

UK National Health Service, European Office – Official response.

EU Consultation on Rare Diseases

Introduction

The **National Health Service (NHS)** is the largest publicly funded health care system in the world providing the majority of healthcare in England. The NHS is committed to the principle of universal access to healthcare which is free at the point of use. Every 36 hours the NHS sees over one million patients who make use of a wide range of health services ranging from primary care, in-patient care, long term health care, ophthalmology and dentistry. The NHS is a major employer in Europe with 1.3 million people on its payroll.

This response has been coordinated by the NHS European office¹ in consultation with NHS organisations.

Background on the UK commissioning approach to rare diseases services

As there are 8,000 or more identifiable 'rare diseases' it is not practical to plan and purchase separately services for each of these. As a result the UK approach has been to provide specialised services around sensible groupings of expertise. For example, rather than purchase separate services for each of the many rare disorders of nerve and muscle, specialised centres delivering expertise in neuromuscular disorders have been established.

In the UK specialised services are not provided by every hospital, they tend to be found in larger hospitals based in big towns and cities. Specialised services are high-cost, low-volume interventions and treatments.

The underlying aim of arrangements to purchase specialised services is to ensure fair access to clinically effective, first class, standard, specialised services across England.

Specialised services are services where patient numbers are small and a critical mass of patients is needed in treatment centres to:

- achieve the best outcomes and maintain clinical competence,
- sustain the training of specialist staff,
- ensure cost-effectiveness in provision,
- make the best use of scarce resources (including staff expertise, high tech equipment, donor organs).

Effective planning and purchasing of specialised services ensures:

¹ The NHS European Office was launched in September 2007. It represents the English National Health Service. Its role is to inform the NHS of EU issues and to ensure that the NHS contribute positively to EU developments.

- the right patient (clear patient selection criteria and referral guidelines) is offered,
- the right treatment (evidence based, clinically and cost effective interventions) by,
- the right provider (monitored against agreed service/clinical quality standards) in,
- the right place (optimising geographical access but avoiding unnecessary duplication of provision) at,
- the right cost (robust costing and information systems and demonstrable value for money),
- with the full involvement of the patient (adequate information to enable supported choice).

Response to consultation questions

Q1 Is the current EU definition of a rare disease satisfactory?

Although the UK does not have an official definition for rare diseases, the prevalence criteria it uses to include a disease in national commissioning is 1 per 50,000. With this in mind the current EU definition of less than 5 per 10,000 which is mathematically equivalent to 25 per 50,000, does not accurately reflect the UK view, or the way health services for rare diseases are organised in the United Kingdom. Many diseases at a prevalence of less than 5 per 10,000 do not require special arrangements for health services to be commissioned.

The European Commission should therefore take into consideration that for the commissioning and provision of healthcare, a rare disease having a prevalence of less than 5 per 100,000 would be more appropriate.

Furthermore the current definition does not convey any criteria for the severity of the disease. Regulation EC 141/2000 for the designation of orphan medicinal products includes the requirement that the disease or condition is 'life-threatening or chronically debilitating'. The current definition should reflect the diverse levels of severity between rare diseases.

Q2 Do you agree that there is a pressing need to improve coding and classification in this area?

The NHS agrees that there is a pressing need to improve coding and classification in the area of rare diseases. Currently the UK uses the International Classification of Disease, 10th revision to code hospital and mortality data. This classification system would benefit from updating and improvement.

Q3 Can a European inventory of rare diseases help your national system to better deal with rare diseases?

A basic inventory of diseases would be a useful reference tool, particularly if it were regularly updated. This would enable the recognition of trends and alert

Member States and their healthcare systems of emerging health needs. However, given the very nature of rare diseases the NHS considers that an internal EU policy may not be sufficient and encourages the EU to collaborate with work also being undertaken at International level.

Q4 Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How?

European reference networks should predominantly privilege the transfer of knowledge. Some patients suffering from rare diseases are severely debilitated by their condition making travel extremely difficult. The Commission must consider that the majority of patients wish to be treated close to home, friends and family, and that travelling abroad to receive healthcare inevitably carries added complications: language barriers, transportability and translation of medical records, prior agreement for care pathway, liability issues.

Nonetheless the NHS recognises that situations may arise where the patient needs to travel to a centre of expertise for their treatment. This is most obvious for those in need of a surgical procedure or intervention, but applies also to patients who need complex medical assessments or technologies which are only available in a small number of locations in Europe. In such instances the work of the European Reference Networks in privileging both the transfer of knowledge and the mobility of patients will be extremely important.

Q5 Should on-line and electronic tools be implemented in this area?

While the NHS welcomes on-line and electronic tools in this area and in particular the empowerment they can bring to patients with rare diseases, we encourage the EU to build on on-line communities of patients with rare diseases and healthcare providers which are already in place. Through investing EU resources in this area, as opposed to creating new tools, the European Commission would not only help to sustain existing online networks but would facilitate the effective knowledge sharing they engage in. This would be a useful tool for the European Commission. As with any on-line and electronic tool however, the Commission must also consider the legal, practical and ethical issues which each of these tools brings.

While basic on-line information sources on rare diseases such as ORPHANET deliver clear benefits to patients and professionals, and are welcomed, more complex information sources such as databases should be properly governed, and may require legal compliance.

Q6 What can be done to further improve access to quality testing for rare disease?

Diagnosis of rare disease depends on both clinical assessment and laboratory investigation. Clinical assessment must be undertaken in expert centres which meet the criteria set out in the report from the Rare Disease Task Force to the High Level Working Group on Health and Health Services. Laboratory services must only be provided by laboratories which participate in external

quality assurance schemes. There are many of these good examples in Europe, the data for which should be accessible via an EU-wide coordinated information tool.

Q7 Do you see a major need in having an EU level assessment of potential population screening for rare disease?

The NHS encourages the Commission to avoid duplication at European level by researching common practices in population screening programmes.

The UK has a national screening programme which advises Ministers and the NHS about all aspects of screening policy and supports implementation. Using research evidence, pilot programmes and economic evaluation, it assesses the evidence for programmes against a set of internationally recognised criteria. Assessing programmes in this way is intended to ensure that they do more good than harm at a reasonable cost.

We would propose these arrangements as a model for other countries to follow and would support the assessment of potential screening for rare diseases to continue to be taken at national level,

Q8 Do you envisage the solution to the orphan drugs accessibility problem on a national scale or an EU scale?

The NHS recognises two aspects to the 'accessibility problem' as set out in the consultation document:

- Willingness of authorisation holders to market their products in specific member states
- Decisions by health systems on whether or not to pay for the product.

The first of these is not a substantive issue in the United Kingdom, but the second is of significant importance, particularly in a system such as the NHS which is funded by the taxpayer. The EU should not have the right to determine where publicly funded EU health systems spend their resources.

We believe that processes for assessment and appraisal of orphan products should be the responsibility of Member States, and must not be dealt with at European level.

Q9 Should the EU have an orphan regulation on medical devices and diagnostics?

We do not believe that there are sufficient problems in the development and commercial marketing of devices and diagnostics to justify the administrative effort and special privileges of orphan regulations.

Q10 What kind of specialised social and educational services for rare disease patients and their families should be recommended at EU level and at national level?

Although we welcome the opportunity to share information on specialised social and educational services for rare disease patients and their families at EU level, we do not believe the EU should make recommendations in this area.

Patients with rare diseases require specialised social and educational services which, by their nature, require provision which is close to home. In this respect decisions concerning their delivery should be made close to the point of delivery and not taken at EU level.

Q11 What model of governance and of funding scheme would be appropriate for registries, databases and biobanks?

Registries, databases and biobanks must fully respect common legal and ethical principles. Registries and databases for very rare conditions would be most conveniently organised on a pan-European basis and as a result should be eligible, subject to normal rules and processes for EU funding.

The development of treatments for orphan conditions frequently depend upon the use of biobank-type data as these processes make efficient use of the relatively scarce information base that is an inherent characteristic of orphan diseases..The intellectual property rights of the individuals who provide the raw material for such developments should be recognised in the governance arrangements of biobanks of orphan diseases.

Where registries contain evidence produced by a commercial enterprise, there should be no obstacle to full and efficient access to evidence contained within such registries. Similarly, access to and utilisation of the data contained in patient registries should not be influenced by specific patient groups.

We recommend that the design, construction, maintenance, analysis and control of registries for orphan conditions should be placed with an independent organisation.

Q12 How do you see the role of partners (industry and charities) in an EU action on rare disease? What model would be most appropriate?

Partnership working between patients and professionals form an important aspect of the treatment of rare diseases and as a result any proposed EU action should reflect this.

Such partnership working should include not only relationships between charities, patients and health professionals, but should also consider the role of industry. Where potential tensions may exist, dialogue must be open and constructive. Transparency would be vital to the success of any partnership working.

Q13 Do you agree with the idea of having action plans? If yes should it be at national or regional level in your country?

The NHS supports Member States developing their own national action in this area, with opportunity for diversity at regional level. Within England arrangements to plan and purchase specialised services will be taken forward by the 'Specialised Commissioning Groups'. The devolved administrations of Scotland, Wales and Northern Ireland will develop their own plans.

Q14 Do you consider it necessary to establish a new European Agency on Rare Disease and to launch a feasibility study in 2009?

The NHS supports the launch of a feasibility study to investigate what measures and additional research may be necessary at EU level to tackle rare diseases effectively. Only after a study has been completed should the EU consider what the best course of action may be.

The study should investigate various courses of action to address rare diseases at EU level, and set out the scope, structures and costs for each one. It should not be assumed that a new European Agency on Rare Diseases is the only possible way forward. In this respect the NHS recommends the European Commission also to investigate what role the European Centre of Disease Prevention and Control may have to play, and to investigate expanding their competencies to deal with rare diseases.

Any final decision should only be made following formal approval by Member States.

This does not detract from the NHS view that some form of additional resources in this area could be helpful in ensuring coherent EU strategies encouraging research, development and marketing of medicines to treat rare diseases and supporting the availability of new orphan drugs to patients in the different Member States.