

## Familial Hypercholesterolemia

Order Name: **Fam Hypercholest**  
Test Number: 5194956  
Revision Date: 03/21/2023

TEST NAME	METHODOLOGY	LOINC CODE
Familial Hypercholesterolemia	Polymerase Chain Reaction	

### SPECIMEN REQUIREMENTS

Specimen	Specimen Volume (min)	Specimen Type	Specimen Container	Transport Environment
Preferred	8.5 mL (3 mL)	Whole Blood	ACD Solution A or B (Yellow Top)	Room Temperature
Alternate 1	8.5 mL (3 mL)	EDTA Whole Blood	EDTA (Lavender Top)	Room Temperature
Alternate 2	1	Saliva	Oragene Dx saliva kit	Room Temperature
Alternate 3	1	Buccal swab	PurFlock buccal swab kit	Room Temperature

**Instructions**

**Specimen Type:** Whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit  
**Specimen Volume:** 8.5 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit  
**Minimum Volume:** 3 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit  
**Collection:** Standard phlebotomy. Follow PurFlock buccal swab kit or Oragene Dx 500 saliva kit collection instructions. Do not eat, drink, smoke, or chew gum 30 min prior to collection.  
**Specimen Storage:** Maintain specimen at room temperature or refrigerate at 4C Do not freeze.  
**Special Instructions:** In cases in which there is a known variant documented in the family, the physician may prefer to order **Targeted Variant Analysis**, test code **5194970**. Test orders must include an attestation that the provider has the patient's informed consent for genetic testing.

### GENERAL INFORMATION

<b>Expected TAT</b>	14 - 21 days In some cases, additional time may be required for confirmatory or reflex tests.
<b>Clinical Use</b>	This test includes the following genes: APOB (556 bp of exon 26), LDLR, LDLRAP1 and PCSK9.
<b>Performing Labcorp Test Code</b>	482261
<b>Lab Section</b>	Reference Lab