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Specialised Services Service Specification: CP166

National Acute Porphyria Services

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Statement

Welsh Health Specialised Services Committee (WHSSC) will commission the National Acute Porphyria Services in accordance with the criteria outlined in this document.

In creating this policy WHSSC has reviewed the requirements and standards of care that are expected to deliver this service.

Disclaimer

WHSSC assumes that healthcare professionals will use their clinical judgment, knowledge and expertise when deciding whether it is appropriate to apply this document.

This document may not be clinically appropriate for use in all situations and does not override the responsibility of healthcare professionals to make decisions appropriate to the circumstances of the individual patient, in consultation with the patient and/or their carer or guardian.

WHSSC disclaims any responsibility for damages arising out of the use or non-use of this document.

1. Introduction

This policy has been developed for the planning and delivery of National Acute Porphyria Services (NAPS) for people resident in Wales. This service will only be commissioned by the Welsh Specialised Services Committee (WHSSC) and applies to residents of all seven Health Boards in Wales.

1.1 Background

Porphyria is an inherited metabolic disorder where there is a problem with the production of haem within the body. Haem is mainly made in the liver and in red blood cells. Haem is used to make haemoglobin in red blood cells which transfer oxygen around the body.

There is a complex process that goes on in the liver and in red blood cells to make haem. The process has various steps and each step is controlled by a special protein called an enzyme. At each step, substances are made that are known as haem precursors. These are substances that are made during the process leading up to the making of haem. They include substances called porphyrins.

There are seven different types of porphyria. In each type, there is a lack of (or partial lack of) one of the enzymes that controls one of the steps in the making of haem. Because this enzyme is lacking, there is overproduction of haem precursors including porphyrins. The porphyrins and other precursors may then build up in the body and cause the various problems associated with porphyria.

Build-up of other haem precursors in the liver and elsewhere in the body causes the symptoms that occur in the acute attacks of porphyria.

The acute porphyrias include:

- Acute intermittent porphyria.
- Aminolaevulinic acid dehydratase porphyria (also known as plumboporphyria).

Symptoms of acute porphyria can vary. The most common symptom is a severe tummy (abdominal) pain. The nervous system is also commonly affected to cause symptoms such as muscle weakness and numbness in parts of the body. Acute porphyria can also cause mental health (psychiatric) problems including agitation, mania, depression and hallucinations.

The incidence and prevalence of this rare condition means that patients that suffer an attack are treated by highly specialised services. Nationally,

across the United Kingdom there was an active caseload of 137 patients¹ during 2016/17.

1.2 Aims and Objectives of the service

The aim of this specification is to enable:

- Immediate clinical support and advice from National Acute Porphyria Service (NAPS) to the acute care physicians in Wales, on the management and treatment of all acute attacks of porphyria.
- Telephone follow-up of patients who have recurrent attacks by the NAPS Porphyria Centre Specialist Nurse (PCSN).
- Access to NAPS clinics in Wales to improve the management of ongoing treatment and complications for patients with recurrent attacks.

The overall objective of this agreement is to improve the clinical management and care of NHS Wales patients with acute porphyria who experience an attack or attacks by providing immediate specialist clinical advice to acute care physicians, as well as clinically appropriate support on follow-up after discharge via telephone advice and specific NAPS clinics.

This service is targeted primarily at patients who experience acute porphyria attacks that require admission to hospital. The relevant patient demographic comprises mostly of young adults, the majority of whom are women, and whose wellbeing and quality of life is severely reduced by active porphyria.

1.3 Relationship with other documents

This document should be read in conjunction with the following documents:

- **NHS Wales**
 - All Wales Policy: [Making Decisions in Individual Patient Funding requests](#) (IPFR).
- **Relevant NHS England policies**
 - [Severe acute porphyria service \(All Ages\), E06/S\(HSS\)/e 2013/14 NHS Standard Contract](#)

¹ NHS England: Highly Specialised Services 2017

2. Service Delivery

The Welsh Health Specialised Services Committee will commission acute porphyria services for Welsh residents, in-line with the criteria identified in this policy.

2.1 Access Criteria

Welsh residents with a diagnosis of porphyria who have an acute attack and require out of hours advice and treatment. Haem arginate is the treatment of choice for severe or recurrent acute porphyria attacks, which is aimed at stopping the attack.

2.2 Service description

In addition to the standards required within the Contract, specific quality standards and measures will be expected. The provider must also meet the standards as set out below.

2.3 Service Components Provided by NAPS

The core components of the service provided by NAPS to NHS Wales are:

- Full access for acute care physicians in Wales to the immediate 24hour clinical advisory service for the management of acute attacks.
- Access to ad-hoc telephone advice to support the ongoing management of patients with recurrent porphyria attacks when necessary.
- Telephone follow-up of patients by the NAPS PCSN.
- Haem arginate required for urgent treatment, which will be couriered from the on-call porphyria centre (University Hospital of Wales, Cardiff or King's College Hospital, London).

2.4 Interdependencies with other services or providers

Service Components Provided by NHS Wales:

- Inpatient care for patients during acute porphyria attacks to be provided by acute care physicians within the patient's local hospital.
- Oversight of follow-up care by a NAPS Consultant with experience of treating porphyria, in collaboration with a named medical consultant in the patient's local hospital.
- Biochemistry Laboratory Services.
- Haem arginate provision this will be billed directly from the NAPS centre which provides the drug to the patient's host Health Board.

2.5 Acceptance Criteria

The service outlined in this specification is for patients ordinarily resident in Wales, or otherwise the commissioning responsibility of the NHS in Wales. This excludes patients who whilst resident in Wales, are registered with a GP practice in England, but includes patients resident in England who are registered with a GP Practice in Wales.

2.6 Patient Pathway (Annex i)

2.6.1 Isolated acute attacks

NHS Wales physicians suspecting an acute porphyria attack will contact the on-call National Acute Porphyria Centre via a 24-hour emergency telephone service² which is publicised in the British National Formulary (BNF)³, to discuss treatment including whether haem arginate if required

Before advising the use of haem arginate the consultant will confirm that:

- the symptoms and signs are consistent with an acute attack
- for new patients evidence of increased urine porphobilinogen excretion has been demonstrated.

The local clinical team will be offered guidance on:

- the management of the attack
- how to access consensus guidelines
- how to access haem arginate, and
- how to administer the drug.

The local clinical team will be offered outreach support from the NAPS Centre via telephone during the acute admission.

Follow-up care

On discharge, new patients will be referred to the NAPS consultant in Wales and will be invited to attend regular reviews with their consultant for two years after the initial attack.

Patients should be followed-up remotely at 6 weeks by the NAPS PCSN; as well as at 6, 12, 18 and 24 months after the initial attack at a NAPS clinic in Cardiff. The NAPS PCSN will feed back any issues to the NAPS consultants in Wales. The aim of the regular follow-up appointments is to:

- ensure appropriate and adequate rehabilitation for any residual neurological disability, delivered by the patients' health board

² National Acute Porphyria Centre 24 hour emergency telephone number via UHW Switchboard: 029 20747 747

³ <https://bnf.nice.org.uk/treatment-summary/acute-porphyrrias.html>

- reduce the risk of further attacks through education and safe treatment of any associated medical conditions
- ensure Clinical Genetics referral for family counselling and screening, and
- provide information about the patient support group (British Porphyrria Association⁴).

The NAPS consultants in Wales will also ensure any further attacks are promptly and appropriately treated through collaboration with a named local consultant and that sufficient information is forwarded to the NAPS PCSN to support the maintenance of a detailed database of all patients, treatments and monitoring for audit and review of outcomes.

Exit point

Patients not on haem arginate are referred back to the sole care of the local consultant after two years of experiencing no further acute attacks.

2.6.2 Recurrent acute attacks

Patients who have had four or more acute attacks requiring haem arginate in one year are followed up by the NAPS consultant in Wales on a long-term basis and a care plan is devised to prevent, or as a minimum, reduce the frequency of severe attacks.

Subsequent acute admissions

The local acute care physician should follow the acute pathway outlined in annex i and inform the patient's named consultant in Wales of the admission.

Follow-up care

Patients are seen at 3-6 monthly intervals in the Welsh NAPS clinic. The NAPS PCSN will provide additional telephone follow-up if required.

2.6.3 Children and Young People

Patients who present with a suspected acute porphyria attack before their 16th birthday will usually initially be assessed by a Paediatric Consultant or senior Paediatric trainee, who will urgently seek advice from an identified Consultant in Paediatric Inherited Metabolic Medicine via telephone. The on-call Consultant in Paediatric Inherited Metabolic Medicine will discuss the case with NAPS colleagues. If haem arginate is required, this will be prescribed and administered locally. Haem arginate will be requested via the NAPS service but billed directly to the patient's health board by the treating hospital.

⁴ <http://www.porphyrria.org.uk/>

Follow-up care

Paediatric patients will have their follow-up care overseen by a Consultant in Paediatric Inherited Metabolic Medicine. Patients between the ages of 16 to 18 years will transition to the adult follow-up pathway. This includes attendance at the NAPS clinics.

Recurrent acute attacks

As per the adult pathway but delivered by a Consultant in Paediatric Inherited Metabolic Medicine.

2.6.4 Inpatient Admission to NAPS

Inpatient admission for Welsh residents to a NAPS specialist centre can be arranged if required. However, this is outside the scope of this service agreement. If inpatient treatment is considered necessary by the acute care physician or named consultant in Wales, the Health Board will pay for it on a cost per case basis. Funding approval for in-patient treatment at a NAPS centre should be sought via IPFR by the referring consultant prior to or in the case of emergencies at the point of referral.

2.7 Service provider/Designated Centre

The National Acute Porphyria Service (NAPS) commissioned by NHS England, provided by Cardiff and Vale University Health Board and King's College London.

2.8 Exceptions

If the patient does not meet the criteria for treatment as outlined in this policy, an Individual Patient Funding Request (IPFR) can be submitted for consideration in line with the All Wales Policy: Making Decisions on Individual Patient Funding Requests. The request will then be considered by the All Wales IPFR Panel.

If the patient wishes to be referred to a provider outside of the agreed pathway, and IPFR should be submitted.

Further information on making IPFR requests can be found at: <http://www.whssc.wales.nhs.uk/individual-patient-funding-requests>

3. Quality and Patient Safety

The provider must work to written quality standard and provide monitoring information to the lead commissioner. The quality management systems must be externally audited and accredited.

The centre must enable the patients, carers and advocates informed participation and to be able to demonstrate this. Provision should be made for patients with communication difficulties and for children, teenagers and young adults.

3.1 Quality Indicators (Standards)

Outcome 1

Improve the overall management and treatment of patients admitted to NHS Wales acute receiving wards with (suspected) sporadic acute porphyria attacks through sustainable NAPS on-call consultant cover for NHS Wales clinicians:

- Length of hospital stay during attack.
- Mortality 12 months after attack onset.
- Absolute number of new sporadic patients resident in Wales.
- Number of clinical telephone enquiries to service from NHS Wales clinicians.

Outcome 2

Improve the overall management and review of treatment of patients suffering recurrent attacks:

- Number of acute porphyria patients in Wales attending 6 monthly outreach clinics.
- Number of acute porphyria patients in Wales receiving follow-up phone calls by the PCNS.

3.2 Other quality requirements

- the provider will have a recognised system to demonstrate service quality and standards
- the service will have detailed clinical protocols setting out nationally (and local where appropriate) recognised good practice for each treatment site
- the quality system and its treatment protocols will be subject to regular clinical and management audit
- the provider is required to undertake regular patient surveys and develop and implement an action plan based on findings.

4. Performance monitoring and Information Requirement

4.1 Performance Monitoring

WHSSC will be responsible for commissioning services in line with this policy. This will include agreeing appropriate information and procedures to monitor the performance of organisations.

For the services defined in this policy the following approach will be adopted:

- Service providers to evidence quality and performance controls
- Service providers to evidence compliance with standards of care

Each provider will provide monitoring returns on a 6 monthly basis, April-September and October to March. These will be submitted by the 21st October and 21st April respectively.

WHSSC will conduct performance and quality reviews on an annual basis

4.2 Key Performance Indicators

The providers will be expected to monitor against the full list of Quality Indicators derived from the service description components described in Section 2.2.

The provider should also monitor the appropriateness of referrals into the service and provide regular feedback to referrers on inappropriate referrals, identifying any trends or potential educational needs.

4.3 Date of Review

This document is scheduled for review before 2024, where we will check if any new evidence is available.

If an update is carried out the policy will remain extant until the revised policy is published.

5. Equality Impact and Assessment

The Equality Impact Assessment (EQIA) process has been developed to help promote fair and equal treatment in the delivery of health services. It aims to enable Welsh Health Specialised Services Committee to identify and eliminate detrimental treatment caused by the adverse impact of health service policies upon groups and individuals for reasons of race, gender re-assignment, disability, sex, sexual orientation, age, religion and belief, marriage and civil partnership, pregnancy and maternity and language (Welsh).

This policy has been subjected to an Equality Impact Assessment.

The Assessment demonstrates the policy is robust and there is no potential for discrimination or adverse impact. All opportunities to promote equality have been taken.

6. Putting Things Right: Raising a Concern

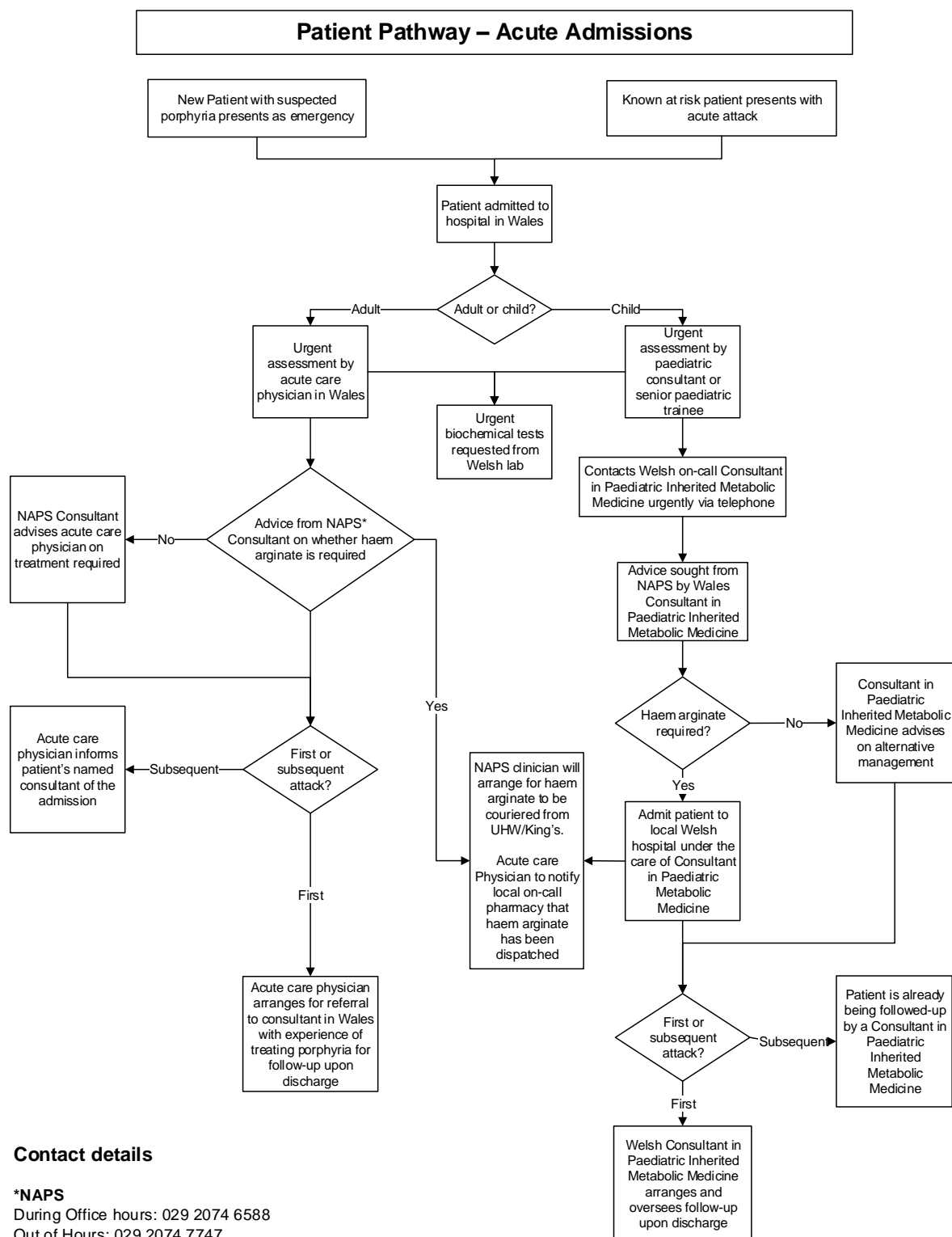
If the patient does not meet the criteria for treatment as outlined in this policy, an Individual Patient Funding Request (IPFR) can be submitted for consideration in line with the All Wales Policy: Making Decisions on Individual Patient Funding Requests. The request will then be considered by the All Wales IPFR Panel.

If an IPFR is declined by the Panel, a patient and/or their NHS clinician has the right to request information about how the decision was reached. If the patient and their NHS clinician feel the process has not been followed in accordance with this policy, arrangements can be made for an independent review of the process to be undertaken by the patient's Local Health Board. The ground for the review, which are detailed in the All Wales Policy: Making Decisions on Individual Patient Funding Requests (IPFR), must be clearly stated

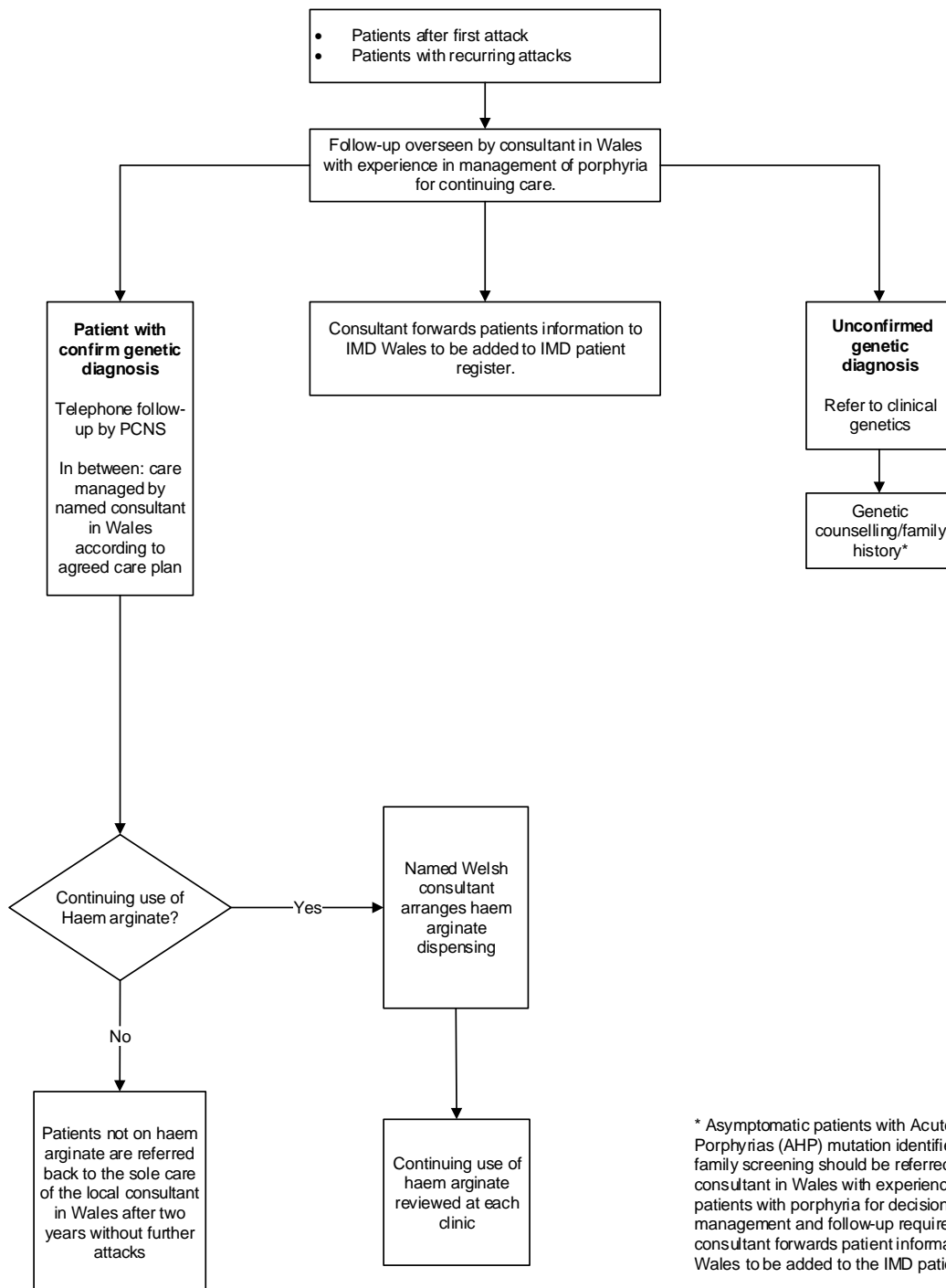
If the patient wishes to be referred to a provider outside of the agreed pathway, and IPFR should be submitted.

Further information on making IPFR requests can be found at: [Welsh Health Specialised Services Committee \(WHSSC\) | Individual Patient Funding Requests](#)

Annex i Patient Pathway



Patient Pathway – Follow-up care



* Asymptomatic patients with Acute Hepatic Porphyrria (AHP) mutation identified through family screening should be referred to a consultant in Wales with experience of treating patients with porphyria for decisions on case management and follow-up required. The consultant forwards patient information to IMD Wales to be added to the IMD patient register.

Annex ii Abbreviations and Glossary

Abbreviations

AWMSG	All Wales Medicines Strategy Group
IPFR	Individual Patient Funding Request
NAPS	National Acute Porphyria Services
PCSN	Porphyria Centre Specialist Nurse
WHSSC	Welsh Health Specialised Services

Glossary

Individual Patient Funding Request (IPFR)

An IPFR is a request to Welsh Health Specialised Services Committee (WHSSC) to fund an intervention, device or treatment for patients that fall outside the range of services and treatments routinely provided across Wales.

Welsh Health Specialised Services Committee (WHSSC)

WHSSC is a joint committee of the seven local health boards in Wales. The purpose of WHSSC is to ensure that the population of Wales has fair and equitable access to the full range of Specialised Services and Tertiary Services. WHSSC ensures that specialised services are commissioned from providers that have the appropriate experience and expertise. They ensure that these providers are able to provide a robust, high quality and sustainable services, which are safe for patients and are cost effective for NHS Wales.