ELEVATED C5OH ACYLCARNITINE
ORGANIC ACID DISORDERS

- Why does the baby need a second test?

The newborn metabolic screen result shows an increased level of C5OH acylcarnitine and follow up samples are required to rule out the possibility of a metabolic disorder. It is possible for C5OH to be raised in the newborn and not be significant in which case the tests will be normal and no further action required. However, in those cases where the C5OH remains elevated (or the initial result indicates), urine and plasma samples will be required to determine whether the baby has a metabolic disorder or not.

- What can an elevated C5OH indicate?

3-methylcrotonyl-CoA carboxylase (3MCC) deficiency (baby or Mum)
Multiple carboxylase deficiency (MCD) due to biotinidase or holocarboxylase synthase deficiency.
3-hydroxy-3-methylglutaryl (HMG)-CoA lyase deficiency
β ketothiolase deficiency
3-methylglutaconic aciduria (3MGA)
2-methyl-3-hydroxybutyric acidaemia (2M3HBA)

All of these are organic acid disorders, caused by a deficiency of the relevant enzyme. Although rare these condition are clinically important and therefore require early diagnosis and treatment.

- Clinical Symptoms

Neonates are usually asymptomatic in 3MCC. In the other conditions metabolic acidosis, lethargy and feeding problems are commonly seen. Multiple carboxylase deficiency (especially if due to biotinidase deficiency) can cause skin rashes, hair loss and seizures. Symptoms of organic acid disorders are common in the neonatal period but later presentations are also possible depending on the condition and severity. All the disorders can result in a metabolic crisis particularly after fasting, illness/infection or high protein intake.
Treatment

During episodes of acute decompensation, patients will need intravenous hydration, correction of acidosis and may even require dialysis in severe cases. Long term treatment includes a low protein diet (or biotin supplementation for biotinidase deficiency). There is no cure and treatment is life long.

Diagnostic Evaluation

The diagnosis of these disorders is confirmed through the analysis of urine organic acids and plasma acylcarnitines (and biotinidase activity) which reveal the metabolites associated with these conditions. The mother’s blood sample may be required as some cases may be due to maternal elevation of C5OH.

What to do after receiving the elevated C5OH result?

a) Contact the family to inform them of the screening result and ascertain the clinical status of the baby.
b) Consult a metabolic clinician for advice if baby is unwell.
c) Collect repeat blood spot or additional diagnostic samples as recommended in the screening report or by the metabolic clinician.
d) If the baby develops symptoms, advise the family to go to the emergency department with the covering letter.