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

Climb™

Supporting those affected by Inherited Metabolic Disorders

Acute Intermittent Porphyria

The porphyrias are a group of eight disorders, each of which is caused by a fault in a specific gene used to produce one of the enzymes which make haem. Although haem is made in all cells of the body, the majority is used in either the bone marrow to make haemoglobin or the liver to support metabolism. Acute intermittent porphyria is one of three autosomal dominant acute porphyrias which results in partial deficiency of the enzyme porphobilinogen deaminase, which in certain situations can slow the production of haem in the liver and lead to a build up the precursor molecules 5-aminolaevulinic acid (ALA) and porphobilinogen (PBG). These molecules are associated with acute attacks with ALA being considered to be the most likely cause of the nervous system damage.

AIP does not cause any skin symptoms in sunlight, unlike most of the other porphyrias. Symptoms occur in acute attacks of physical pain and neurological problems and usually develop after puberty. The salt content of the blood may also fall.

Synonyms

Alternative names for this condition are:

- AIP, Swedish Porphyria

Further information about this condition is available from Climb.

Disclaimer

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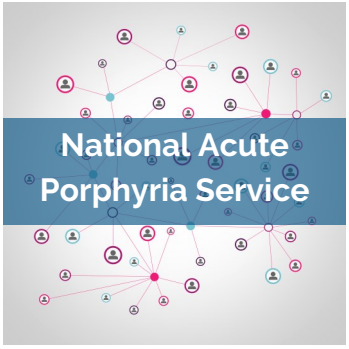

Updated 04/05/17

Telephone: **0845 241 2173** Freephone: **0800 652 3181**

Email: contact@climb.org.uk Website: www.climb.org.uk