

Urinary Porphyrins Genetic Testing and Neurological Impacts of Acute Intermittent Porphyria

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ABOUT THE STUDY

Acute Intermittent Porphyria (AIP) is a rare genetic disorder characterized by intermittent episodes of severe abdominal pain, neurological disturbances, and psychiatric symptoms due to a deficiency in the enzyme porphobilinogen deaminase. This enzyme is necessary in the heme synthesis pathway, and its deficiency leads to the accumulation of toxic porphyrin precursors. AIP is inherited in an autosomal dominant pattern, meaning a single mutated gene from either parent can trigger the condition. Symptoms typically arise during acute attacks, which can be precipitated by factors such as stress, certain medications, or hormonal changes. Diagnosing AIP involves a combination of biochemical tests to detect elevated porphyrin precursors and genetic testing to confirm the presence of mutations. Management focuses on alleviating symptoms and preventing attacks through medications like intravenous heme and lifestyle adjustments. Understanding and managing AIP are important for improving the quality of life and reducing the frequency of debilitating episodes.

Role of urinary porphyrins

It plays a pivotal role in diagnosing Acute Intermittent Porphyria (AIP). During an acute attack, the deficiency of porphobilinogen deaminase leads to the accumulation of porphyrin precursors, which are excreted in the urine. Elevated levels of Porphobilinogen (PBG) and Aminolevulinic acid (ALA) in the urine are key indicators of AIP. Diagnostic testing typically involves measuring these porphyrin precursors using specialized assays. Increased urinary PBG levels are particularly significant in distinguishing AIP from other porphyrias. Additionally, urine samples may be analyzed for total porphyrin content, but PBG and ALA are more specific markers for acute episodes. Accurate measurement of these compounds helps confirm the diagnosis, guide treatment, and monitor disease progression. Given their sensitivity and specificity, urinary porphyrin levels are important in both diagnosing acute episodes and assessing the effectiveness of ongoing management strategies.

Genetic testing for AIP confirmation

It involves analyzing DNA to confirm the presence of mutations in the HMB synthase (HMBBS) gene, which encodes the enzyme porphobilinogen deaminase. This enzyme's deficiency leads to the accumulation of porphyrin precursors, causing the symptoms of AIP. The testing process typically includes sequencing the HMBBS gene to identify specific mutations known to cause the disease. This molecular diagnosis is particularly useful for confirming AIP in individuals with suggestive symptoms and abnormal biochemical test results. Genetic testing also helps identify asymptomatic carriers and allows for family counselling and risk assessment. Early and accurate genetic diagnosis can guide appropriate management strategies, such as avoiding known triggers and implementing preventive measures. By pinpointing the exact genetic mutation, clinicians can better understand the disease's inheritance pattern and customized treatment and surveillance plans to improve patient outcomes.

Managing acute AIP attacks

It involves a combination of urgent care and preventive strategies to alleviate symptoms and prevent further episodes. The primary treatment for acute attacks is intravenous administration of heme, typically in the form of hematin or heme arginate. Heme therapy helps reduce the production of toxic porphyrin precursors by supplying a synthetic heme that inhibits the enzyme overproduction. Pain management is also necessary, often requiring analgesics to relieve severe abdominal and neuropathic pain. Additionally, corresponding triggers such as fasting, stress, or certain medications is need to prevent exacerbation. Supportive care may include hydration, electrolyte balance, and monitoring for complications. Long-term management involves educating patients about avoiding known precipitants, maintaining a balanced diet, and managing stress. Regular follow-ups with a healthcare provider are important to monitor liver function and adjust treatment plans as necessary.

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Neurological implications of AIP

Acute Intermittent Porphyria (AIP) can have significant neurological implications due to the accumulation of porphyrin precursors, which are neurotoxic. During acute attacks, patients may experience a range of neurological symptoms, including peripheral neuropathy, muscle weakness, and autonomic dysfunction. Neuropathy often manifests as pain, tingling, or numbness in the extremities, while muscle weakness can impact

mobility and daily activities. Autonomic disturbances may lead to symptoms such as fluctuating blood pressure, rapid heart rate, and gastrointestinal issues. Central nervous system effects can include confusion, hallucinations, and cognitive impairments. These neurological symptoms arise because the excess porphyrin precursors interfere with nerve function and disrupt normal neurotransmission. Early diagnosis and prompt treatment are important to mitigate neurological damage.