ELEVATED TYROSINE LEVELS

- Why does the baby need a second test?

The newborn metabolic screen result shows an increased level of tyrosine and a repeat blood spot is required to rule out the possibility of a metabolic disorder. In most cases this will be transient and the repeat test will be normal requiring no further action. However, in those cases where the tyrosine remains elevated (or the initial result indicates), further samples will be required to determine whether the baby has a metabolic disorder or not.

- What can an elevated phenylalanine indicate?

Transient Tyrosinaemia of the newborn
Tyrosinaemia type 1
Tyrosinaemia type 2 or 3
Liver Disease

Patients with tyrosinaemia have problems breaking down tyrosine. The most common type of tyrosinaemia found by newborn screening is Transient Tyrosinaemia. This is a harmless condition, often seen in newborns due to immaturity of the liver. Tyrosinaemia type 1 is due to a deficiency in the enzyme fumarylacetoacetase. If raised tyrosine is due to liver disease then other amino acids such as phenylalanine and methionine are usually elevated.

- Clinical Symptoms

Tyrosinaemia type I causes severe liver failure. It may present acutely with vomiting, hypoglycaemia, bleeding or septicemia. A chronic onset is also seen with hepatomegaly, cirrhosis, growth retardation and phosphaturic rickets. Neurological signs occur due to porphyria-like crises. Tyrosinaemia type II causes corneal lesions which give rise to lacrimation, photophobia and corneal scarring. Hyperkeratosis is seen on the soles of feet and the palms of hands. It is not clear if there is an effect on mental development. Tyrosinaemia type III has an unclear phenotype. It does not affect liver, eye or skin. All forms of tyrosinaemia are very rare.

Contact Details: National Expanded Newborn Screening Lab
B1, Children’s Tower, KKH
100 Bukit Timah Rd, Singapore, 229899
Tel: (65) 6394 5049
Fax: (65) 6394 3773
- **Treatment**

  Transient tyrosinaemia will resolve without treatment. In Type I a medication called NTBC is used as well as a diet that is low in phenylalanine and tyrosine. Diet alone is used for types II and III.

- **Diagnostic Evaluation**

  The diagnosis is confirmed by analysis of plasma amino acids and urine organic acids which reveal the metabolites associated with these conditions. Enzyme confirmation is not usually performed as liver biopsy is required. DNA mutation analysis may be used for confirmation.

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