



CIRCULOGENE

One Tube
One Week
Complete Results

PRECISION MEDICINE FOR FH-ASSOCIATED CHD

- ✓ Outstanding test performance with >99% sensitivity and >99% specificity
- ✓ Full gene coverage ensures all exons of every gene are included in analysis
- ✓ Industry-leading cost and time-efficiency
- ✓ Orthogonal technology for variant call confirmation

FAMILIAL HYPERCHOLESTEROLEMIA (FH)

About 1 in 50 people with high cholesterol are born with Familial Hypercholesterolemia (FH), a hereditary disorder that causes very high cholesterol. People with high cholesterol and FH are 22 times more likely to develop coronary heart disease (CHD) than those with normal cholesterol and no FH. FH is a common cause of premature cardiovascular disease and is often undiagnosed in young people.

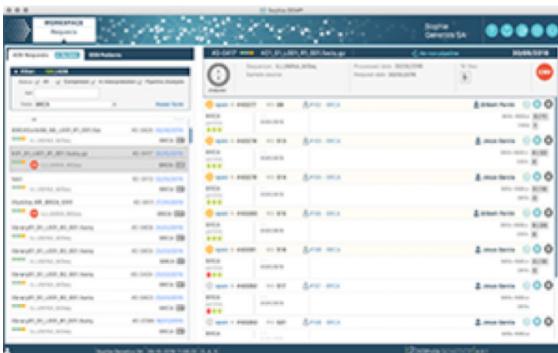
Too Many: 1 in 250 people have familial hypercholesterolemia.

Too Young: FH is responsible for an estimated 20% of myocardial infarctions before age 45 and 5% between age 45 and 60.

Too Predictable: Each first-degree relative of an individual with FH has a 50% chance of having a genetic mutation. Circulogene FH genetic test can confirm clinical diagnosis of FH and facilitate family testing (cascade screening), which can further improve treatment management in children, adolescents and young adults.

CIRCULOGENE FH GENETIC TESTING

- One tube, one week, complete results, with insurance coverage providing a comprehensive analysis of the four genes associated with FH: LDLR, APOB, PCSK9 and LDLRAP1.
- Individuals with unexplained elevated cholesterol or early cardiovascular disease could benefit from the confirmation of an FH diagnosis through our testing to enable appropriate medical management and more targeted therapy.
- Early and appropriate medical intervention can reduce the risk of cardiovascular events. Through our FH full-gene sequencing analysis, at-risk relatives could be identified and preventive therapy could be initiated.
- Guideline-based cardiovascular risk reductions could include targeted therapy to reduce LDL cholesterol (i.e., Statins, Ezetimibe, PCSK9 inhibitors).



Data Analysis & Interpretation

Sequenced data analysis, annotated and pre-classified variants, and 200,000 community variant call consensus offer you the best tool for the most accurate results.



Variant Report

All the variants are called in different pathogenicity classes, i.e., from pathogenic to benign. This AI machine-learning pre-classification significantly simplifies interpreting variants and allows you to systematically evaluate each variant.

Any reportable pathogenic, likely pathogenic or VUS variant will be confirmed by high-sensitivity, allele-specific, real-time PCR when: (1) the variant depth of coverage is <300X; and (2) the variant frequency is between 10-25% or 70-85%, where the allele zygosity status can't be clearly determined.