Dear Doctor Date:

**Version 0.1 (2014)**

**Re: Positive Newborn Screening for Hereditary Tyrosinaemia Type 1 (HT1)**

[name of child], [date of birth], [NHS number]

[name of child] has been detected on newborn screening to have a positive (abnormal) test for hereditary tyrosinaemia type 1 (HT1). HT1 is a rare disorder of tyrosine metabolism in which a baby or child has a problem breaking down tyrosine, resulting in accumulation of succinylacetone which causes liver damage.

A child with this condition is at risk of severe liver disease including acute liver failure, chronic liver disease with the risk of hepatocellular carcinoma; renal tubular dysfunction, growth failure, and rickets; and neurologic crises, associated with neuropathic pain, acute mental disturbance, peripheral neuropathy and respiratory failure. Death occurs in untreated patients due to liver failure, neurologic crisis or hepatocellular carcinoma. However, HT1 can be effectively managed in the longer term with medical and dietary therapy.

The positive test so far is a screening test, and therefore it is essential for the Specialist Metabolic Team to urgently review [name of child] to confirm the diagnosis and start treatment. The team will meet with the family to further explain the condition.

Infants may be symptomatic prior to this screening test result being known. If [name of child] is already an inpatient, transfer will be organised to the local Specialist Metabolic Centre at [name of IMD Centre] for further assessment and management.

Infants who are currently asymptomatic may also be at risk. If [name of child] is at home, urgent review at your local Paediatric Centre, [name of local hospital], will be organised today. Early intervention is essential for best outcomes.

Even if [name of child] is clinically well, initial same-day assessment of liver function and blood clotting will be arranged, and if these indicate that they are affected with HT1 they will be admitted and treatment commenced while awaiting results of the definitive diagnostic tests. If [name of child] is clinically well and the initial liver function tests and clotting are normal, further close monitoring will be undertaken until results of the definitive diagnostic tests are available. The diagnostic results will be available within 5 working days. If [name of child] is unwell, admission to the [name of hospital] will be arranged and definitive testing performed and treatment commenced. Treatment will include a low protein/ low tyrosine diet with medication with nitisinone that blocks the production of the toxic succinylacetone.

If the parents would like to discuss any matters prior to this review, [name of clinician] may be contacted on [contact number].

The long-term prognosis is dependent on initiating and continuing appropriate treatment. Immunisation should be undertaken as normal, general care is unaltered. The condition is inherited in an autosomal recessive fashion, with a 1 in 4 risk of recurrence in each pregnancy. Once the diagnosis has been confirmed, screening of any siblings will be offered.

You will be or may have already been contacted by [name of clinician] to discuss contact with the family. If you have any further questions, please do not hesitate to contact [name of clinician]. A further letter will be sent to you following review by the Specialist Metabolic Team.

Guidance for the longer-term management of HT1 is available in the guidelines section of the BIMDG website: [NBS Guidelines - BIMDG](https://bimdg.org.uk/guidelines/nbs-guidelines/)

Further information can be found on the following website:

NHS Newborn Blood Spot Screening Programme ([www.gov.uk/government/collections/newborn-blood-spot-screening-programme-supporting-publications](https://www.gov.uk/government/collections/newborn-blood-spot-screening-programme-supporting-publications))

With kind regards

Yours sincerely

Enclosed: Specialist Metabolic Team contact details

HT1 is suspected leaflet