AAVBC

AMERICAN ACADEMY OF VALUE BASED CARE

Porphyria

Quick Reference Guide

2026

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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]

HCC/RAF V28 Mapping

ICD-10 CODE	HCC (V28)	RAF (CNA)	DOCUMENTATION ANCHOR
E80.21 Acute intermittent (hepatic) porphyria (AIP)	HCC 50—Amyloidosis, Porphyria, and Other Specified Metabolic Disorders	0.648	Document "acute intermittent porphyria" or "AIP." State the diagnostic basis (such as elevated urine PBG during an attack, or genetic testing confirming an HMBS gene mutation). This supports the medical necessity for givosiran (Givlaari)
E80.29 Other Prophyria (HCP, VP, EPP, X-linked protoporphyria)	HCC 50	0.648	Includes: Hereditary coproporphyria (HCP) and ALAD-deficiency porphyria (ADP). Note: Do not use for EPP/XLP (which belong in E80.0) or Variegate Porphyria (which belongs in E80.20). Specify the subtype clearly in the clinical note.
E80.1 Porphyria cutanea tarda (PCT)	HCC 50	0.648	Document "porphyria cutanea tarda" or "PCT." Establish the active-management context (e.g., ongoing therapeutic phlebotomy schedules, or low-dose oral hydroxychloroquine therapy) to substantiate active status
E80.0 (Hereditary erythropoietic porphyria)	HCC 50	0.648	Includes: Congenital erythropoietic porphyria (CEP/Gunther disease), Erythropoietic protoporphyria (EPP), and X-linked protoporphyria (XLP). Note: EPP and XLP must be coded here. **E80.0** is the mandatory primary code required to **secure prior authorization** for afamelanotide (Scenesse) implants
E80.20 (Unspecified porphyria - AVOID)	HCC 50	0.648	Acceptable **only** while the subtype is **pending**; replace with the specific subtype code once confirmed

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; AIP = Acute Intermittent Porphyria; CEP = Congenital Erythropoietic Porphyria; CNA = Community Non-Dual Eligible, Aged; HCC = Hierarchical Condition Category; HCP = Hereditary Coproporphyria; HCQ = Hydroxychloroquine; ICD-10-CM = International Classification of Diseases, 10th Revision, Clinical Modification; PCT = Porphyria Cutanea Tarda; RAF = Risk Adjustment Factor; V28 = Version 28 CMS-HCC model; VP = Variegate Porphyria *RAF values represent the Community Non-Dual Eligible Aged (CNA) coefficient from the 2026 CMS-HCC model;11 values vary across patient populations based on eligibility and care setting

Risk-Adjusted Care Resources per Patient/Year[11,12]

Risk-adjusted care resource allocation — MA base rate (~$10,402.34) × RAF coefficient

Porphyria
~$6,741

HCC 50 · RAF 0.648

2 RECOGNITION AND DIAGNOSIS

Medicare Screenings and Surveillance (Adult and Geriatric)

TEST/SURVEILLANCE	POPULATION AND TIMING	PURPOSE
Spot (random) urine PBG + ALA + creatinine[7]	Any patient with unexplained recurrent severe abdominal pain, especially during an active episode; sample protected from light, normalized to creatinine	First-line screen/diagnosis for AHP; ALA and PBG are elevated **>=5x** the upper limit of normal during an attack[13]
Urine, plasma, or erythrocyte porphyrins (type-directed)[7]	When a cutaneous porphyria is suspected; plasma/urine uroporphyrin for PCT, erythrocyte protoporphyrin for EPP[14]	Establishes and subtypes cutaneous porphyria; **not a substitute** for PBG in AHP screening
Liver ultrasound (HCC surveillance)[7]	Every **6 months from age 50** for ALL AHP patients, regardless of cirrhosis status[15]	Detects AHP-associated hepatocellular carcinoma, which occurs without cirrhosis and is **missed by standard cirrhosis-based protocols**
Serum creatinine/eGFR[7]	Baseline then at least annually for all AHP patients; every 3 months for the first 6 months on givosiran[16]	Detects **porphyria-associated CKD** (up to **59%** of symptomatic AIP) and drug-related renal change[17]
Blood pressure[7]	Every visit for chronic management; frequently during an acute attack	**Hypertension** affects about **43%** of AHP patients and contributes to CKD and cerebrovascular risk[18]

TEST/SURVEILLANCE	POPULATION AND TIMING	PURPOSE
Comprehensive medication review against APF database[7]	Every encounter and at every medication change	**Identifies porphyrinogenic drugs** - the single most preventable trigger in polypharmacy

ABBREVIATIONS: AGA = American Gastroenterological Association; AHP = Acute Hepatic Porphyria; AIP = Acute Intermittent Porphyria; ALA = delta-Aminolevulinic Acid; APF = American Porphyria Foundation; CKD = Chronic Kidney Disease; eGFR = estimated Glomerular Filtration Rate; EPP = Erythropoietic Protoporphyria; HCC = Hepatocellular Carcinoma; IPNET = International Porphyria Network; PBG = Porphobilinogen; PCT = Porphyria Cutanea Tarda; ULN = Upper Limit of Normal

Subtle Early Signs in Older Adults (>65)

SUBTLE SIGN IN OLDER ADULTS	WHY IT IS MISSED	CLINICAL SIGNIFICANCE
Recurrent severe abdominal pain with a benign abdominal exam and normal imaging[7]	Attributed to functional GI disease, adhesions, or aging bowel; pain is severe yet imaging shows no peritoneal signs	**Hallmark of an AHP neurovisceral attack**; pain out of proportion to exam, often with ileus, should prompt urine **PBG**
Tachycardia and elevated systolic blood pressure during pain episodes[13]	Read as anxiety, pain response, or essential hypertension	Autonomic features of an **acute attack**; the combination with abdominal pain is a key clue
Hyponatremia, sometimes with seizures[13]	Attributed to thiazides, SIADH, or poor intake common in elders	The triad of **seizures, abdominal pain, and hyponatremia** is highly suggestive of acute porphyria
Progressive limb weakness, foot drop, or wrist drop[13]	Mistaken for stroke, radiculopathy, or deconditioning	**Motor neuropathy from an attack**; may persist and signals severe disease needing urgent treatment
New psychiatric symptoms - confusion, anxiety, hallucinations, low mood[19]	Misread as dementia, delirium, or late-life depression	**Neurovisceral attacks** produce psychiatric features; **46%** of recurrent-attack patients report daily symptoms[10]

SUBTLE SIGN IN OLDER ADULTS	WHY IT IS MISSED	CLINICAL SIGNIFICANCE
Blistering, fragility, and scarring on sun-exposed skin (hands, face)[14]	Attributed to age-related skin fragility, eczema, or trauma	Classic for PCT, the porphyria of this age group; prompts **plasma/urine porphyrins**

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; GI = Gastrointestinal; PBG = Porphobilinogen; PCT = Porphyria Cutanea Tarda; SIADH = Syndrome of Inappropriate Antidiuretic Hormone

Geriatric Risk Factors

RISK FACTOR/TRIGGER	MECHANISM	GERIATRIC RELEVANCE
Porphyrinogenic medications[7]	Induce hepatic **ALAS1**, increasing **ALA/PBG** production and precipitating attacks[15]	**The most modifiable trigger**; risk is amplified by polypharmacy **(commonly 5-12 agents**) in older adults
Caloric restriction/fasting[15]	Glucose deprivation de-represses **ALAS1**	Relevant with **poor intake, acute illness, or procedural NPO orders**; coordinate nutrition and minimize fasting windows
Alcohol use[14,15]	Directly porphyrinogenic; **triggers both AHP attacks and PCT**	Common and modifiable; **abstinence** is first-line PCT therapy
Iron overload, HCV, estrogen, smoking, HFE variants[14]	Drive uroporphyrinogen decarboxylase inhibition in the liver **(PCT pathogenesis)**	All concentrate in the Medicare-age PCT population; **HCV** is present in roughly **two-thirds of PCT patients**
Physiologic/psychological stress (infection, surgery)[15]	Stress hormones and inflammation induce **ALAS1**	Perioperative and infection-related attacks **require porphyria-safe anesthesia and antibiotic choices**
PEPT2 *1/*1 genotype[20]	**Independently worsens** porphyria-associated **renal outcomes**	Associated with an odds ratio of about **6.85 for eGFR <60**, compounding age-related nephron loss

RISK FACTOR/TRIGGER	MECHANISM	GERIATRIC RELEVANCE

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; ALA = delta-Aminolevulinic Acid; ALAS1 = delta-Aminolevulinic Acid Synthase 1; eGFR = estimated Glomerular Filtration Rate; HCV = Hepatitis C Virus; HFE = Homeostatic Iron Regulator gene; NPO = Nil Per Os (nothing by mouth); PBG = Porphobilinogen; PCT = Porphyria Cutanea Tarda; PEPT2 = Peptide Transporter 2

Diagnostic Thresholds (AGA 2023/IPNET 2026)

DIAGNOSTIC STEP	CRITERION/THRESHOLD	NOTES
First-line biochemical test (AHP)[7]	Spot urine **PBG**, ALA, porphyrins, and creatinine; a 24-hour collection is not required	**ALA and PBG ≥5x ULN during an attack confirm AHP**; PBG is elevated in all AHP except ALAD porphyria
Sample handling[7,13]	**Protect from light**; normalize results to urine creatinine	**Light exposure and failure to normalize** are common technical causes of false results
Timing/repeat testing[7]	Collect during **active symptoms**; **15-44%** of sporadic AIP patients have normal ALA/PBG between attacks	**Normal between-attack** values **do not** exclude AHP; repeat during an attack
Confirmatory genetic testing[7]	Sequence HMBS, CPOX, PPOX, ALAD **after positive** biochemical testing	Genetic testing **confirms subtype**; it is **NOT a first-line screen** and may not be covered without prior biochemical evidence
Cutaneous porphyria (PCT)[14]	**Elevated** plasma and urine **porphyrins** (uroporphyrin, heptacarboxylporphyrin) with **skin fragility/blistering**	Plasma fluorescence peak distinguishes **PCT (619-622 nm)** from **VP (626 nm)**[4]
Clinical subgroup classification (no numeric score)[7]	Sporadic **(<4 attacks/yr)**; recurrent (**≥4/yr**); asymptomatic high excretor; latent carrier	The **≥4 attacks/year** threshold triggers consideration of prophylactic therapy

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; AIP = Acute Intermittent Porphyria; ALA = delta-Aminolevulinic Acid; ALAD = ALA-Dehydratase-Deficiency porphyria; PBG = Porphobilinogen; PCT = Porphyria Cutanea Tarda; ULN = Upper Limit of Normal; VP = Variegate Porphyria

Clues to Dig Deeper

CLUE TO DIG DEEPER	WHAT TO DO NEXT
Severe abdominal pain with a normal/benign exam and nondiagnostic imaging, poorly responsive to analgesia[7,13]	Order spot urine **PBG + ALA + creatinine** during the episode;[1] do not stop at total porphyrins
Abdominal pain plus tachycardia, hypertension, and hyponatremia in the same patient[4,15]	Treat as a probable acute attack and **test PBG urgently**; **watch sodium** closely
Recurrent unexplained hospitalizations with negative standard GI work-up[7]	Review for prior porphyria testing; **obtain urine PBG** and a careful medication and family history
Dark red or brown urine reported during pain episodes[4]	Supports **porphyrin precursor accumulation**; collect a protected spot urine for PBG/ALA
Blistering skin disease on the hands/face with fragility and HCV, alcohol, or iron-overload risk[14,21]	Send plasma/urine porphyrins for PCT; **screen for HCV and check ferritin/transferrin saturation**
Family history of porphyria or unexplained acute attacks[4]	Counsel and arrange **biochemical screening**; most porphyrias are autosomal dominant with low penetrance

ABBREVIATIONS: AGA = American Gastroenterological Association; ALA = delta-Aminolevulinic Acid; GI = Gastrointestinal; HCV = Hepatitis C Virus; IPNET = International Porphyria Network; PBG = Porphobilinogen; PCT = Porphyria Cutanea Tarda

Common Oversights

CLINICAL OVERSIGHT	CONSEQUENCE	BETTER PRACTICE
Ordering total urine porphyrins instead of urine PBG to screen for AHP[7,13]	Mild nondiagnostic elevations cause over-diagnosis; true attacks are missed	Order spot urine **PBG + ALA + creatinine**; reserve total porphyrins for cutaneous work-up
Attributing chronic abdominal pain, neuropathy, or psychiatric symptoms in an elder to common geriatric causes[4]	Diagnostic delay that can span years and miss treatable disease	Keep porphyria on the differential for **unexplained recurrent neurovisceral symptoms**

CLINICAL OVERSIGHT	CONSEQUENCE	BETTER PRACTICE
Treating HCC surveillance as cirrhosis-only (standard AASLD protocol)[7]	AHP-associated HCC, which occurs **without cirrhosis**, goes undetected	Biannual liver ultrasound from age 50 for ALL AHP patients regardless of fibrosis; AFP is insensitive in non-cirrotic, early stage HCC[22]
Assuming normal between-attack urine PBG excludes AHP[7]	Sporadic AIP missed in **15-44%** of asymptomatic intervals	Repeat biochemical testing during an active attack **before** excluding the diagnosis
Overlooking declining eGFR and rising blood pressure as porphyria sequelae	Progressive CKD (up to **59%**)[17] and hypertension (**43%**)[7] go unmonitored	Track eGFR and BP at least annually; refer nephrology for **eGFR decline >5 mL/min/1.73m2/year**[24]

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Key Differentials in Elderly

DIFFERENTIAL	DISTINGUISHING FEATURES
Other acute hepatic porphyrias (AIP vs HCP vs VP vs ALAD)[4,7]	Clinically indistinguishable attacks; **differentiated by urine/fecal/plasma porphyrin patterns**; VP and HCP may add photosensitivity
Guillain-Barre syndrome[4]	Ascending weakness **without preceding abdominal pain**; PBG normal and CSF protein elevated (opposite of AHP)
Lead poisoning[4]	Anemia **(absent in AHP)**, elevated ALA and coproporphyrin III with normal PBG, decreased erythrocyte ALA-dehydratase
Common causes of acute abdomen (surgical, biliary, obstructive)[7]	**Peritoneal signs and abnormal cross-sectional imaging point away from porphyria**, which has a benign exam
Pseudoporphyria[14]	**Bullae resembling PCT but with normal plasma/urine porphyrins**; often NSAID-related or seen in renal failure
Secondary porphyrin elevations (liver disease, iron deficiency, CKD)[4]	Mild **non-specific** porphyrinuria without a matching porphyria pattern;[3] interpret in clinical context

DIFFERENTIAL	DISTINGUISHING FEATURES

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; AIP = Acute Intermittent Porphyria; ALA = delta-Aminolevulinic Acid; ALAD = ALA-Dehydratase-Deficiency porphyria; CKD = Chronic Kidney Disease; CSF = Cerebrospinal Fluid; HCP = Hereditary Coproporphyria; NSAID = Nonsteroidal Anti-Inflammatory Drug; PBG = Porphobilinogen; PCT = Porphyria Cutanea Tarda; VP = Variegate Porphyria

Comorbidity Screening

COMORBIDITY TO SCREEN	FREQUENCY/SIGNAL IN PORPHYRIA	ACTION
Chronic kidney disease[17]	Up to **59%** of symptomatic AIP; annual eGFR decline about 1 mL/min/1.73m^2; **PEPT2 variant OR ~6.85 for eGFR <60**[17,20]	**Annual creatinine/eGFR**; nephrology referral for accelerated decline
Hepatocellular carcinoma[22]	Annual incidence about **0.3%** in AHP (~0.4% in AIP), rising to **1.7-2.1%/year** in biochemically active AIP >50; risk increases ~**80-fold** after age 50[4,14,15]	**Biannual liver ultrasound from age 50**; expedite hepatology for any new lesion (AFP unreliable)
Hypertension and cerebrovascular disease	Hypertension in **43%**; elevated U-PBG carries an adjusted HR of about **1.40** for cerebrovascular disease[7,18]	**BP at every visit**; porphyria-safe antihypertensives (ACE inhibitor, CCB)
Chronic neuropathy[4,7]	Over **50%** of recurrent-attack patients report chronic neurologic symptoms; **35%** have diagnosed neuropathy	Distinguish from **diabetic/B12 causes**; document residual deficits
Psychiatric comorbidity (depression, anxiety, insomnia)	High rates in recurrent attacks; **46%** report daily symptoms[9,18]	**Screen and treat with porphyria-safe agents**; avoid **misattribution to dementia/delirium**
Kidney cancer (non-hepatic malignancy)	Higher in AHP than general population (**0.8% vs 0.2%**, p<0.001 in a Swedish cohort)[18]	**Maintain surveillance awareness**; investigate unexplained hematuria or renal masses

ABBREVIATIONS: AFP = Alpha-Fetoprotein; AHP = Acute Hepatic Porphyria; AIP = Acute Intermittent Porphyria; BP = Blood Pressure; CCB = Calcium Channel Blocker; eGFR = estimated Glomerular Filtration Rate; HR = Hazard Ratio; OR = Odds Ratio; PBG = Porphobilinogen; PEPT2 = Peptide Transporter 2; U-PBG = Urinary Porphobilinogen

⚠ RED FLAG — EMERGENCY/ACTIVATE CRISIS RESPONSE

Activate emergency transfer for any of the following in a known or suspected porphyria patient:

- Severe or progressive **motor neuropathy** with limb or **respiratory-muscle weakness** (risk of **respiratory failure**)
- **Seizures** during an acute attack, which may signal **posterior reversible encephalopathy syndrome** — **phenytoin** and **barbiturates** are porphyrinogenic and **contraindicated**; use **benzodiazepines** or **levetiracetam**
- **Severe hyponatremia** (**Na <125 mEq/L**) with **altered mental status**
- **Bulbar weakness** or **respiratory compromise** requiring **ICU-level care**

⚠ RED FLAG — URGENT (24-72 HOURS)

Arrange urgent (same-day to 72-hour) specialist contact for:

- Confirmed acute attack (**urine PBG ≥5× ULN**) with moderate-to-severe pain — **IV hemin** should begin within **~24 hours**
- New **tachycardia** and **hypertension** with abdominal pain in a known porphyria patient
- Acute **psychiatric disturbance** (psychosis, confusion, hallucinations) that may represent a **neurovisceral attack** rather than primary psychiatric illness
- New **liver lesion** on surveillance ultrasound in an AHP patient **≥50 years** — expedite cross-sectional imaging
- **Recurrent attacks**
    - Refer for prophylaxis evaluation
    - **Earlier referral is preferred** over waiting for a fixed attack threshold, to reduce cumulative hospitalization and complication risk

3 MEAT DOCUMENTATION ESSENTIALS

Porphyria is documented well when the note includes naming the **subtype**, tracking **organ complications**, and re-checking **medication safety**. The **MEAT** framework (**Monitor, Evaluate, Assess/Address, Treat**) provides a structured approach to documenting ongoing clinical management of the diagnosis.

*Case vignette: A female patient, 71, with biopsy- and biochemistry-confirmed acute intermittent porphyria after several younger-adult attacks, now attack-free, presents for an annual visit. Her note records "acute intermittent porphyria (AIP), **E80.21**, attack-free, actively managed," documents the year's liver ultrasound and eGFR, lists a medication review against the APF Drug Safety Database with no porphyrinogenic agents, and notes **counseling to avoid fasting and alcohol**. One year later a covering clinician sees the active AIP problem, checks a new antibiotic*

against the database before prescribing, and avoids a sulfonamide that would have risked an attack.

MONITOR: "Acute intermittent porphyria (AIP), **E80.21**, attack-free since young adulthood. Urine ALA/PBG: stable, last checked (date). Renal function: **eGFR 68 mL/min/1.73m^2** (CKD-EPI), stable from prior. Blood pressure: **132/78 mmHg**. Liver ultrasound (date): no new lesions, surveillance continued per AHP protocol. Medication review against APF Drug Safety Database: no porphyrinogenic agents identified."

EVALUATE: "ALA/PBG remain within normal limits between attacks — not a high excretor. eGFR stable over the past 12 months, no evidence of progressive decline. Blood pressure at goal (**<140/90 mmHg**). Liver ultrasound unchanged from prior surveillance imaging; no new findings warranting cross-sectional follow-up."

ASSESS: "Acute intermittent porphyria (AIP), attack-free, actively managed — **E80.21**. No active neurovisceral attack. No evidence of CKD progression or hepatic complication on current surveillance."

TREAT: "Continued trigger avoidance counseling: fasting, alcohol, and porphyrinogenic medications reviewed and reinforced. Medication reconciliation completed against APF database prior to any new prescription. **Follow-up: annual liver ultrasound, annual eGFR/blood pressure check, and urine ALA/PBG if symptomatic.** Patient educated to contact the clinic before starting any new medication, including from covering or specialist clinicians."

Clinical Documentation Elements

- **Name the specific porphyria subtype** (e.g., "acute intermittent porphyria") and its **diagnostic basis** - biochemical, genetic, or both - rather than "porphyria" alone
- **State current activity status:** attack-free, sporadic attacks, recurrent (**≥4/year**), or asymptomatic high excretor; the recurrent threshold drives prophylaxis decisions
- **Document the year's medication reconciliation against the APF Drug Safety Database**, noting that every new prescription was checked
- **Record complication surveillance actually performed:** biannual liver ultrasound from age 50, annual eGFR, and blood pressure
- **For PCT, document the trigger profile** (HCV, alcohol, estrogen, iron overload) and the **active treatment** (phlebotomy or low-dose HCQ)
- **Link each managed complicatio**n (CKD, hypertension, neuropathy) to its own code and plan so the record reflects the full clinical complexity

Reframing Common Documentation Shortcuts

COMMON SHORTCUT	WHY IT FALLS SHORT	STRONGER DOCUMENTATION
"History of porphyria"	Reads as **resolved/inactive** and **breaks annual documentation** and **drug-safety visibility**	"Acute intermittent porphyria (AIP), currently attack-free, actively managed - medications reviewed against APF database"
"Porphyria" without a subtype	Defaults to **E80.20** and tells the care team nothing about drug safety or monitoring	**Name the subtype** (AIP, HCP, VP, PCT) with **its diagnostic basis**
"Avoid unsafe drugs"	A vague instruction in a patient on 5-12 medications; **hundreds of agents are porphyrinogenic**	"All new prescriptions checked against the **APF Drug Safety Database**; no porphyrinogenic agents on current list"
"HCC screening per routine"	Implies **cirrhosis-based AASLD screening**, which misses non-cirrhotic AHP HCC	"Biannual liver ultrasound from age 50 for AHP regardless of cirrhosis; AFP unreliable"
"Stable" with no detail	Does not show monitoring, evaluation, or active management	Record the **eGFR, BP, ultrasound result**, and medication review that justify the assessment

ABBREVIATIONS: AASLD = American Association for the Study of Liver Diseases; AFP = Alpha-Fetoprotein; AHP = Acute Hepatic Porphyria; AIP = Acute Intermittent Porphyria; APF = American Porphyria Foundation; HCC = Hepatocellular Carcinoma; HCP = Hereditary Coproporphyria; PCT = Porphyria Cutanea Tarda; VP = Variegate Porphyria

DOCUMENTATION REMINDER IS PATIENT SAFETY

Documentation that names the porphyria subtype, its activity status, and the year's medication-safety review is what keeps a covering clinician from prescribing a porphyrinogenic drug. **Write the subtype, the activity status, and the database check at least once a year.** Accurate documentation here is patient safety.

4 TREATMENT AND REFERRAL QUICK GUIDE

Treatment approaches differ by porphyria family and should not be conflated. Acute hepatic porphyria is managed by removing triggers, treating attacks (**IV dextrose** for mild episodes, **IV hemin** for moderate-to-severe episodes), and preventing recurrent attacks with **givosiran**. Porphyria cutanea tarda is managed by removing iron and other precipitating triggers through **phlebotomy** or low-dose **hydroxychloroquine**.

Therapy Escalation Criteria

ESCALATION TRIGGER	THRESHOLD	ACTION*
Mild acute attack[7]	Oral-manageable pain, no paresis or hyponatremia[13]	IV dextrose (10% glucose, **≥300 g/day**); discontinue porphyrinogenic drugs; safe analgesia/antiemetics
Moderate-to-severe acute attack[7,15]	IV opioids required, motor neuropathy, hyponatremia, seizures, or psychiatric/autonomic features	IV **hemin 3-4 mg/kg/day x 3-5 days** via central access; treat electrolytes, BP, pain
Recurrent attacks[7]	**≥4 attacks/year**, biochemically and genetically confirmed AHP	Refer for prophylaxis: subcutaneous **givosiran 2.5 mg/kg** monthly (or off-label prophylactic hemin)[25]
Refractory disease[7]	Intractable attacks despite optimized therapy	**Refer to hepatology/transplant center**; liver transplantation can be curative
PCT, all patients[14]	At diagnosis	**Cease alcohol, estrogen, smoking**; treat HCV if present (can resolve disease)[21]
PCT, active disease[14]	Symptomatic skin disease or elevated porphyrins	Phlebotomy or low-dose **hydroxychloroquine** (100 mg twice weekly) until porphyrins normalize[26]

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; BP = Blood Pressure; HCV = Hepatitis C Virus; IV = Intravenous; PCT = Porphyria Cutanea Tarda
***Consult FDA labels for the most up-to-date dosage information, contraindications, and drug-drug interactions**

AGA/IPNET Guideline-Aligned Treatment by Patient Profile

PATIENT PROFILE	GUIDELINE-ALIGNED THERAPY*	NOTES AND GERIATRIC MODIFICATIONS
AHP, mild acute attack[7,15]	IV dextrose (10% glucose **≥300 g/day**); stop porphyrinogenic drugs; opioids and ondansetron for symptoms	Dose opioids conservatively for fall risk and renal clearance; **watch for SIADH/diuretic-related hyponatremia**
AHP, moderate-to-severe attack[7]	IV **hemin** (Panhematin) **3-4 mg/kg/day x 3-5 days** via PICC or central port	Central access can be hard in frail patients; cumulative **iron overload** with repeated courses - monitor ferritin
AHP, recurrent (≥4/year)[25]	**Givosiran 2.5 mg/kg SC monthly** (reduce to 1.25 mg/kg if ALT ≥3x ULN)[27]	ENVISION: **74%** reduction in annualized attack rate (3.2 vs 12.5/yr, P <0.001); consider B6 for elevated homocysteine
AHP, refractory[7]	Liver transplantation (**curative**);	Full frailty assessment before transplant evaluation in elderly
PCT, all patients (Step 1)[14,21]	Cease alcohol, estrogen, smoking; treat HCV; sun protection	**HCV therapy is well tolerated in older adults and can resolve PCT**
PCT, iron depletion[26]	**Phlebotomy 450 mL every 2 weeks** until ferritin ~20 ng/mL (median remission ~6.9 months)	**Poorly tolerated in frail, anemic, or cardiovascularly compromised elders** – prefer HCQ in these cases
PCT, drug therapy[26]	Low-dose **hydroxychloroquine 100 mg TWICE WEEKLY** (median remission ~6.1 months)	**NOT** the standard 400 mg/day rheumatologic dose, which is hepatotoxic in PCT; baseline + annual ophthalmic exam

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; ALT = Alanine Aminotransferase; eGFR = estimated Glomerular Filtration Rate; HCQ = Hydroxychloroquine; HCV = Hepatitis C Virus; PCT = Porphyria Cutanea Tarda; PICC = Peripherally Inserted Central Catheter; SC = Subcutaneous; SIADH = Syndrome of Inappropriate Antidiuretic Hormone; ULN = Upper Limit of Normal
***Consult FDA labels for the most up-to-date dosage information, contraindications, and drug-drug interactions**

CLINICAL PEARL - PCT HYDROXYCHLOROQUINE DOSE

The standard rheumatologic hydroxychloroquine dose (400 mg/day) is **hepatotoxic in PCT**. The porphyria-specific dose is **100 mg twice weekly only** - equally effective as phlebotomy.

Non-Pharmacologic Care and Supportive Interventions

INTERVENTION	RATIONALE	PRACTICAL NOTE
Systematic drug-safety auditing[7]	Porphyrinogenic exposure is the leading preventable AHP trigger	Check the **APF Drug Safety Database** for every new prescription at every encounter[23]
Nutrition/avoid fasting[15]	**Caloric restriction, fasting**, and **low-carb/keto diets** precipitate attacks; sulfur/preservative-rich foods and garlic **may also trigger**	Coordinate dietitian support; **minimize procedural NPO windows** and **provide carbohydrate cover**
Alcohol cessation[14,15]	Directly porphyrinogenic and a PCT driver	First-line PCT therapy; manage cessation carefully in frail elders
Sun protection (cutaneous porphyrias)[14]	Photosensitizing porphyrins cause skin injury	Physical barriers and **visible-light protection**; **essential** for PCT and EPP
Medical alert identification[15]	Ensures emergency teams avoid porphyrinogenic drugs	**Carry a card/bracelet naming the porphyria type** and a specialist contact

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; APF = American Porphyria Foundation; EPP = Erythropoietic Protoporphyria; NPO = Nil Per Os (nothing by mouth); PCT = Porphyria Cutanea Tarda

Medication Safety and Key Interactions

DRUG/CLASS	PORPHYRIA STATUS	ACTION*
Phenytoin, phenobarbital, carbamazepine[7,13]	**UNSAFE** - potent ALAS1 inducers (approved agents, contraindicated in AHP)	**Avoid in AHP**; for attack-related seizures use benzodiazepines or levetiracetam; verify in APF database[23]
Sulfonamide antibiotics (e.g., trimethoprim-sulfamethoxazole)[14,23]	**UNSAFE** - porphyrinogenic (approved agent, avoid in AHP)	Use **amoxicillin** or a **cephalosporin** for common infections; a frequent high-risk substitution error in elders

DRUG/CLASS	PORPHYRIA STATUS	ACTION*
Rifampin[15,21]	**UNSAFE** - potent CYP450 inducer (approved agent, avoid in AHP)	Consult a porphyria specialist if needed for **active TB**
Estrogen/progestin[14,21]	**UNSAFE** - hormonal ALAS1 induction; direct PCT trigger	**Avoid in AHP**; discontinue in PCT as part of Step 1
Givosiran[25,27]	**Approved** (indicated for confirmed AHP with **≥4 attacks/year**)	**ALT ≥3x ULN in 15%**; monitor LFTs, creatinine, homocysteine, amylase/lipase
Hydroxychloroquine for PCT[14,26]	**Approved but dose-critical**	**100 mg twice weekly only** - 400 mg/day is hepatotoxic in PCT; ophthalmic monitoring
Opioids (morphine, oxycodone, hydromorphone, fentanyl)[15]	**SAFE - no porphyrinogenic risk**	Required for attack pain; dose conservatively for renal function/fall risk; fentanyl preferred in renal impairment
ACE inhibitors/calcium channel blockers; ondansetron[15,23]	**Generally SAFE but with intra-class variation** — captopril, lisinopril, and diltiazem showed minimal porphyrin accumulation; **enalapril and nifedipine were substantially porphyrinogenic in vitro**[28]	First-line **antihypertensives and antiemetic**; confirm the specific agent before prescribing

ABBREVIATIONS: ACE = Angiotensin-Converting Enzyme; AHP = Acute Hepatic Porphyria; ALAS1 = delta-Aminolevulinic Acid Synthase 1; ALT = Alanine Aminotransferase; APF = American Porphyria Foundation; CYP450 = Cytochrome P450; LFT = Liver Function Test; PCT = Porphyria Cutanea Tarda; TB = Tuberculosis; ULN = Upper Limit of Normal
***Consult FDA labels for the most up-to-date dosage information, contraindications, and drug-drug interactions**

When to Refer

SITUATION	REFER TO	TIMING
Confirmed acute attack needing hemin[29]	Porphyria specialist or hepatologist	Same day; hemin within ~24 hours
Recurrent attacks (≥4/year)[7]	Porphyria specialist (for givosiran)	**Expedited** (within days)
New liver lesion on surveillance, or HCC concern[7]	Hepatology	**Expedited** cross-sectional imaging

SITUATION	REFER TO	TIMING
Declining eGFR (>5 mL/min/1.73m2/year)[30]	Nephrology	Routine-to-expedited
Suspected new porphyria with negative standard work-up[13]	Porphyria specialist after urine PBG/ALA testing	**Expedited**
PCT with HCV or significant iron overload[31]	Hepatology/phlebotomy service	Routine

ABBREVIATIONS: ALA = delta-Aminolevulinic Acid; eGFR = estimated Glomerular Filtration Rate; HCC = Hepatocellular Carcinoma; HCV = Hepatitis C Virus; PBG = Porphobilinogen; PCT = Porphyria Cutanea Tarda

Follow-Up Timing

CONTEXT	PARAMETER	INTERVAL
Known AHP, stable[7]	eGFR/creatinine, BP, urine ALA/PBG (if high excretor)	At least annually
AHP, age ≥50[7]	Liver ultrasound (HCC surveillance)	Every **6 months**
On givosiran[27]	LFTs, then creatinine/homocysteine/amylase/lipase	LFTs **monthly x6 months**, others every **3 months x6 months**
PCT on phlebotomy[31]	Ferritin/transferrin saturation	Every **2-4 weeks** until target
PCT, disease activity[32]	Plasma/urine uroporphyrin	Every **3 months** and **at suspected relapse**
PCT on hydroxychloroquine[4,33]	Ophthalmic examination	Baseline, then annually
Any porphyria, polypharmacy[7]	Medication reconciliation vs APF database	**Every encounter**

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; ALA = delta-Aminolevulinic Acid; APF = American Porphyria Foundation; BP = Blood Pressure; eGFR = estimated Glomerular Filtration Rate; HCC = Hepatocellular Carcinoma; LFT = Liver Function Test; PBG = Porphobilinogen; PCT = Porphyria Cutanea Tarda

Comorbidity Management

COMORBIDITY	PRIMARY-CARE ROLE	PORPHYRIA-SPECIFIC CAUTION
Hypertension (43%)[7]	Treat to **<140/90 mmHg**; choose porphyria-safe agents	**ACE inhibitors and CCBs generally safe**; verify each agent in the APF database[28]

COMORBIDITY	PRIMARY-CARE ROLE	PORPHYRIA-SPECIFIC CAUTION
Chronic kidney disease (up to 59%)[7]	**Annual eGFR**; manage as standard CKD; nephrology for decline	Givosiran can worsen eGFR - reassess risk/benefit in pre-existing CKD[27]
Chronic neuropathy[7]	Document and manage neuropathic pain with safe agents	**Distinguish from diabetic/B12 causes**; avoid porphyrinogenic neuropathic agents
Depression/anxiety/ insomnia[7]	Screen and treat; coordinate behavioral health	Select **porphyria-safe psychotropics**; avoid misattribution to dementia
Iron overload (iatrogenic, repeated hemin)[7]	**Track ferritin**; arrange phlebotomy if tolerated[1]	**Phlebotomy may be poorly tolerated in frail** or anemic elders

ABBREVIATIONS: ACE = Angiotensin-Converting Enzyme; APF = American Porphyria Foundation; B12 = Vitamin B12; CCB = Calcium Channel Blocker; CKD = Chronic Kidney Disease; eGFR = estimated Glomerular Filtration Rate

Cost-Smart Options

BRAND (EST. COST)	COST-SMART OPTION	EST. SAVINGS	COST-SMART TIP
Givlaari (givosiran) SC injection, brand (~$42,884/vial cash price)[34]	No generic exists; **IV hemin** (Panhematin) prophylaxis, lower-cost alternative for recurrent AHP (**~$9,126–$11,689/vial** cash price)[35]	Real-world cost-of-care study found **hemin averaged 78% lower annual total cost** than givosiran (**$134,798 vs $616,911/ year**)[36]	Reserve givosiran for confirmed recurrent AHP (**≥4 attacks/year**); requires biochemical/ genetic confirmation and prior authorization
Brand hydroxychloroquine (Plaquenil) (~$654/30-day supply)[37]	Generic **hydroxychloroquine** (**~$13–$116/30-day supply** cash)[38]	**~$540–640/month**	**Generic hydroxychloroquine is bioequivalent to brand**; no clinical indication favors brand-name prescribing
Generic hydroxychloroquine, ongoing therapy (~ $13–116/month)[38]	**Therapeutic phlebotomy**, CPT **99195** (**~$87/session**, Medicare non-facility national average)[39]	Variable; phlebotomy avoids recurring drug cost	Phlebotomy and low-dose HCQ show comparable efficacy for iron depletion in PCT; reserve HCQ for patients who cannot tolerate phlebotomy (poor venous access, anemia)

BRAND (EST. COST)	COST-SMART OPTION	EST. SAVINGS	COST-SMART TIP
Brand Mavyret (glecaprevir/ pibrentasvir) for HCV-associated PCT (~$12,877–$14,101 discounted cash price per 8–12-week course)[40,41]	**No generic currently available** in the US	N/A	Treating underlying HCV in PCT can resolve cutaneous disease and reduce long-term liver risk; coordinate antiviral selection with hepatology

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; HCQ = Hydroxychloroquine; HCV = Hepatitis C Virus; PCT = Porphyria Cutanea Tarda; SC = subcutaneous; IV = intravenous; CPT = Current Procedural Terminology; WAC = wholesale acquisition cost

Patient Education and Adherence

- Teach the **cardinal warning signs** of an AHP attack: **sudden severe abdominal pain**, **dark red or brown urine**, **muscle weakness or difficulty walking**, and **rapid heart rate with elevated blood pressure**. Instruct patients to seek **emergency care** immediately if these occur
- Every patient should carry **medical-alert identification** naming the **porphyria subtype** and a **specialist contact**
- Counsel on **trigger avoidance**: no alcohol, no fasting, and no skipping meals
- Any new medication (**prescription or over-the-counter**) must be checked against the **APF Drug Safety Database** before use; patients should be empowered to do this themselves and to contact the clinic before accepting a prescription from any provider, including covering clinicians and specialists

DOCUMENTATION REMINDER - PATIENT EDUCATION

Document the patient-education you provided; trigger avoidance, attack warning signs, and the medical-alert plan, so the whole care team can reinforce it. Document that the patient was instructed to ask any prescriber or pharmacist to check the **APF Drug Safety Database** before starting a new medication.

Quality Metrics Tie-In

MEASURE (PROGRAM)	STANDARD	RELEVANCE TO PORPHYRIA
Controlling Blood Pressure (HEDIS CBP; MIPS #236)	**Denominator:** patients 18-85 with hypertension (≥2 outpatient visits with HTN diagnosis in MP or year prior) **Numerator:** most recent BP <140/90 mmHg during MP **Exclusions:** ESRD, hospice, palliative care, pregnancy	**Hypertension affects up to 43% of acute hepatic porphyria (AHP) patients**, likely related to recurrent catecholamine surges during attacks.[7] Document use of a porphyria-safe antihypertensive (e.g., labetalol, amlodipine); avoid agents with porphyrinogenic risk per APF database
Comprehensive Medication Review (Part D MTM CMR)	**Denominator:** Part D enrollees meeting MTM eligibility criteria (multiple chronic conditions, multiple Part D drugs, annual cost threshold) **Numerator:** annual CMR completed with written summary provided to patient **Exclusions:** plan-specific opt-out	For AHP patients this is the single most actionable safety check — every CMR should include a cross-check of all medications against the APF unsafe drug database
Medication Reconciliation Post-Discharge (HEDIS TRC sub-measure 4; MIPS #46)	**Denominator:** patients ≥18 seen in outpatient setting within 30 days of inpatient discharge **Numerator:** discharge medication list reconciled with current medication list documented in outpatient record **Exclusions:** none specified	**Hospitalization introduces high risk of porphyrinogenic drug exposure** (anesthetics, antibiotics, anticonvulsants) prescribed without porphyria history flagged. Post-discharge reconciliation must include an explicit APF database check, not just standard med-rec
Kidney Health Evaluation (HEDIS KED; MIPS #488)	**Denominator:** patients 18-85 with diabetes diagnosis **Numerator:** eGFR AND uACR both completed during MP **Exclusions:** ESRD, dialysis	**MIPS #488 is diabetes-gated and does not directly apply to porphyria patients without comorbid diabetes** — flag as an internal benchmark instead: annual eGFR (plus uACR where clinically indicated) for all AHP patients, given CKD prevalence up to 59% in this population from recurrent attack-related nephrotoxicity[7]

MEASURE (PROGRAM)	STANDARD	RELEVANCE TO PORPHYRIA
Plan All-Cause Readmissions (CMS Stars PCR; MIPS #479)	**Denominator:** enrollees ≥18 with acute inpatient discharge **Numerator:** unplanned readmission within 30 days (lower is better) **Exclusions:** planned readmissions, hospice, psychiatric	Recurrent acute attacks are a major readmission driver in AHP; document attack diagnosis, prophylactic therapy initiation (e.g., givosiran), and a structured follow-up plan to reduce recurrence

ABBREVIATIONS: AHP = Acute Hepatic Porphyria; APF = American Porphyria Foundation; BP = Blood Pressure; CKD = Chronic Kidney Disease; eGFR = estimated Glomerular Filtration Rate; HEDIS = Healthcare Effectiveness Data and Information Set; MTM = Medication Therapy Management; uACR = urine Albumin-to-Creatinine Ratio

QUALITY OUTCOME

There is no porphyria-specific Star or HEDIS measure, but porphyria patients gain the most from the medication-safety and chronic-disease measures already in place. The highest-impact tactics for PCPs:

- **Check the APF Drug Safety Database** before every new prescription and at annual medication review — the single most effective attack-prevention behavior
- **Treat hypertension to goal** using porphyria-safe agents (e.g., labetalol, amlodipine)
  - Directly supports **CBP** performance
- **Monitor renal function** at recommended intervals to catch decline early
  - Directly supports **KED** performance
- **Reinforce trigger avoidance and warning-sign recognition** at every visit to reduce ED visits and avoidable readmissions

AAVBC PERSPECTIVE

Porphyria is diagnosed late not because it is biologically rare, but because the right tests are not always ordered at the right clinical moment. AAVBC supports recognition of the clinical trigger pattern that should prompt this action: recurrent severe abdominal pain accompanied by neurological symptoms such as weakness, psychiatric changes, or seizures; red or brown urine in the context of abdominal pain; and photosensitive skin changes in an adult without an established dermatological diagnosis. ***Porphyria is not a single disease, it encompasses distinct acute and cutaneous subtypes with different mechanisms, presentations, and management pathways, and mixed presentations can occur in the same patient.*** *In elderly patients, acute porphyria may present atypically as chronic abdominal pain, psychiatric symptoms, or neuropathy that is readily misattributed to more common geriatric conditions.*

AAVBC also encourages providers to avoid substituting genetic testing or total urine porphyrins for spot urine PBG, as neither is an appropriate first-line diagnostic step for an acute neurovisceral presentation. ***This helps ensure the right patients are tested with the right tool at the right time.***

5 CODING REMINDERS AND CASE EXAMPLES

Coding Specificity

ICD-10 CODE	DOCUMENTATION REQUIREMENT	COMMON ERROR
E80.21(Acute intermittent (hepatic) porphyria)	Name "acute intermittent porphyria" or "AIP" with biochemical/genetic basis	Defaults to **E80.20** when the subtype is known but not named
E80.29(Other porphyria (HCP, VP, EPP, X-linked))	Name the specific subtype (hereditary coproporphyria, variegate porphyria, etc.)	Used as a catch-all without naming the subtype
E80.1(Porphyria cutanea tarda)	Document "PCT"; specify familial vs. acquired if known; note active treatment	Missed when blistering skin is attributed to dermatoses without a metabolic work-up
E80.0(Hereditary (congenital) erythropoietic porphyria)	Document "congenital erythropoietic porphyria" or "CEP"	Confused with erythropoietic protoporphyria (EPP, coded **E80.29**)
E80.20(Unspecified porphyria)	Use only while the subtype is pending	Overused as a permanent code; replace once the subtype is confirmed

ABBREVIATIONS: AIP = Acute Intermittent Porphyria; CEP = Congenital Erythropoietic Porphyria; CNA = Community Non-Dual Eligible, Aged; EPP = Erythropoietic Protoporphyria; HCC = Hierarchical Condition Category; HCP = Hereditary Coproporphyria; ICD-10 = International Classification of Diseases, 10th Revision; PCT = Porphyria Cutanea Tarda; RAF = Risk Adjustment Factor; V28 = Version 28 CMS-HCC model; VP = Variegate Porphyria

Annual Clinical Review and Confirmation

- **Porphyria is a lifelong diagnosis that must be reaffirmed as a current, actively managed condition at least once each year** to support accurate HCC 50 documentation and to keep the diagnosis visible to every clinician who prescribes for the patient

- At the annual review, **restate the specific subtype by name** (not "history of porphyria" and not the unspecified **E80.20**), **record the current activity status, and document that medications were reconciled against the APF Drug Safety Database**
- **Tie the diagnosis to the surveillance actually performed -** biannual liver ultrasound from age 50, annual eGFR, and blood pressure - and to any complication being co-managed, each carrying its own code (for example **N18.-** for CKD, I10 for hypertension, **C22.0** for hepatocellular carcinoma)

Good Documentation - EHR Tips

EHR TIP	HOW IT HELPS	IMPLEMENTATION
Put the named subtype on the active problem list	Keeps **E80.21/E80.1** (not **E80.20**) visible to every prescriber	Pin the subtype with its diagnostic basis; **remove any "history of porphyria" wording**
Build a porphyria drug-safety alert	Prompts an **APF database** check at prescribing	Best-practice alert tied to the **E80.x** problem
Create a surveillance reminder set	Prevents missed biannual ultrasound and annual eGFR	Standing orders/recall for liver US from age 50 and annual renal labs
Use a SmartPhrase for the annual note	Documents subtype, activity status, and database check consistently	Template: **subtype + activity + APF review + surveillance results**
Flag givosiran monitoring	**Avoids missed LFT/renal/ homocysteine checks**	Order set: LFTs monthly **x6 months**; creatinine, homocysteine, amylase/ lipase **q3 months**

ABBREVIATIONS: APF = American Porphyria Foundation; eGFR = estimated Glomerular Filtration Rate; EHR = Electronic Health Record; LFT = Liver Function Test; US = Ultrasound

Brief Case Examples

SUCCESS - Named Subtype, Active Management, Coordinated Drug Safety

Patient: Female patient, 71, with biochemically and genetically confirmed acute intermittent porphyria (AIP), attack-free for years.

Documentation: Annual note reads "acute intermittent porphyria (AIP), **E80.21**, attack-free, actively managed"; records the year's liver ultrasound and eGFR; documents a medication reconciliation against the APF Drug Safety Database with no porphyrinogenic agents; notes counseling to avoid alcohol and fasting.

Outcome: When a covering clinician later considers an antibiotic, the active AIP problem and database habit prompt avoidance of a sulfonamide, preventing an attack. HCC 50 is accurately documented and the care team shares one clear plan.

PITFALL - Unspecified Coding and Omitted Complexity

Documentation as written: "History of porphyria - stable" carried forward each year, coded **E80.20** (unspecified).

Consequence: The note does not name the subtype, so the care team cannot tell which drug-safety protocol applies; biannual HCC surveillance is never scheduled because the record implies a resolved, inactive condition.

RAF Impact: E80.20 still maps to HCC 50 (CNA RAF 0.648), but the unspecified code and "history of" framing understate the active clinical complexity and break annual documented and care coordination.

Fix: Replace with the confirmed subtype code **(E80.21)**, document it as current and actively managed, record the APF database review, and order the biannual liver ultrasound from age 50.

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