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Research protocol Validation of the Danish National FH database And DNPR diagnostic FH codes V.1

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We use this protocol and it's working

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Abstract

Background:
Familial Hypercholesterolemia (FH) is the most common inherited disease of the lipid metabolism with 1/217. FH patients are at high risk of premature atherosclerotic cardiovascular disease (ASCVD), and it is paramount to initiate lipid-lowering treatment to achieve low-density lipoprotein cholesterol (LDL-C) treatment goals. The diagnosis is made by clinical criteria and genetic test. However, only few of the 30.000 estimated FH patients remains found in Denmark, and many are suboptimal treated. The National Database for FH (DFH) was established in 2020. The purpose of the DFH is to monitor diagnostics, detection and treatment of FH patients in Denmark. Data is gathered by the local lipid clinics and from other national registries. Currently the DFH covers 50% of FH diagnosis registered in the Danish National Patients Registry (DNPR). However, the validity of the FH diagnosis registered in the DNPR and the DFH is unknown.

Aim:
This study aim to validate the FH diagnosis in a random sample in both the DFH and the DNPR to ensure a validated database for future management of FH and research originating from the DFH. We aim to prove a degree of coverage of >90% of FH diagnosis in the DFH after validation of the FH diagnosis in both registries.

Method:
A random sample of 800 patients with a diagnosis of FH will be validated against patient records calculating Dutch Clinical Lipid Network (DLCN) score (clinical and family history of ASCVD, cholesterol levels, arcus cornea and tendon xanthomas) and reviewing genetic test. A DLCN >6 is considered diagnostic for FH. The 800 patient will be compared with data in the DFH calculating sensitivity and positive predicitive value of the DFH and the DNPR including degree of coverage after validation of the diagnosis of the DFH.

Limitations:
The study is limited by the risk of low validity of the diagnosis registered in the DNPR and risk of lacking of registration of variables in the DFH.

Expected clinical impact:
Validation of the DFH is paramount to secure a valid database for future management of FH regarding prioritizing resources and initiatives for this patient group. The quality of future research originating from the DFH depends on a sufficient valid database.

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