AAVBC
AMERICAN ACADEMY OF VALUE BASED CARE

Porphyria

Quick Reference Guide

2026

Table of Contents

1 CLINICAL SNAPSHOT

Definition: The **porphyrias** are a group of rare, mostly inherited metabolic disorders, each arising from a deficiency in **one of the eight enzymes of the heme biosynthesis pathway**, which causes **toxic accumulation of porphyrin precursors or porphyrins**.[1] They divide into **two clinically distinct families** that should never be merged. The **acute hepatic porphyrias** (AHP) (including acute intermittent porphyria (AIP), hereditary coproporphyria (HCP), variegate porphyria (VP), and ALA-dehydratase-deficiency porphyria (ALAD)) present with acute neurovisceral attacks driven by accumulation of **delta-aminolevulinic acid (ALA)** and **porphobilinogen (PBG)**.[1] The **cutaneous (photocutaneous) porphyrias** - most importantly **porphyria cutanea tarda (PCT)** (plus erythropoietic protoporphyria (EPP), X-linked protoporphyria, and congenital erythropoietic porphyria) present with photosensitive skin disease.[2] These families differ in pathway enzyme, precursor profile, and target organ.[3] **AIP is the most common** AHP and **PCT is the most common porphyria** overall.[4] In the Medicare population, the active attack phase has usually passed, so primary care manages long-term complications rather than acute crises.

ICD-10 Codes:

All porphyrias are reported within ICD-10-CM category **E80** (disorders of porphyrin and bilirubin metabolism). The clinically meaningful subcodes are **E80.21** (acute intermittent [hepatic] porphyria), **E80.29** (other porphyria - used for HCP, VP, EPP, or X-linked protoporphyria, with the specific subtype named in the note), **E80.1** (porphyria cutanea tarda), and **E80.0** (hereditary erythropoietic porphyria, i.e., congenital erythropoietic porphyria/Gunther disease). **E80.20** (unspecified porphyria) is a trap code: it should be used only while the subtype is pending and replaced with the specific code once biochemical or genetic confirmation establishes the type. Commonly co-occurring conditions are coded separately - for example the long-term sequelae of AHP such as chronic kidney disease **(N18.-)**, hepatocellular carcinoma **(C22.0)**, essential hypertension **(I10)**, and polyneuropathy **(G62.-)**. Subtype specificity is not a billing nicety; the named subtype drives drug-safety protocols, monitoring requirements, and complication-surveillance level.

Prevalence and Burden: Overall porphyria prevalence ranges from roughly **1 in 500 to 1 in 50,000** depending on type and geography.[6] Symptomatic AIP, the most common AHP, has a US prevalence of about **1 in 100,000**, although pathogenic HMBS variants are carried by approximately **1 in 1,700** individuals with clinical penetrance near **1%** in the general population (rising toward 23% in families with an affected proband).[7,8] Acute AHP attacks occur predominantly in **women aged 15-50** (roughly **90%** of symptomatic patients are female)[3] and are rare after menopause, the 65+ population is defined less by attacks than by accumulated organ injury.[7] Among patients with recurrent attacks, **65%** report chronic symptoms and **46%** experience daily symptoms, and symptomatic patients reach disability pension at a median age of about **45 years.**[9,10]