

低密度脂蛋白
毫摩尔/升

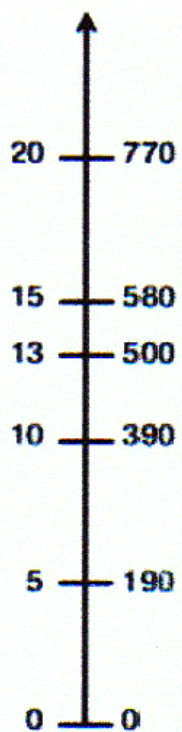
临床诊断

遗传分析

Cholestérol LDL
mmol/L / mg/dL

**Diagnostic
clinique**

**Analyse
génétique**



纯合子FH
Homozygous FH

异合子FH
Heterozygous FH

Common
hypercholesterolaemia

普通高胆固醇血症

纯合子低密度脂蛋白LDL-阴性受体

**Homozygous LDL-receptor
negative**

LDL 纯合子-缺陷受体或
LDLRAP1/ARH纯合子

**Homozygous LDL-receptor
defective or homozygous
LDLRAP1/ARH**

**Homozygous APOB defect/
PCSK9 gain of function**

缺陷的APOB纯合子
/PCSK9增强功能

**Compound heterozygous
LDL-receptor APOB/PCSK9**

混杂的LDL异合子-
APOB/PCSK9受体