
Sir,
The case of acute coproporphyria in a 13 years old female, who presented with neuropsychiatric symptoms (visual hallucinations and peripheral neuropathy) and abdominal pain has previously been described. We report an Irish kindred of 8 additional patients with previously undiagnosed hereditary coproporphyria, who were defined following studies of the family of the index case.

Case Report
The first step in the investigation of suspected acute porphyria is measurement of urinary a-aminolaevulic acid (ALA) and porphobilinogen (PBG). When raised, acute porphyria is suggested. These tests do not discriminate between the various types of porphyria, and they return to normal limits within 7-10 days of onset of symptoms. Genetic analysis subsequently confirmed a previously undiagnosed mutation of the coproporphyrinogen oxidase gene (c.995G>A, R332 Q) at a locus associated with hereditary coproporphyria.

The treatment of mild episodes of acute porphyria involves highenergy intake, with oral or intravenous glucose. For more severe cases, haem arginate (Normosangfi, Leiras Medica, Finland) should be infused through a large bore cannula. This treatment should be instituted early, as it can prevent the development of neurological effects, but it cannot reverse established neurological effects. Both treatments induce clinical and biochemical remission by reducing ALA synthesis. Urine ALA and PBG results return rapidly to normal after treatment is commenced.

As the inheritance of coproporphyria is autosomal dominant, the extended family was invited for screening; 10 accepted, of whom 8 have porphyria. The affected individuals span 3 generations. The youngest diagnosed individual was 3 years old. Following successful family screening, affected family members were educated and counselled, and relevant hospital and community medical services were informed about these family members. Education included provision of lists of medications that are thought to be safe and unsafe for individuals with porphyria, and family planning advice for female patients. In porphyria, barrier methods of contraception are recommended, as hormonal methods are recognised precipitants of acute porphyrria.

Symptoms possibly attributable to porphyria have since been recognised in 2 of these family members. Shortly after starting the oral contraceptive pill (a known precipitant of porphyria), a 23 years old female developed abdominal pain, without associated neuropsychiatric symptoms. This episode resolved following discontinuation of the likely offending medication and she has been well since. A second female, aged 16 years, required hospital admission with abdominal pain, without associated neurological or psychiatric symptoms. This episode resolved after instituting a course of haem arginate. Urinary ALA and PBG subsequently confirmed acute porphyrria in both.

Including the index case, 5 patients in the paediatric age range have been diagnosed with porphyria, 2 have had episodes of acute porphyrria. Due to the falling age of menarche (menarche is a recognised precipitant of porphyria) we suggest that more patients with acute porphyria can be expected to present to paediatricians.

Family screening and diagnosis allowed timely medical treatment of 2 episodes of acute porphyrria in these kindred. Thus unnecessary, expensive, prolonged investigations and hospital admission were prevented, and treatment was started before the patients developed any neurological effects. Family screening should be part of the routine management of any individual diagnosed with porphyria.

CS O’Gorman, D Gill, C Darby, V Crowley, MJ Mahony

Department of Paediatrics, Mid-Western Regional Hospital, Dooradoyle, Limerick.

Irish Porphyrin Laboratory, St James’s Hospital, James’s St, Dublin 8.

Department of Paediatric Nephrology, The Children’s University Hospital, Temple Street, Dublin 1.

Correspondence: MJ Mahony

Consultant Paediatrician, Limerick Regional Hospital, Dooradoyle, Limerick.

References