



Porphyria

Quick Reference Guide

2026

Table of Contents

1. CLINICAL SNAPSHOT	3
HCC/RAF V28 Mapping	4
2. RECOGNITION AND DIAGNOSIS	5
Medicare Screenings and Surveillance (Adult and Geriatric)	5
Subtle Early Signs in Older Adults (>65)	6
Geriatric Risk Factors	7
Diagnostic Thresholds (AGA 2023/IPNET 2026)	8
Clues to Dig Deeper	9
Common Oversights	9
Key Differentials in Elderly	10
Comorbidity Screening	11
3. MEAT DOCUMENTATION ESSENTIALS	12
Clinical Documentation Elements	13
Reframing Common Documentation Shortcuts	14
4. TREATMENT AND REFERRAL QUICK GUIDE	15
Therapy Escalation Criteria	15
AGA/IPNET Guideline-Aligned Treatment by Patient Profile	16
Non-Pharmacologic Care and Supportive Interventions	17
Medication Safety and Key Interactions	17
When to Refer	18
Follow-Up Timing	19
Comorbidity Management.....	19
Cost-Smart Options	20
Patient Education and Adherence	21
Quality Metrics Tie-In	22
5. CODING REMINDERS AND CASE EXAMPLES	24
Coding Specificity	24
Annual Clinical Review and Confirmation	24
Good Documentation - EHR Tips.....	25
Brief Case Examples.....	25
REFERENCES	26

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}

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