Factor XII SNP Analysis

FXII

Collection / Transport

Collection Requirements:

Whole Blood (preferred sample type): 1 FULL vacutainer with ACD A or B, EDTA, or Heparin (ACD is preferred but not required)

Buccal Swabs: Collect two foam-tipped swabs prior to food or drink, or after thoroughly rinsing mouth with water. [Preferred Swab: Puritan cat. no. 25-1506 1PF available from Andwin Scientific at 951.674.5646 for distribution information.]

Specimen Preparation:

Buccal Swab: Collect 2 buccal swabs from inside of cheek prior to food or drink. If not possible, rinse mouth thoroughly with water prior to swabbing. Collect buccal swab samples by brushing the inside surface of 1 cheek with moderate pressure, approximately 20 times. Repeat this process with a second swab on the opposite cheek. Clearly label the sterile swab sleeve with collection date, the patient's first and last name, and patient ID number or birth date. Return swabs to sterile packaging, seal with tape.

Preferred Volume: 5mL Whole Blood collected in an EDTA (lavender-top) tube

Pediatric Collection:

Minimum volume: 1mL whole blood or 2 buccal swabs

Storage Transport Temp:

Whole Blood: Send blood Priority Overnight via FedEx and in a well insulated container on an ice pack.

Buccal Swabs: Place swabs in sterile packaging, seal with tape, and ship samples in a padded envelope at room temperatures.

Storage = Store blood samples for no longer than 1 week at room temperature or no longer than 1 month at 2-8 degrees C. Store blood sample long-term at or below -20 degrees C on a 24 hour monitored freezer.

Notes:

Hereditary angioedema (HAE) due to missense mutations in Factor XII of the complement system have been described by Bork, K et al. (Biochemical and biophysical research communications 2006 343: 1286-1289). This form of HAE has been termed HAE Type III. HAE Type III occurs primarily in women and differs from angioedema due to normal levels of C1-inhibitor. The primary variant that has been described in patients with HAE Type III is a change from the amino acid Threonine to Lysine at position 309 (T309K). A second variant at the same location has also been described, specifically Threonine to Arginine at position 309 (T309R). The Factor XII SNP sequencing assay detects both nucleotide variants (rs118204456) resulting in the amino acid changes described above by Bork, K et al.

Informed Consent is required prior to completing any of the following tests. Consent must be obtained by the provider and maintained in the patient medical record. Download and print an informed consent form here.

Overview
**Performed:**
Twice a month

**Methodology:**
PCR followed by DNA Sequencing

**Reported:**
Within 14 days from when specimen is received in the laboratory.

**Lab Department:**
Molecular Diagnostics Lab

**Synonyms:**
Factor twelve; HEREDITARY ANGIOEDEMA TYPE III; FXIIA

### Result Interpretation

**Reference Interval:**
By Report

### CPT Codes

**CPT Code:**
81403

Test Name: Factor XII SNP Analysis
Test Code: FXII