# Acute intermittent porphyria

**Acute intermittent porphyria** is congenital which is' 'autosomal dominantly inherited' *with a mutation in the enzyme* hydroxymethylbilane synthase *or PBGD.*

## Symptoms

Symptoms occur especially in adulthood and are more common in women than in men. The most common are *attacks of neurovisceral symptoms'* . The attacks last for several days and often require hospitalization. Severe attacks can last longer and can be fatal, early diagnosis is important. The most common symptom is abdominal pain, which is long-lasting and poorly localized. Symptoms of mental disorders also occur frequently. The initial warning and identification mark may be *'dark urine'* , which is colored red to purple, looking like blood at first glance.

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### More symptoms:

- tachycardia,
- Arterial hypertension,
- anxiety,
- sweating,
- nausea,
- vomitus,
- headaches, neckache, chest pain,
- dysuria,
- dysfunction of urine bladder,
- ileus – can last for more than a week.

Peripheral Neuropathy may develop, which is manifested by muscle weakness that begins most often in the upper limbs. It can affect all limbs, respiratory muscles. The acute neurological manifestations of acute porphyrias are convulsions, such as the manifestation of *hyponatremia* . Hyponatremia can be caused by electrolyte deletion during vomiting, diarrhea, renal sodium loss, [[ADH] antidiuretic hormone] secretion]. There is an increased risk of hepatocellular carcinoma in this and other acute porphyrias.

## Metabolic abnormalities

Acute intermittent porphyria is caused by a loss of "porphobilinogene deaminase" activity, an enzyme in heme biosynthesis. Inheritance is autosomal dominant, but about 50% of the enzymatic activity of the enzyme is due to the normal allele. Porphobilinogene deaminase deficiency alone does not significantly impair hepatic heme synthesis. With increased demands on liver heme (drugs, hormones, nutritional factors), the deficient enzyme may become limiting. Hepatic "ALAS1" is indicated, 5-aminolevulinic acid and porphobilinogen accumulate. The non-enzymatic conversion of porphobilinogen and the enzymatic conversion of 5-aminolevulinic acid result in an excess of porphyrins.

## Diagnostic tests

In a patient with acute intermittent porphyria, there is a marked "" increase in porphobilinogen secretion. Concentrations are higher than 6 mg / l with a color scale for evaluating higher concentrations. Increased excretion of porphobilinogen occurs between two attacks and only after a longer period does it return to normal. Stool porphyrins are generally normal or minimally elevated in acute intermittent porphyria. Slightly elevated are porphyrins in plasma. Decreased porphobilinogene deaminase activity in erythrocytes will help confirm the diagnosis of acute intermittent porphyria.

## Treatment a prognosis

As a specific treatment for acute attacks, "hemin" is used, which is administered intravenously. It reduces the activity of ALAS1 and also reduces the urinary excretion of 5-aminolevulinic acid and porphobilinogen. Hemin therapy will also help with symptoms such as vomiting, hyponatremia and nausea. To reduce ALAS1 activity, 10% glucose is also administered intravenously, but much less frequently than hemin. The preferred form of hemin is "hemarginate". Hematin degradation products may have a transient anticoagulant effect. Hemin is most often given at 3-4 mg / kg intravenously once a day for 4 days. Most acute attacks require hospitalization, intravenous administration of hemin, and monitoring for neurological complications. Analgesics are commonly used for abdominal, back or limb pain. Occasionally, *diazepam* is given if mild sedatives are needed. For mild attacks, carbohydrates are given instead of hemin, in an amount of 300 g / day. Further attacks can be prevented by taking medication and adequate nutrition.

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# References

## External reference

- Osmosis: Acute intermittent porphyria (video) (https://www.youtube.com/watch?v=DcgBWKBRTQM)
- Genetic and Rare Diseases Information Center: Acute intermittent porphyria (https://rarediseases.info.nih.gov/diseases/5732/acute-intermittent-porphyria)

## Reference