



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



Supporting those affected by  
Inherited Metabolic Disorders

# Citrullinaemia Type 1

Citrullinaemia Type 1 is a Urea Cycle Disorder, in which the body's ability to manage dietary protein is impaired. 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 Urea Cycle Disorders one of the six enzymes does not function properly and ammonia is not removed from the blood stream. In Citrullinaemia Type 1 there is a deficiency or absence of the enzyme argininosuccinate synthetase. This prevents ammonia from being converted into urea and leads to an accumulation of ammonia and citrulline.

The severity of the disorder varies. Individuals with the classical form often present with symptoms soon after birth. Symptoms can include refusal to feed, vomiting, lethargy, irritability, a lack of muscle tone, an enlarged liver, breathing difficulties, seizures and the accumulation of fluid in the brain. Individuals with this disorder may progress to coma due to high ammonia in the blood. Unless this disorder is diagnosed and managed quickly it can be fatal. A less common and milder form can develop later in childhood or adulthood. There are also some individuals with the Citrullinaemia gene defect who do not show symptoms.

## Synonyms

Alternative names for this condition are:

- Citrullinuria

Further information about this condition is available from Climb.



## Disclaimer

[Please read our disclaimer and information on data protection.](#)

Updated 02/08/17

Telephone: **0845 241 2173**  
Email: [contact@climb.org.uk](mailto:contact@climb.org.uk)

Freephone: **0800 652 3181**  
Website: [www.climb.org.uk](http://www.climb.org.uk)

Children Living with Inherited Metabolic Diseases is a charity registered in England and Wales (1089588) in Scotland (SC044634) and a Company Limited by Guarantee 4267454



ClimbHQ



ClimbHQ



TeamClimb