

The porphyrias are a group of rare metabolic disorders, either inherited or acquired, resulting from defects in the heme biosynthetic pathway, and characterized by a specific pattern of accumulation of heme precursors with typical clinical manifestations.

Erythropoietic protoporphyria (EPP) is caused by a ferrochelatase deficiency, the last enzyme in the heme biosynthesis pathway, leading to accumulation of protoporphyrin IX in erythrocytes.

EPP is a semi-dominantly inherited porphyria, most commonly presenting with photosensitivity during early childhood.

Adult onset EPP has scarcely been reported. Such an onset could be attributed either to an acquired aggressive form associated with hematological disease or to a phenotypically mild inherited form. Both will be discussed during this talk.

Thus, EPP should be considered in the differential diagnosis of chronic photosensitivity in adults.