Tyrosinaemia Type 1

Other names that may be used for this condition:
- Congenital Tyrosinosis
- Fumarylacetoacetase Deficiency
- Hepatorenal Tyrosinaemia
- Hereditary Tyrosinaemia Type 1

Tyrosinaemia Type 1 is a rare metabolic disorder characterised by raised levels of the amino acid, tyrosine in the blood. The disorder occurs when there is an absence or a deficiency of an enzyme known as fumarylacetoacetate hydrolase which is needed to break down tyrosine. If this amino acid is not broken down there is an accumulation of tyrosine breakdown products in the body especially the liver, the kidneys and the brain. Individuals with this condition present with either an acute or a chronic form. The forms are based on the age of the child and the severity of his or her symptoms.

This information has been kindly checked by Dr P. McKiernan (Birmingham) and is correct as of 29/04/13. This is a short internet summary only. For a full summary and further more detailed information please contact CLIMB by email, letter or telephone.

Disclaimer
This information about metabolic diseases is provided by Climb and is intended for educational purposes only. It should not be used for diagnostic or treatment purposes. Should you require more detailed information please contact Climb by email (info.svcs@climb.org.uk) or by telephone (0800 652 3181). For specific medical information regarding a particular disease or individual please contact your GP or Paediatrician.

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