Theme 1: The future of diagnosis: new hopes, promises and challenges

Poster Number 277





Swiss

Society for Porphyria

Abdominal Pain «Without a Good Reason» Think <u>Acute Hepatic Porphyria</u>!



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- <u>Acute hepatic porphyrias are (not so...) rare metabolic disorders affecting adult carriers</u>
- Massive abdominal pain, sometimes accompanied by dark reddish / brown urine, is the main manifestation of an acute porphyria attack

- Untreated, the acute attack can become disabling and life threatening
- Triggers include commonly used pharmaceutical drugs, stress, lack of carbohydrates, anesthetics
- A simple and specific urine test can support the diagnosis



<u>Acute hepatic porphyrias (AHP)</u> are inborn errors of metabolism. They affect the synthesis of the red blood dye «heme» in the liver.

In the liver, heme is essential to neutralize toxins and break down pharmaceutical drugs. In the healthy liver in case of a higher demand, e.g., drug intake, more heme is produced. In such instances precursors of heme accumulate in AHP sufferers.

All precursors of heme are toxic. During a so called «<u>acute attack</u>» they can cause severe abdominal pain, nausea, vomiting, constipation or diarrhea, muscle pain, numbness and paralysis, anxiety, confusion and hallucinations, high blood pressure and changes in the electrolyte composition.

Trigger factors of acute attacks are many commonly used pharmaceutical drugs - including pain killers (!) - stress, fasting, infections and oher unkown factors.





phase, the substances <u>aminolevulinic</u> acid and porphobilinogen are massively elevated in the urine. <u>Abnormally colored urine</u> can be a sign of an acute attack.

If an acute hepatic porphyria is suspected, please consult with a national porphyria reference center: European Porphyria Network: <u>www.porphyria.eu</u>

The Porphyrias Consortium: <u>www.rarediseasesnetwork.org/cms/porphyrias</u>

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