Sterols, Plasma

**Condition Description**

The post squalene biosynthesis of cholesterol from lanosterol is a multi-step enzymatic process. Intermediate enzyme deficiencies may cause decreased cholesterol biosynthesis, and result in several different metabolic disorders. This analysis detects sitosterolemia (elevated sitosterol and campesterol), an absorption disorder which results in xanthomas and early onset coronary artery disease. Lathosterolosis and desmosterolosis are severe, early onset multiple malformation syndromes (similar to Smith-Lemli-Opitz syndrome), with only a few cases reported of each condition. Elevated cholestanol is a biochemical marker for cerebrotendinous xanthomatosis (CTX), a neurological disorder that can present with chronic diarrhea during infancy, progressing to include juvenile cataracts, ataxia, xanthomas, and dysarthria.

Reference:


**Indications**

This test is indicated for:

- The identification of patients affected with sitosterolemia, desmosterolosis, or cerebrotendinous xanthomatosis.

**This test will not reliably detect carriers of any of these conditions.**

**Methodology**

Qualitative and quantitative determination performed by gas chromatography/mass spectrometry.

**Detection**

Patients receiving lipids as dietary supplementation may show elevations in several plasma sterols, in the absence of a disorder of cholesterol metabolism. Elevated stigmasterol can be used in some cases to distinguish between affected individuals and dietary artifacts. A detailed dietary history can aid in the interpretation of results.

**Reference Range**

Reference ranges are:

- Cholestanol: \( \leq 8.5 \mu g/mL \)
- Desmosterol: \( \leq 4.0 \mu g/mL \)
- Lathosterol: \( \leq 4.0 \mu g/mL \)
- Campesterol: \( \leq 7.5 \mu g/mL \)
- Stigmasterol: \( \leq 1.0 \mu g/mL \)
- Sitosterol: \( \leq 4.5 \mu g/mL \)

**Specimen Requirements**

**Type: Plasma**

Specimen Requirements:

In sodium heparin (green top) tube: 1-5 ml. Sample should be collected while fasting or 2-4 hours postprandial. Centrifuge to separate plasma and freeze.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.