Acatalasemia

Other names that may be used for this condition are:

- Acatalasia
- Inherited Catalase Deficiency
- Takahara’s Disease
- Acatalasaemia

This disorder is characterised by an inherited deficiency in an enzyme known as catalase. Catalase is involved in the breakdown of hydrogen peroxide to oxygen plus water. Catalase activity can be measured in the red blood cells (erythrocytes). The disorder is caused by a genetic defect on the short arm chromosome 11 (11p13). There are a number of different variants of Acatalasemia, most cases are classified as Japanese, Hungarian or Swiss. The Japanese variant which has the least enzyme activity and less than half of individuals from the earlier cases may suffer from mouth ulcerations. The Swiss variant is the least severe and often those affected do not show symptoms (asymptomatic). The prevalence of Acatalasemia in Hungary is 0.05 in 1000.

This information has been kindly checked by Dr. László Góth (Hungary) and is correct as of 16/01/13. 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.

Climb accepts no responsibility for any errors or omissions nor does Climb assume any liability of any kind for the content of any information contained within this summary or any use that you may make of it.