ELEVATED C3 (PROPIONYL Carnitine)  
PROPIONIC ACIDAEMIA AND METHYL MALONIC ACIDAEMIA

- Why does the baby need a second test?

The newborn metabolic screen result shows an increased level of C3 (propionylcarnitine) and a repeat blood spot is required to rule out the possibility of a metabolic disorder. Elevated C3 may be transient and therefore repeat test may be normal, requiring no further action. However, in those cases where the C3 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 C3 indicate?

Propionic acidaemia (PA)  
Methylmalonic acidaemia (MMA) including cobalamin (B12) defects and transport  
Maternal B12 deficiency.

Both PA and MMA are organic acid disorders. PA is caused by a lack of propionyl-CoA carboxylase and MMA by either a lack of MMA-CoA mutase or vitamin B12. If Mum has severe B12 deficiency this can give rise to the initial abnormal results in the baby.

- Clinical Symptoms

Most newborns do not have symptoms at birth but they can develop within the first few weeks of life, in severe cases within the neonatal period. These include metabolic acidosis, failure to thrive, vomiting, dehydration and hyperammonaemia. Illness can be triggered by infections or fasting. If left untreated, these disorders may cause serious medical problems.

- 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 and carnitine supplementation. There is no cure and treatment is life long.
- **Diagnostic Evaluation**

The diagnosis of PA or MMA is confirmed through the analysis of urine organic acids and plasma acylcarnitines which reveal the metabolites associated with these conditions. To determine which form of MMA, whether due to the primary disorder, a cobalamin disorder, or maternal B12 deficiency, additional studies must be done. These include plasma total homocysteine and vitamin B12 analysis.

- **What to do after receiving the elevated C3 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.