

COMMON LABORATORY TESTS IN FATTY ACID OXIDATION DISORDERS

Alkaline Phosphatase: enzyme produced by many cells in the body. It becomes elevated when there is not enough vitamin D or when there is a blockage in the excretion of bile.

AST and ALT- transaminases: these are liver function enzymes. When the liver is stressed, they increase. They also increase when the muscle is sick and CK is elevated.

Bilirubin: derives from the breakdown of red blood cells and is eliminated in the bile. Increases when the liver cannot do its job properly or when bile ducts are blocked.

BNP: Brain natriuretic peptide measures levels of a protein made by the heart and blood vessels. BNP levels increase with heart failure.

CK: this lab is high when the muscle or the heart are stressed.

Creatinine and BUN: are removed by the kidney. Increase when the kidney shuts down. Can increase with dehydration.

Electrolytes: Sodium (Na), Potassium (K), Chloride (Cl), bicarbonate (CO₂): they determine if there is enough salt and the proper acidity. They need to be monitored when giving intravenous fluids.

Glucose: blood sugar utilized by all cells in the body. Becomes low in fatty acid oxidation disorders because the body cannot utilize other energy sources as a fuel.

Hemoglobin A1c: this lab reflects the average blood glucose over the past 3 months. It is high in diabetes and can be low (<5) when there is hypoglycemia.

Urine analysis: gives us an idea of how kidneys are functioning. It can detect hemoglobin (derived from red blood cells) and myoglobin (from muscle breakdown such as in rhabdomyolysis). Cannot distinguish between hemoglobin and myoglobin.

BIOCHEMICAL TESTS

Plasma carnitine free and total (Plasma): carnitine is necessary for fatty acid utilization. This test measures adequacy of carnitine supplies (free should be in the normal range or close to it).

Plasma acylcarnitine profile: tells us what is attached to carnitine: in fatty acid oxidation disorders there is accumulation of acylcarnitines specific for each condition.