Clinical Interpretation and Relevance of Porphyrin Precursor Results

Scenario 1

If Urine PBG is BELOW threshold (< 0.89umol/mmolcreatinine) the following Interpretive comment will be applied:

- Urine PBG:Creatinine ratio is WITHIN the reference range
- This level is BELOW threshold for acute porphyria attack.
- Note Pre-symptomatic porphyria cannot be ruled out.
- Further information is available on Labmed User Guide
- http://search.stjames.ie/Labmed/ search term PORPHYRINS.
- NB: Results can be affected by direct porphyria treatments

Clinical Relevance:

What is the rationale for requesting porphyrin analysis?

1. **Diagnosis of suspected acute porphyria attack/ ongoing clinical symptoms due to acute porphyria**

- A Urine PBG:Creatinine level < 0.89 ***umol/mmolcreatinine*** is not indicative of an acute porphyria attack. Therefore the patient's present clinical status cannot be attributed to a concurrent acute porphyria episode.
- However, if the patient had been recently treated with Haem Arginate or Glucose / Carbohydrate loading prior to the test sample being taken, then the interpretation of this result may have been affected by this action.
- In addition, if a suspected porphyria attack occurred in a timeframe extending a week or more before the date of sampling then interpretation of this result is more complex and would need to be discussed with the Porphyria service, Biochemistry Department, St James's Hospital
- Detailed interpretative information is also available through http://stjsharepoint:83/#3/21 (Prescribing guidelines)

2. Screening for Acute Hepatic Porphyria (AHP) in an asymptomatic patient

- This Urine PBG:Creatinine level < 0.89 ***umol/mmolcreatinine*** does not rule out the presence of a Pre-symptomatic porphyria i.e. an underlying AHP genetic susceptibility.
- If there is a definite family history of a confirmed AHP (including Acute Intermittent Porphyria, Variegate Porphyria or Hereditary Coproporphyria) then the appropriate screening for this condition should be undertaken in consultation with the Porphyria service, Biochemistry Department, St James's Hospital
- In addition, if there is a past history of an AHP diagnosis in this patient then appropriate screening for this condition should be undertaken in consultation with the Porphyria service, Biochemistry Department, St James's Hospital
- If indicated further investigation should include collecting a full range of biochemical and genetic samples i.e. Urine, Faecal, Plasma & Erythrocyte porphyrins and Porphyria genetics
- Consult LabMed User handbook for further information http://search.stjames.ie/Labmed/

Clinical Interpretation and Relevance of Porphyrin Precursor Results

<u>**Scenario 2**</u>

If Urine PBG is in a BODERLINE range (0.89 -2.36 umol/mmolcreatinine) the following Interpretive comment will be applied:

- Urine PBG:Creatinine ratio is in a BORDERLINE range.
- Suggest further porphyrin investigations AND
- Advise discussion with Porphyrin Laboratory.
- Information is also available on Labmed User Guide
- http://search.stjames.ie/Labmed/ search term PORPHYRINS.
- NB: Results can be affected by direct porphyria treatments.

Clinical Relevance:

What is the rationale for requesting porphyrin analysis?

1. **Diagnosis of suspected acute porphyria attack/ ongoing clinical symptoms due to acute porphyria**

- While a Urine PBG:Creatinine level between ***0.89 -2.36 umol/mmolcreatinine*** is NOT directly indicative of an acute porphyria attack per se, it does suggest the possible presence of an underlying Acute Hepatic Porphyria susceptibility (see comment under Screening for AHP). Therefore the patient's present clinical status cannot be directly attributed to a concurrent acute porphyria episode without further investigation.
- However, if the patient had been recently treated with Haem Arginate or Glucose / Carbohydrate loading prior to the test sample being taken, then the interpretation of this result may have been affected by this action.
- In addition, if a suspected porphyria attack occurred in a timeframe extending a week or more before the date of sampling then interpretation of this result is more complex and would need to be discussed with the Porphyria service, Biochemistry Department, St James's Hospital.
- Detailed interpretative information is also available through http://stjsharepoint:83/#3/21 (Prescribing guidelines)

2. Screening for Acute Hepatic Porphyria (AHP) in an asymptomatic patient

- This Urine PBG:Creatinine level between ***0.89 -2.36 umol/mmolcreatinine*** may indicate the presence of a Pre-symptomatic porphyria i.e. an underlying AHP genetic susceptibility.
- Therefore appropriate screening for AHP (including Acute Intermittent Porphyria, Variegate Porphyria or Hereditary Coproporphria) should be undertaken in consultation with the Porphyria service, Biochemistry Department, St James's Hospital
- Further investigation should include collecting a full range of biochemical and genetic samples i.e. Urine, Faecal, Plasma & Erythrocyte porphyrins and Porphyria genetics
- Consult LabMed User handbook for further information http://search.stjames.ie/Labmed/

Clinical Interpretation and Relevance of Porphyrin Precursor Results

<u>**Scenario 3**</u>

If Urine PBG is ELEVATED (>2.36 umol/mmolcreatinine) the following Interpretive comment will be applied:

- Urine PBG:Creatinine ratio is ELEVATED above the reference range.
- Result suggests an underlying acute porphyria disorder
- AND could represent an acute porphyria attack OR
- could be related to "Asymptomatic High Excretor" status.
- Advise discussion of result with Porphyrin Laboratory.
- Information is also available on Labmed User Guide
- http://search.stjames.ie/Labmed/ search term PORPHYRINS.
- NB: Results can be affected by direct porphyria treatments.

Clinical Relevance:

What is the rationale for requesting porphyrin analysis?

1. **Diagnosis of suspected acute porphyria attack/ ongoing clinical symptoms due to acute porphyria**

- A urine PBG:creatinine level >2.36 ***umol/mmolcreatinine*** strongly suggests the presence of an underlying Acute Hepatic Porphyria susceptibility
- While, this result could indicate that the patient is suffering an acute porphyria attack, perpetually elevated levels of Urine PBG:Creatinine are commonly seen in patients with a known confirmed AHP diagnosis where the condition is not clinically active. This is a state referred to as "Asymptomatic High Excretor" status
- Therefore, interpretation of elevated Urine PBG:Creatinine levels is complex and dependent on factors including the extent of the elevation, previous levels of PBG:Creatinine ratio, the current clinical presentation and any previous history of acute porphyria attacks.
- Further action based on this result should be undertaken in consultation with the Porphyria service, Biochemistry Department, St James's Hospital
- Please note if the patient had been recently treated with Haem Arginate or Glucose / Carbohydrate loading prior to the test sample being taken, then the interpretation of this result may have been affected by this action.

- In addition, if a suspected porphyria attack occurred in a timeframe extending a week or more before the date of sampling then interpretation of this result is more complex and would need to be discussed with the Porphyria service, Biochemistry Department, St James's Hospital
- Detailed interpretative information is also available through http://stjsharepoint:83/#3/21 (Prescribing guidelines)

2. Screening for Acute Hepatic Porphyria (AHP) in an asymptomatic patient

- This Urine PBG:Creatinine level >2.36 ***umol/mmolcreatinine*** strongly suggests the presence of a Pre-symptomatic porphyria i.e. an underlying AHP genetic susceptibility.
- Therefore appropriate screening for AHP (including Acute Intermittent Porphyria, Variegate Porphyria or Hereditary Coproporphria) should be undertaken in consultation with the Porphyria service, Biochemistry Department, St James's Hospital
- Further investigation should include collecting a full range of biochemical and genetic samples i.e. Urine, Faecal, Plasma & Erythrocyte porphyrins and Porphyria genetics
- Consult LabMed User handbook for further information http://search.stjames.ie/Labmed/