



RARE DISEASE UK: THE CASE FOR A NATIONAL STRATEGY FOR RARE DISEASE

What is the issue?

There are over 6000 rare diseases affecting over 300,000 people in Scotland and over 3.5 million people in the UK (1 in 17). Collectively, rare diseases are not rare.

Services to support people with rare diseases remain patchy and poorly integrated, meaning that hundreds and thousands of families with rare diseases struggle to access the help and support that they need.

What is Rare Disease UK asking for?

- We called on the UK Government to support the recent Council Recommendation on a European action in the field of Rare Diseases. The Recommendation, which was adopted on 9th June 2009, calls on EU Member States to implement plans or strategies for rare disease, to aim to ensure that patients with rare diseases have access to high quality care. **Achieved.**
- We would like to work in partnership with the Scottish Government and colleagues in NHS Scotland to develop and implement a National Strategy for integrated service delivery for families affected by rare disease in Scotland and across the UK.

How would this benefit Scotland and the UK?

A National Strategy for integrated service delivery for families affected by rare disease would aim to create efficiency in expenditure and service allocation. It would:

- help to make the most efficient use of the limited resources available in the health services;
- support the health research agenda, including strengthening translational research;
- facilitate early and correct diagnosis and timely treatment, which could offer improved health and quality of life for people with rare diseases;
- ensure the nations of the UK keep pace with other EU countries that have adopted national plans on rare disease.

What is Rare Disease UK?

It is the National Alliance of key stakeholders brought together in response to unmet health care needs of hundreds and thousands of families who currently struggle to get access to integrated care and support from the NHS. It is a joint initiative of the Genetic Interest Group (the UK alliance of patient organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders) and others.

Why do we need a National Strategy for integrated service delivery for patients with rare disease?

Much progress has been made in this field in the past decade, with innovative commissioning strategies and the establishment of centres of excellence delivering multidisciplinary care for some conditions. However, there is currently no coherent strategy for care and support for people affected by rare disease.

Many patients with rare conditions have difficulties accessing high quality care and services, in many cases due to the lack of communication and coordination of services that are sometimes already in existence. Due to the rarity of many conditions, health professionals often have little or no experience in supporting patients in order to find optimum care pathways which can often be difficult or impossible. This leaves patients and families feeling frustrated and 'stuck' in the system.

A National Strategy for integrated service delivery is needed to improve the current fragmentation of services and enable patients and health professionals to provide and use best practice care. This will ensure that all patients with rare disease can not only be diagnosed quickly, but also have timely access to the care and support that they need.



IMPROVING HEALTHCARE SERVICES

How could a National Strategy for integrated service delivery improve healthcare services for people with rare disease?

Care and services for families and patients with rare disease are currently patchy and fragmented. A National Strategy for integrated service delivery could ensure that the hundreds and thousands of individuals and families who are affected by rare genetic disorders have ready access to integrated care and support from the health services.

How would this benefit Scotland and the UK?

A National Strategy for integrated service delivery would help NHS Scotland make the most efficient use of scarce expertise by targeting health care resources to maximise the benefits for patients and families affected by rare disease.

Empowering patients and families by improving information on rare diseases would also contribute to the vision of a patient-led NHS.

A National Strategy for integrated service delivery should include commitments to:

- identify national or regional centres of expertise, and create them where they do not exist;
- ensure clear healthcare pathways for patients with rare disease through the cooperation between relevant experts;
- provide coordinated care for patients with rare disease within a multidisciplinary team approach model of care, to counter fragmentation across organisational boundaries (this is important because many rare disease affect a number of systems within the body, so patients need to see healthcare professionals from several different specialties during their lifespan);
- improve information on rare diseases for patients and the public.

How much would this cost?

Recommendations such as those outlined above are largely about reorganisation of resources, rather than an increase in resources. They would enable a more efficient use of resources and maximised benefits at minimum cost to government.

PROMOTING RESEARCH INTO RARE DISEASE

How could a National Strategy for integrated service delivery promote research into rare disease?

This is a timely opportunity to ensure:

- that Scotland and the UK are attractive locations for rare disease research. That means providing the right framework and environment to support this research – this could include, for example, patient registries to facilitate clinical trials.
- better coordination of the rare disease research that is already taking place, for example by building research networks.

How could this benefit Scotland and the UK?

A clear strategy on rare disease research, embracing both the academic and private sectors, would help to reinforce our position as world leaders in biomedical research. Coordinating and fostering rare disease research would:

- mean more possibilities for diagnosis and treatment;
- make Scotland and the UK more attractive research locations;
- lead to knock-on benefits for medical research in general, particularly for conditions with a genetic component, such as asthma and diabetes.



A National Strategy for integrated service delivery would open up opportunities for greater international research cooperation, which is particularly important for rare disease with very low number of patients. This is an opportunity that we need to grasp in order to keep pace with other countries and keep investment here, and it is closely aligned with the health research agenda, for example in:

- supporting translational research;
- developing a thriving research culture;
- maintaining and attracting investment by the pharmaceutical and biotechnology industries.

A National Strategy for integrated service delivery should include commitments to:

- identify current research projects and existing research resources;
- identify needs and priorities for basic, clinical and translational research in the field of rare disease;
- foster research in the field of rare disease.

How much would this cost?

The aim of the National Strategy for integrated service delivery would be to create efficiency in current expenditure.

ENABLING HEALTH GAIN

How would a National Strategy for integrated service delivery lead to increased health gain?

By enabling early diagnosis and timely access to appropriate treatment. This is particularly important for the many rare diseases that are progressive in nature.

Alstrom Syndrome is one of the rare conditions for which there is coordinated care, with specialised multi-disciplinary clinics which make a real difference to families' lives. However, many other patients with rare diseases do not have access to coordinated specialist care. This can result in misdiagnosis or non-diagnosis, which are barriers to improving quality of life. For example, it is very common for families with Gorlin Syndrome (of which one of the main characteristics are skin cancers called basal cell carcinomas, or BCCs), to remain undiagnosed for several generations. Some people with Gorlin Syndrome are still misdiagnosed and given radiotherapy, which accelerates the BCCs.

Where there are treatments for rare conditions, not all patients are able to access the right treatment, either at all or at the right time. Medicines for such small numbers of patients are unlikely to meet cost-effectiveness criteria, but even if they do, decisions to fund treatments are then often made on a case by case basis. As well as causing undue worry for patients, wasting health service resources and leading to geographical variations, the subsequent delays in treating patients can reduce the health benefits of the treatment.

How would this benefit Scotland and the UK?

The more effective and efficient the diagnosis and treatment of rare disease, the more people with rare conditions can live healthier, longer lives and make a contribution to society and the economy.

A National Strategy for integrated service delivery should include commitments to:

- improve diagnostic facilities and genetic testing services;
- foster greater awareness of rare disease among the medical profession, to ensure early diagnosis;
- ensure that there is a systematic way of introducing new interventions for health gain;
- establish a robust, transparent, evidence-based system to assess new medicines for rare diseases, involving all stakeholders;
- commission treatment of rare diseases at regional or national level;
- ensure adequate access to cost-effective treatments.



How much would this cost?

There is a misconception that the healthcare budget would be overwhelmed with a 'tidal wave' of new medicines for rare diseases. There are several reasons why that would not be the case:

- Each year around 100 medicines in development receive orphan designation (official recognition that they would be used to treat rare disease). However, only 10 per cent of these eventually become licensed medicines.
- At any one time it is unlikely that there would be more than 150 orphan medicinal products on the market in Europe under exclusivity arrangements (meaning that directly competitive similar products cannot normally be placed on the market at the same time).

A EUROPEAN COMPARISON

The nations of the UK are in danger of slipping behind other countries who are taking action. France has implemented a comprehensive plan, the Netherlands has implemented a national strategy and Italy has adopted regional strategies. Bulgaria and Portugal have also adopted national plans. This now needs to be considered as a positive opportunity for our patients.

NEXT STEPS

Rare Disease UK aims to create a draft National Strategy for integrated service delivery for families affected by rare disease, with input from expert stakeholders from research, patient organisations, clinicians, the pharmaceutical industry, commissioners and government. The Strategy will bring together research into all aspects of rare disease, prevention and diagnosis of rare disease, best practice in the treatment of rare disease, and the dissemination of information on rare disease to the public, patients and professionals.

We look forward to working with the governments in the UK to take this forward and make a real difference to the hundreds and thousands of families living with rare disease in this country.

PATIENTS' STORIES

"It has taken over a year to get a diagnosis for our 1 year old daughter - meanwhile untold damage was being done as a result of her infantile spasms". **Family with child with Tuberous sclerosis, Ayrshire**

"As a parent of two children with Laurence Moon Bardet Biedl Syndrome, a rare and complex genetic condition, it is very stressful to be made aware time and time again that consultants have little experience or knowledge about this condition." **Family with LMBBS, Dunbartonshire**

"I was misdiagnosed as having asthma rather than a degenerative lung condition of genetic origin and thus I was not treated correctly." **Patient with Alpha 1 anti-trypsin deficiency, Dundee**

"Our son was tested at 3, but we didn't find out what his diagnosis was until he was 23, where we came across it by accident when we asked to read his file. In our experience, after diagnosis, it was like the door was shut." **Family with an adult son with multiple learning disabilities and epilepsy, caused by a rare chromosomal disorder**

I was diagnosed with Neurofibromatosis when I was about 13 years old (I'm now in my 40s). I was never offered any support or thorough checks. I also feel that if you have a rare disease you don't always see the correct doctors. For many years I was only ever seen by a Dermatologist despite the fact that Neurofibromatosis is a complex condition that affects the nervous system, is often associated with learning disabilities and can affect other parts of my body as well. **Patient with Neurofibromatosis, Glasgow**

It took me 12 years of GP visits with back pain before I was finally sent to see a Rheumatologist who diagnosed my Alkaptonuria. I am used to being met with the question from healthcare professionals, "Alkaptonuria, what is that? I've never heard of it." I feel that I know more about my genetic disorder than they do. **Patient with Alkaptonuria, Orkney**