

Genetic Backgrounds and Diagnosis of Familial Hypercholesterolemia

FAMILIAL HYPERCHOLESTEROLEMIA(FH)

Heterozygous Familial
Hypercholesterolemia
(HeHF)

Homozygous Familial
Hypercholesterolemia
(HoHF)

1:311

1:160,000-1:360,000

Single allelic mutation of
LDLR, APOB, PCSK9

Bi-allelic mutation of LDLR,
APOB, PCSK9 or LDLRAP1

LDL- C over 190mg/dL

LDL-C over 400mg/dL

Onset of CVD in 30-60
years

Onset of CVD in childhood

DIAGNOSTIC CRITERIA

Dutch Lipid Network (DLCN), Simon Broome (SB)
Register, Make Early Diagnosis, Prevent Early Deaths
(MEDPED), simplified Canadian definition of FH, The
Montreal-FH-Score (MFHS, Japan Atherosclerosis
Society (JAS) FH criteria

FAMILIAL HYPERCHOLESTEROLEMIA (FH)

Genetic disorder that causes
extremely high levels of LDL
cholesterol.

1:250

Individuals globally

Lipid disorders play a critical role
in the intricate development of
atherosclerosis and its clinical
consequences, such as coronary
heart disease and stroke.

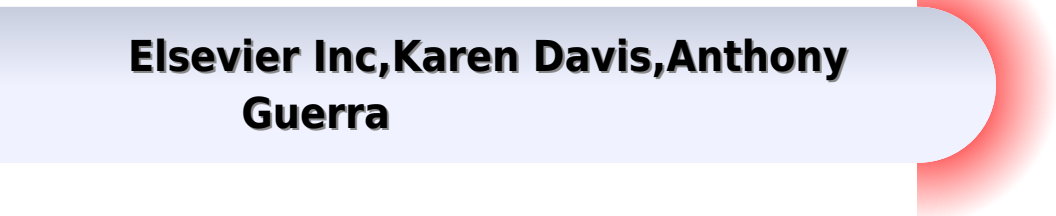
Responsible for a significant
number of deaths in many adult
populations worldwide.

>90%

FH is undiagnosed

Unit 4 Hypercholesterolemia Key Terms

**Elsevier Inc, Karen Davis, Anthony
Guerra**



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