Porphobilinogen (urine)

Description	Porphobilinogen (PBG) is a water-soluble, monopyrrole porphyrin precursor and part of the haem biosynthetic pathway. It is formed from two molecules of 5-aminolevulinic acid (ALA) in a reaction catalysed by the enzyme PBG synthase. Four molecules of PBG polymerase form the linear tetrapyrrole hydroxymethylbilane (HMB) in a reaction catalysed by the enzyme HMB synthase.
Indication	Measurement of urinary PBG is indicated in suspected cases of acute porphyria.
Additional Info	The porphyrias are a group of disorders resulting from the partial deficiency of enzymes involved in the haem biosynthetic pathway. The porphyrias can be divided into the acute porphyrias, associated with acute neuropsychiatric attacks, and the non-acute porphyrias, associated with cutaneous photosensitivity. The acute porphyrias include: acute intermittent porphyria (AIP), hereditary coproporphyria (HCP) and variegate porphyria (VP). Acute attacks occur when hepatic haem synthesis is induced in the presence of HMB synthase deficiency. In AIP there is a primary HMB synthase deficiency and in VP and HCP the deficiency is secondary. HMB synthase deficiency results in the accumulation of PBG and ALA during acute attacks. The renal threshold for PBG is low and so acute attacks are associated with excessive urinary excretion of PBG while concentrations remain low in the circulation. Measurement of urinary PBG is therefore used as a screening test in suspected cases of acute porphyria. Symptoms of acute attacks include: abdominal pain, nausea, vomiting, constipation, paraesthesia, weakness, confusion, depression, hallucinations and psychosis.
Concurrent Tests	n/a
Dietary Requirements	n/a
Interpretation	**During symptomatic acute attack:** Increased excretion of PBG is specific for the diagnosis of acute porphyria. Further confirmatory tests will be required to

	determine the specific diagnosis. If PBG excretion is normal when the patient is symptomatic then this probably excludes a diagnosis of acute porphyria. **After an acute attack or when patient is asymptomatic:** PBG excretion may be normal in asymptomatic patients and between acute episodes and so a diagnosis of acute porphyria cannot be excluded in these cases. If clinical suspicion of acute porphyria remains or if there is a family history then further tests will be required. **Please contact the laboratory for additional interpretation of results and advice on further investigations.**
Collection Conditions	A freshly voided, random urine sample should be collected during or immediately after an acute attack. The sample should be collected into a universal tube and **protected from light**. Please send the sample to the laboratory **immediately**.
Frequency of testing	As required.