| Agenda Item |
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# Joint Commissioning Committee

## Commissioning of Highly Specialised Services for Very Rare Diseases

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| **Dyddiad y Cyfarfod / Date of Meeting** | 21/01/2025 |
| **Statws Cyhoeddi / Publication Status** | Open/ Public |
| | Choose an item. |
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| **Pwrpas yr Adroddiad / Report Purpose** | For Approval |

| **Engagement (internal/external) undertaken to date (including receipt /consideration at Committee/Group)** | | |
|---|---|---|
| **Committee / Group / Individuals** | **Date** | **Outcome** |
| Management Group on 21.11.2024 | 21/11/24 | Endorsed |

| **Acronyms / Glossary of Terms** | |
|---|---|
| CPAG | Clinical Prioritisation Advisory Group |
| IPFR | Individual Patient Funding Requests |
| JCC | NHS Wales Joint Commissioning Committee |
| NHSE | National Health Service England |
| NICE | National Institute for Health and Clinical Excellence |
| NSD | National Services Directorate |
| RDIN | Rare Disease Implementation Network |
| WHSSC | Welsh Health Specialised Services Committee (former) |

## 1. SITUATION

The purpose of this report is to highlight to the JCC Members in relation to the number of medicines for very rare diseases in the NICE appraisal pathway, for which in Wales there is currently no clinical or financial planning. Although there is a widespread perception within Health Boards (HBs) that the JCC commissions highly specialised services for very rare diseases, the commissioning responsibilities are unclear. These medicines for very rare diseases will require funding and thus the options are that JCC commission these very rare highly specialised services; or HBs undertake the commissioning themselves, accepting that most HBs will have small numbers, typically 0 or 1 patient.

## 2. BACKGOUND

The former Welsh Health Specialised Services Committee (WHSSC), now the NHS Wales Joint Commissioning Committee (JCC) commissioned specialised health services for the people of Wales including some rare and very rare diseases. However, there was no rationality over which services WHSSC commissioned, and many highly specialised services for very rare diseases were not explicitly commissioned.

There are an increasing number of transformational medicines for very rare diseases, but without identified commissioning responsibility neither the JCC nor the HBs can effectively financially plan for these new treatments. With its expertise in rare diseases and medicines optimisation, this report suggests that the JCC is best placed to commission very rare diseases in Wales.

### 2.1 Rare and very rare diseases

Many specialised services are for people with rare conditions. A rare disease is defined as a condition which affects fewer than 1 in 2,000 people. Although rare diseases are individually rare, they are collectively common, with one in 17 people being affected by a rare disease at some point in their lifetime. In the UK, this amounts to over 3.5 million people and equates to about 190,000 people in Wales.

Within specialised services there are a group of highly specialised services for very rare diseases affecting a smaller number of patients. A very rare diseases is defined as a condition that affects fewer than 1 in 50,000 people and usually affecting no more than 500 patients in the UK (equates to 25 patients in Wales).

### 2.2 The burden of rare disease

Rare diseases can be both life-limiting and life-threatening, and disproportionately affect children. Seventy-five per cent of rare diseases affect children and more than 30% of children with a rare disease die before their fifth birthday. Those impacted with rare diseases are often diagnosed in early in life but are now living longer and more fulfilling lives due to significant developments in treatments.

## 2.3 Strategic context

On 9 January 2021, the UK Rare Diseases Framework was published https://www.gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework and included a joint Ministerial foreword by all four respective UK Health Ministers. The Framework was based on the outcomes of the 'National Conversation on Rare Diseases', launched in 2019. The conversation gathered views from across the rare disease community on the major challenges faced by people affected by rare conditions across the UK.

The Framework outlines the key priorities for the next five years to improve the lives of people living with rare diseases. Each nation committed to creating their own Action Plan to implement these priorities.

The Priorities are:

- Helping patients get a final diagnosis faster
- Increasing awareness of rare diseases among healthcare professional
- Better coordination of care, and
- Improving access to specialist care, treatments & drugs.

To turn these priorities into action, governance and oversight groups were established:

- UK Rare Disease Framework Board: Oversees the response to the Framework, and
- UK Rare Disease Forum: Focuses on engagement, with representatives from all four nations attending meetings to ensure collaboration and oversee the development of the Action Plans. The Forum uses a 'deep dive' format to tackle key priorities in depth, drawing on various spheres of expertise.

For Wales, the Rare Disease Implementation Network (RDIN) was formed. This network includes clinical experts, representatives of patient advocacy groups, NHS Wales, and third sector representatives, all working together to develop Wales's Action Plan. In June 2022, the Wales Rare Disease Action Plan was launched https://executive.nhs.wales/functions/networks-and-planning/rare-diseases/rdig-documents/rare-diseases-action-plan/ following on from the development of the Four Nations UK Rare Diseases Framework.

Specialised services nationally, often on a UK basis. Due to the small number of patients accessing such services, they are most appropriately delivered and coordinated nationally through a very small number of expert centres. This model of delivery makes it easier to recruit appropriately qualified professionals and to ensure that they receive the level of training needed to maintain their expertise; this ensures patient safety.

It also ensures the most effective use of resources through efficient management of patient care and ensuring access to the technology necessary to allow delivery of the services. In addition, it generates sufficient numbers of patients to make

clinical research in the UK viable. Examples of highly specialised services include liver transplant services, atypical haemolytic uraemic syndrome (adults and children), and lysosomal storage disorders service (children and adults).

Given the nature of very rare diseases and highly specialised services, it is helpful for the JCC (& NHS Wales) to get expert advice on these services from advisory groups established at a UK level.

### 2.5 Highly specialised services for very rare diseases in the UK

Until recently NHS England (NHSE) Specialised Commissioning commissioned 149 specialised services, with a spend in 2022/23 of £22.9 billion. However, from April 2023, NHSE set out a roadmap for devolving many of the less specialised services to regional commissioning, with integrated care boards given a bigger role in commissioning the higher volume services such as cardiac surgery. There remain however a residual 80 services that are considered to be so highly specialised that they are deemed worthy of commissioning at a national level and will remain under the auspices of NHSE specialised commissioning. Monitoring of clinical outcomes is a key responsibility of the NHSE Highly Specialised Commissioning team and a high level of clinical outcome monitoring is in place for these remaining 80 services. The highly specialised commissioning team work closely with the individual services and the NHS England Quality Team to ensure data completeness.

In Scotland the National Services Directorate (NSD) commissions more than 60 national designated specialist services on behalf of Scottish Government and NHS Scotland's HBs, but also specifically commissions the 80 highly specialised services as defined by NHSE.

In contrast most of the 80 highly specialised services recognised by NHSE and NSD are not formally commissioned in Wales, although the perception in many HBs is that they are. This results in considerable uncertainty, a lack of service or financial planning and inequity of access for patients.

If a medicine receives a positive Technology Assessment (TA) from NICE, there is a requirement for the medicine to be available for patients in Wales within 60 days. For conditions commissioned by the JCC, it will be responsible for commissioning the new medicine, otherwise the default would be for the HBs to provide the medicine.

### 2.6 The Bluteq system for highly specialised medicines

Most high cost medicines come with substantial rebates that need to be claimed back from the pharmaceutical company. The JCC utilise the online Blueteq system to track and ensure appropriate use of high cost medicines that it commissions. The use of Bluteq is central to identifying patients (and rebates) and pharmaceutical companies often mandate the use of Bluteq for rebates. Rebates can be greater than £300,000 for a single patient and thus accurate data collection is imperative to ensure all rebates are collected. The use of the Bluteq

system is standard within specialised services within NHS England. Most HBs in Wales have limited experience of use of Bluteq.

## 3. KEY RISKS / MATTERS FOR ESCALATION

### 3.1 Consequences of absence of highly specialised services policy

Historically, people with rare diseases were often therapeutic orphans. In the absence of specific or disease modifying treatments, care was usually just supportive. Thus, the absence of specialised commissioning for Welsh patients with very rare diseases was often imperceptible. The increasing number of novel, disease modifying and precision medicines for rare and very rare disease now puts the commissioning of highly specialised services centre stage. Because many highly specialised services have not been formally commissioned by the JCC, there has been no horizon scanning or financial planning within NHS Wales for these new treatments.

For example, epidermolysis bullosa (EB) is a very rare inherited skin disorder which can result in severe disability and decreased life expectancy. It is one of the 80 highly specialised services that NHSE Specialised Commissioning (and NHS Scotland) commissions – there are just 2 specialised paediatric centres in the UK. Although a highly specialised service, the former WHSSC did not commission EB services apart from in relation to an occasional Individual Patient Funding Request (IPFR).

In January 2024, NICE approved topical birch bark extract for moderate to severe EB (HST28) to be delivered in the two paediatric centres. Welsh Government requires that all medicines with a positive Technology Appraisal from NICE should be available within 60 days. Because EB was not commissioned by the former WHSSC or the JCC (even though HBs may have believed that they did), there was no clarity of planning or commissioning responsibility. Given the rarity of EB and that very few patients were eligible for treatment (there are three eligible patients in Wales) and the requirement for Bluteq approval, the JCC appeared to be best placed to take on the commissioning of EB. The JCC took on the commissioning of this and a policy is now in place.

Since birch bark extract for EB, there have been 3 further medicines for very rare diseases commissioned by NHSE Specialised Commissioners:

1. Burosumab for X-linked hypophosphataemic rickets in adults
2. Evinacumab for homozygous hypercholesterolaemia, and
3. Belzutifan for von Hippel Landau disease.

All have positive Technology Appraisals from NICE, and thus all need to be made available in Wales within 60 days.
Currently the JCC does not commission these very rare diseases there has been no horizon scanning, clinical planning or financial planning for these medicines to date.

Members should note that the advent of transformational medicines for rare and very rare diseases is set to significantly alter disease outcomes and patient experiences. It is critical the funding stream / principles for accessing treatments, associated clinical care and responsible commissioner are agreed in order to minimise the risks to patients, enabling the required financial planning and the implementation of the rare diseases strategy in Wales.

There is a steady flow of medicines for very rare diseases in the NICE appraisal pathway, for which in Wales there is currently no clinical or financial planning.

**The options are clear, Members are asked to approve that either the JCC plans for and commissions all of the highly specialised services; or HBs take on the commissioning themselves, accepting that most HBs will have 0 or 1 patients.**

Irrespective of the option chosen, the medicines will have to be made available to Welsh patients, and thus there are financial implications for HBs. Given the scale of some of the rebates, and the risk of failing to capture accurate data, the use of the Bluteq system is considered essential. It is recommended that JCC provide a clear directory of services and therapies commissioned by JCC and take on responsibility for the commissioning of very rare diseases in Wales in line with the proposed next steps. Members should note that this matter was discussed at the Management Group meeting on 21 November 2024 and they were very supportive that JCC was the appropriate body to commission very rare diseases and highly specialised services.
For people in Wales with very rare diseases it is suggested that:

- the JCC is best placed to plan and commission the highly specialised services they require
- the very rare diseases and highly specialised services as defined by NHSE and NHS Scotland are incorporated into the JCC portfolio
- the JCC Team horizon scan new medicines or technologies for very rare diseases
- the JCC develop a needs assessment of the number of patients with very rare diseases in Wales likely to eligible for new medicines or technologies
- the JCC financially plan for new medicines or technologies for very rare diseases in Wales, and
- the JCC commission the delivery of new medicines or technologies for very rare diseases and produce a policy position statement to confirm this.

## 4. ASSESSMENT

| **Objectives / Strategy** | |
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| **Dolen i Amcan (au) Strategol CBC / Link to JCC Strategic Objectives(s)** | Improve Equity and Population Health |
| | All JCC strategic objectives are applicable<br>Maximise value; ensure quality; reduce duplication; improve equity and population health. |

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| **Dolen i Ddeddf Llesiant Cenedlaethau'r Dyfodol – Nodau Llesiant / Link to Wellbeing of Future Generations Act – Wellbeing Goals** *150623-guide-to-the-fg-act-en.pdf (futuregenerations.wales)* | A Healthier Wales |
| | If more than one applies please list below:<br>A more equal Wales |
| **Dolen i Hwyluswyr Ansawdd** *(Canllawiau Statudol Dyletswydd Ansawdd (llyw.cymru)) /* **Link to Enablers of Quality** *(Duty of Quality Statutory Guidance (gov.wales))* | Data to Knowledge |
| | If more than one applies please list below:<br>All enablers of quality are applicable<br>Leadership; Culture and valuing people; learning, improvement and research; whole systems perspective |
| **Dolen i Feysydd Ansawdd** *(Canllawiau Statudol Dyletswydd Ansawdd (llyw.cymru)) /* **Link to Domains of Quality** *(Duty of Quality Statutory Guidance (gov.wales))* | Effective |
| | If more than one applies please list below:<br>All domains of quality are applicable<br>Efficient; equitable; person centred; timely and safe |
| **Effaith Amgylcheddol/ Cynaliadwyedd (5R) / Environmental /Sustainability Impact (5Rs)** | No - Not Applicable |
| | If more than one applies please list below: |

| **Impact Assessment** | | |
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| **Ansawdd** *Ydych chi wedi ymgymryd â Sgrinio Asesiad o'r Effaith ar Ansawdd? /* **Quality** *Have you undertaken a Quality Impact Assessment Screening?* | Yes: ☒ | No: ☐ |
| | Outcome:<br>As part of the policy development | If no, please include rationale below: |
| **Cydraddoldeb** *Ydych chi wedi ymgymryd â Sgrinio Asesiad o'r Effaith ar Gydraddoldeb? /* **Equality** *Have you undertaken an Equality Impact Assessment Screening?* | Yes: ☒ | No: ☐ |
| | Outcome:<br>An equality impact assessment was undertaken during the policy development. | If no, please include rationale below: |
| **Cyfreithiol / Legal** | Yes (Include further detail below) | |
| | There is a risk to the JCC as the policies extant are ambiguous and inequitable. | |

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| **Enw da / Reputational** | Yes (Include further detail below) |
| | Particularly to maintain services for patients in Wales to have the same access as other areas of the UK |
| **Effaith Adnoddau** *(Pobl /Ariannol) /* **Resource Impact** *(People / Financial)* | Yes (Include further detail below) |
| | The resource impact will depend on the commissioning decisions which are taken. The policy which went to stakeholder consultation was considered to be resource neutral.<br>The impact on resources will be dependent on the commissioning decisions made by the appropriate Commissioning group. |

## 5. RECOMMENDATIONS

The JCC is asked to:

- **Note** the report
- **Note** there and are number of new medicines for very rare diseases in the NICE appraisal pathway, for which in Wales there is currently no clinical or financial planning, and
- **Approve** a recommendation that Joint Commissioning Committee is the preferred option for the planning and commissioning of highly specialised services for Wales.

## 6. NEXT STEPS

If approved, the JCC Team will plan and commission these very rare diseases and highly specialised services as defined by NHSE and NHS Scotland and will:

- incorporate into the JCC portfolio and develop the directory of services
- utilise the Bluteq system for rebates
- horizon scan new medicines or technologies for very rare diseases
- develop a needs assessment of the number of patients with very rare diseases in Wales likely to eligible for new medicines or technologies
- financially plan for new medicines or technologies for very rare diseases in Wales, and
- commission the delivery of new medicines or technologies for very rare diseases and produce a policy position statement to confirm this.