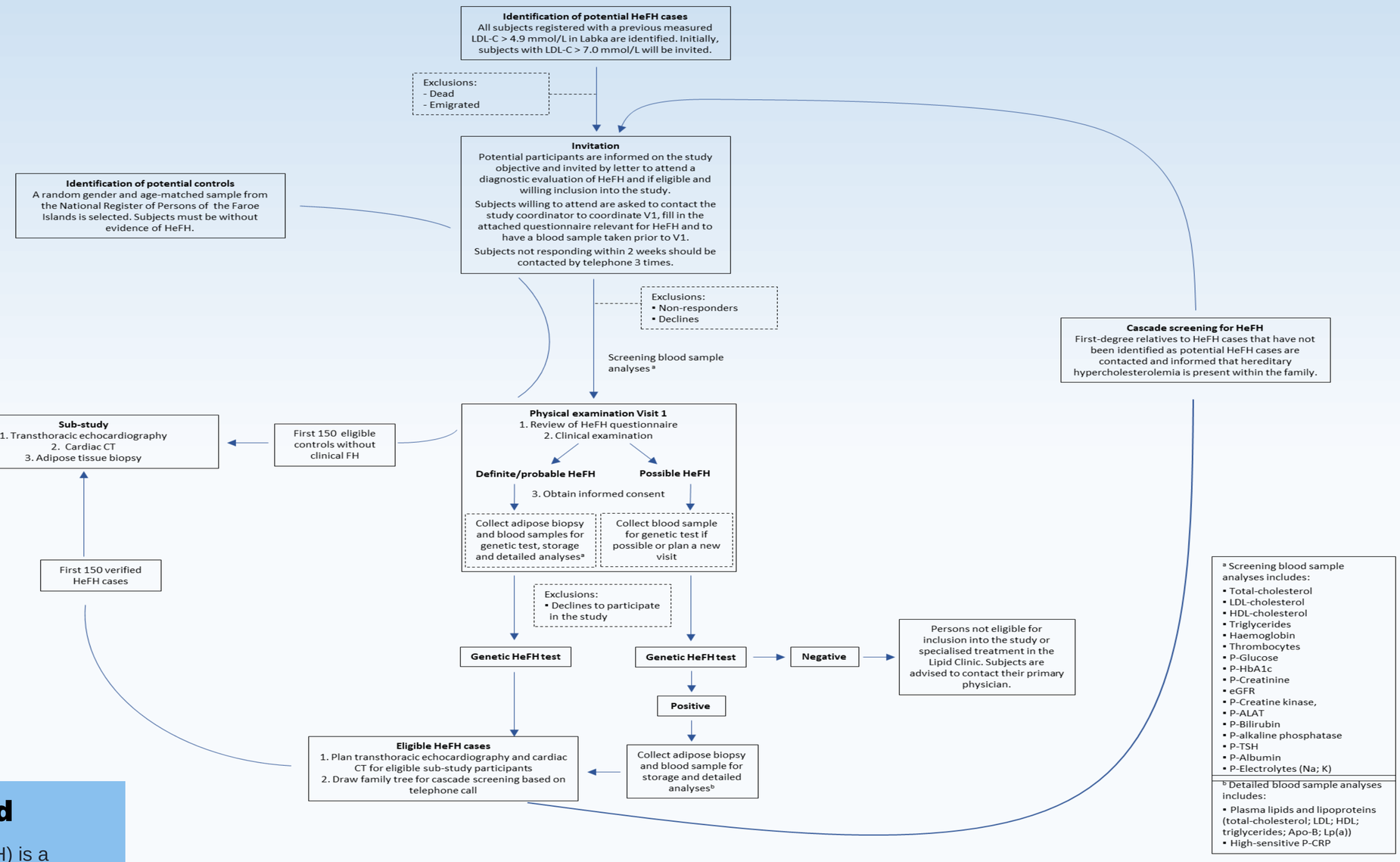

Background

Familial hypercholesterolemia (FH) is a monogenic autosomal dominant genetic disease characterized by highly elevated blood levels of low-density lipoprotein cholesterol (LDL-C). LDL-C is considered a causal factor in the development of atherosclerosis and subjects with FH not receiving appropriate lipid-lowering treatment have a substantial higher risk of developing premature coronary artery disease (CAD) and death. FH is widely underdiagnosed and undertreated in most countries of the world.

The prevalence of FH in the Faroe Islands is not known, but might be higher than in other Nordic countries due to genetic drift.

Identification of subjects with FH and detection of markers of subclinical disease may be of great value for risk stratification and to identify those subjects with FH that require additional and more aggressive treatment to lower their risk of developing manifest atherosclerotic cardiovascular disease (ASCVD).

Objective

The overall objective of this project is to investigate whether potential early ECG and echocardiographic indicators of subclinical myocardial ischemia differ between subjects with and without FH.

We hypothesize that subjects with FH and especially those naïve to statins have a higher prevalence of abnormal frontal T-wave axis and a lower global longitudinal strain (GLS) as indicators of subclinical disease compared to subjects without FH.

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Methods

Potential subjects with FH will be identified based on review of previously measured LDL-C values compatible with FH. Potential participants aged >18 years will be invited to attend a physical examination including diagnostic evaluation for FH. Subjects with possible FH will have blood samples for genetic testing and detailed lipid analyses taken. Also, information on medical history, lifestyle, social factors and medication use will be collected and a resting electrocardiograph (ECG) will be obtained. Cascade screening within the families of those subjects with verified FH will be performed.

The first 150 identified subjects with verified FH and 150 gender and age matched controls without FH aged <75 years without known ASCVD or atrial fibrillation/flutter will be invited to have an echocardiography of the heart performed. Study participants are currently being enrolled into the study.

Perspectives

Identification of subjects with FH is important to lower their risk of ASCVD by appropriate lipid-lowering treatment, which is cost-beneficial and has been associated with a substantial risk reduction. This project is expected to contribute with knowledge on whether potential abnormal echocardiographic GLS measures and ECG patterns such as frontal T-wave are more prevalent in subjects with FH compared to controls and thus may be used to identify those subjects with subclinical ASCVD that may benefit from more aggressive medical treatment and surveillance.

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