

Purpose of Bile Acid testing by FAB-MS

To identify patients with genetic defects in primary bile acid metabolism (bile acid synthesis disorders, BASD). Patients positive for a primary bile acid defect may benefit from primary bile acid therapy.

Test Description

The diagnosis of bile acid synthesis disorders is achieved using mass spectrometry by detecting the presence of unique and specific atypical bile acids and/or intermediates associated with each genetic defect concomitant with a lack of normal primary bile acids. Early diagnosis and treatment is important to limit disease progression and to prevent end-stage liver disease.

Results

- Positive results may mean a patient has a defect in primary bile acid metabolism.
- Negative results may mean, within limitations of the test, a patient does not have a defect in primary bile acid metabolism.
- Uncertain results may mean that further testing/evaluation is necessary to ascertain whether a patient does or does not have a defect in primary bile acid synthesis.

Specimen Retention

Only the ordered test will be performed on this specimen. Specimens are typically retained for up to 60 days. If applicable (e.g. positive), specimens may be retained indefinitely for further workup or analysis.

Approved Signature: _____ Date: _____

Not approved

Limitations of FAB-MS Testing

Bile acid testing by FAB-MS assesses phenotypic, biochemical response to a genetic disorder and does not sequence genes.

- It is possible that genetic testing may indicate a bile acid synthesis disorder that is not reflected in the biochemical response observed during FAB-MS testing.
- Ursodeoxycholic acid (UDCA) therapy (URSO® or ACTIGALL®) may biochemically mask a defect in primary bile acid synthesis.

Potential Risks

No laboratory test is 100 % accurate.

- A suggested genetic diagnosis may be incorrect or ambiguous.
- The patient's true diagnosis may or may not be defined by this test.

Confidentiality

- Results of the Bile acid testing by FAB-MS become part of the patient's official medical record. If you would like to understand how the patient's protected health information found in the official medical record is used by Cincinnati Children's Hospital Medical Center that information can be found in CCHMC's Notice of Privacy Practices, a copy of which is on CCHMC's website at www.cincinnatichildrens.org
- As it relates to the report that is generated as a result of the bile acid testing by FAB-MS, the lab will report test results to the physician or health care providers who ordered the test.

Signatures

You may wish to obtain professional genetic counseling prior to giving consent.

I acknowledge that I have discussed the benefits, limitations, and risks of this test with my physician. I consent to bile acid testing by FAB-MS.

Patient/Legal Guardian:

Printed Name : _____ **Signature:** _____ **Date:** _____

I have explained bile acid testing by FAB-MS to this individual. I have addressed the limitations of the test and have answered all stated questions. I understand that interpretation of these results within a clinical context is ultimately my responsibility.

Physician:

Printed Name: _____ **Signature:** _____ **Date:** _____