Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Citrullinaemia Type 2

When the body digests protein it is broken down into small molecules known as amino acids. Excess amounts of these amino acids are converted into a toxic substance known as ammonia. In the liver, the ammonia is converted to urea and excreted in the urine. In Citrullinaemia Type 2 there is a defect in the SLC25A13 gene which provides instruction for a protein called citrin. A deficiency of citrin inhibits the urea cycle and other processes, causing a build up of ammonia and other toxins. Symptoms usually begin in adulthood and mainly affect the nervous system. Symptoms can be triggered by certain drugs, illness or infection, trauma, and alcohol.

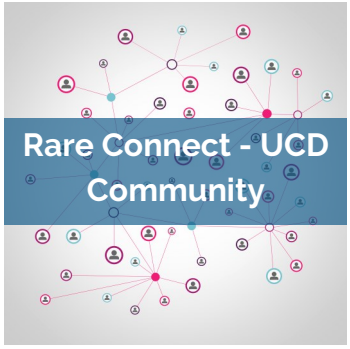
Citrullinaemia Type 2 can also present in infants. If this is the case the condition is known as Neonatal Intrahepatic Cholestasis caused by Citrin Deficiency (Also known as NICCD or Neonatal-Onset Type II Citrullinemia). This condition affects bile flow and prevents the body from using certain nutrients properly. Symptoms often resolve with time. However, in rare cases children may develop a further condition called Failure to Thrive and Dyslipidemia Caused by Citrin Deficiency (Also known as FTTDCD). In some cases, those with NICCD or FTTDCD develop the features of the adult form of Citrullinaemia Type 2 later in life.

Synonyms

Alternative names for this condition are:

- Citrin Deficiency

Further information about this condition is available from Climb.



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