

Suspected Porphyria:

How to proceed with testing

Symptoms suggestive of an acute porphyria attack (known or new porphyria patient, usually in the emergency room setting)

1) Urine Porphyria Screen (PORSU)- random urine sample protected against light - these results will be available within 4 hours and treatment can be initiated. No dietary restrictions for this test and the sample should be collected as soon as possible.

Symptoms suggestive of a chronic porphyria, with skin lesions or a history of an acute attack or current acute attack (new porphyria patient, usually in the outpatient or clinic setting but can also be collected in the emergency setting during an acute attack for the typing and diagnosis of porphyria)

2) Porphyria Study (PORST)- if the PORSU screening test is positive we recommend further testing on the urine and the collection of stool and blood.

PORST has the following dietary restrictions and are recommended for all stable and outpatient sample collections:

- **Duration of diet:** 3 days prior to collection of specimens and on the day of collection of specimens, if possible.
- **What NOT to eat:** No pork, red meat - including raw, cooked, biltong and extracts like marmite, oxo, soup.
- **Medicine not to take (if possible):** Griseofulvin, Acriflavine, Ethoxazene, Haemoglobin, Oxytetracycline, Phenazopyridine, Sulfamethoxazole, Tetracycline, Aminophylline, Antipyretics, Apronalide, Barbiturates, Carbromal, Cloral Hydrate, Chlordiazepoxide, Chlorpropamide, Diazepam, Dichlorophenazone, Ergot, Preparations, Estrogens, Ethanol, Ethoxazene, Glutethimide, Hexachlorobenzene, Hydantion, Derivatives, Isopropyl, Dipyrone, Meprobamate, Methyl dopa, Metyrapone, Phentazocine, Phenylhydrazine, Pregnancy, Progestogens, Succinimide, Sulfomethane, Sulfonamides, Vitamin K.

Family History of porphyria variegata (Outpatient setting)

3) Porphyria Variegata (POVPCR)- the South African Founder mutation R59W, this blood sample does not have to be protected against light. Useful for testing members of a family with a history of porphyria variegata (skin lesions and acute symptoms). This test is performed in Molecular Genetics.