

Evaluating Familial Hypercholesterolemia Prevalence In Pakistan's National Laboratory Network

Introduction

The rising burden of cardiovascular diseases (CVDs) in LMICs calls for strategies to target modifiable risk factors to combat the disease. Familial Hypercholesterolemia (FH) is one such disease characterized by elevated total and low-density lipoprotein cholesterol (LDL-C) since childhood. Utilizing extensive laboratory data presents an opportunity to understand FH prevalence in Pakistan, whereby specific data is notably missing.

Methodology

Data on lipid profiles was obtained from the Laboratory Information System (LIS) of the Agha Khan University Hospital (AKUH) Clinical Laboratories. It comprised lipid profiles collected from 1st January 2009 to 30th June 2018. The Make Early Diagnosis to Prevent Early Death (MEDPED) criteria was utilized to diagnose patients with FH.

Table 1: Study Methodological Framework

Measurement of LDL-C Levels	Number of tests	Data excluded
Individuals getting serum LDL- C measurement	N = 1, 542, 281	Excluded repeat testing N = (496, 259)
LDL- C measurement done for the first time	N = 1, 046, 022	Excluded Age < 10 years (N = 2,205) Age > 70 years (N = 55, 441) Missing gender/age entry (N = 42)
LDL-C measurement done for the first time (10-70 years)	N = 988, 334	Excluded potentially erroneous values (LDL – C < 10 mg/dL) N = 28
Analysis done of individual's data N = 988, 306		

Results

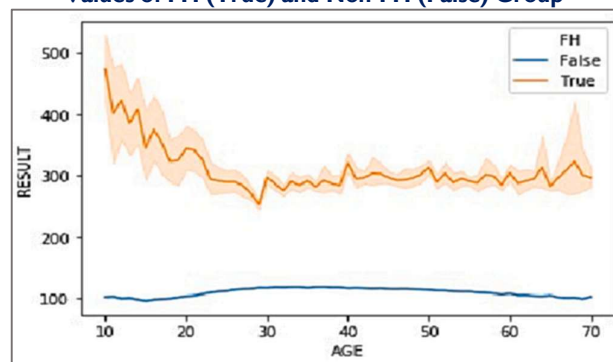
The MEDPED criteria were fulfilled by a total of 2,416 individuals (0.24%). Out of these, 77.6% had LDL \geq 250 mg/dL; 33% had LDL \geq 300 mg/dL and 11.7% had LDL \geq 400 mg/dL.

Prevalence of high LDL-C levels: Across all age groups, 2.3% or higher of the study population showed LDL-C \geq 190 mg/dL.

Salient Features

1. The overall prevalence of FH in Pakistan accounts to 0.24% which is lower as compared to European and Gulf Countries.
2. The prevalence rate of FH decreases with increasing age of the individuals, with a higher rate of diagnosis amongst females.
3. Incidence rates of FH vary according to geographical regions, with highest prevalence in Sindh.

Figure 1: Graphical Representation of Median LDL-C Values of FH (True) and Non-FH (False) Group



Age: The prevalence of FH decreased with increasing age of the sample with the highest prevalence found in individuals aged 10-19 years.

Gender: There was a higher prevalence of FH in females compared to males in the \geq 40-year age group, highlighting a later diagnosis in females.

Geolocation: The majority of patients with FH belonged to the Sindh province followed by Punjab.

Table 2: Province Wise Distribution of Positive FH Results

Province	FH Count	FH % as a subset of total samples from a given province
Sindh	1505	62.29%
Punjab	715	29.59%
KP	125	5.17%
Baluchistan	52	2.15%
Azad Kashmir	13	0.54%
Northern Areas	5	0.21%
Federal Capital Territory	1	0.04%

Recommendations

1. There is an age discrepancy in the diagnosis of FH. Thus, systematic screening programs can be implemented to identify individuals with FH at an early age, particularly during adolescence (20 years or less).
2. Since treatment of FH is costly, access to more economically affordable medication precisely Lipid lowering therapies like statins may be ensured for efficient treatment.
3. Due to limited medical knowledge of FH, health information systems that facilitate the tracking of patients may be developed.

This report was made possible with support from Bill & Melinda Gates Foundation (BMGF). The contents are the responsibility of Research and Development Solutions, and do not necessarily reflect the opinion of BMGF. This policy brief was published in collaboration with Citric Health Data Sciences, Agha Khan University.