Climb™

Supporting those affected by Inherited Metabolic Disorders

Acatalasaemia

Acatalasaemia is a disorder that is caused by a deficiency of an enzyme called catalase. Catalase is needed for the breakdown of hydrogen peroxide in the body. Acatalasaemia prevents these toxic concentrations from being broken down into oxygen and water. Acatalasaemia is very rare with an estimated prevalence of 1 in 31,250 in the general population. The condition is inherited in an autosomal recessive pattern.

There are several different variants of Acatalasaemia, although most cases are Japanese, Swiss or Hungarian forms which are of varying severity. The Japanese form being the most severe. Severity depends on the amount of enzyme activity present, if there is very little activity, the symptoms are more severe. Genetically the Hungarian type of Acatalasaemia has been characterised by 12 syndrome-causing mutations.

Synonyms

Alternative names for this condition are:

- Catalase Deficiency
- Takahara's Disease

Further information about this condition is available from Climb.

Disclaimer

Updated 07/11/16

Telephone: **0845 241 2173** Freephone: **0800 652 3181**
Email: contact@climb.org.uk Website: 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