

Improving Lives, Optimising Resources: How a strategy for rare diseases will benefit the people of Scotland



RARE DISEASE | UK

The National Alliance for people with rare diseases & all who support them

In Scotland, 300,000 people are likely to be affected by a rare disease at some point in their lives (1 in 17 people). Collectively, rare diseases are not rare and they represent a significant health burden to the health services in Scotland.

Rare Disease UK's (RDUK) report *Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy* describes how currently there are a number of obstacles preventing patients and families being able to access the high quality services, treatment, care and support that would ensure they benefit from optimum health outcomes. The report sets out recommendations developed in collaboration with a broad range of stakeholders in Scotland and across the UK to address these issues. These recommendations should form the basis of an effective strategy for rare diseases.

A strategy for rare diseases would ensure that patients receive the highest quality healthcare whilst ensuring the most efficient and effective use of NHS resources.

*The Healthcare Quality Strategy for NHS Scotland*¹ made a number of commitments to patients in Scotland. RDUK supports those commitments and the ultimate aim to deliver the highest quality of healthcare to the people of Scotland. RDUK looks forward to working in partnership with the Scottish Government and key stakeholders to develop an effective strategy to ensure that the aims laid out in the *Healthcare Quality Strategy* are realised for people with rare diseases.

How RDUK's recommendations for a strategy for rare diseases would enable the Scottish Government to meet its aims:

Aim: 'individual health encounters that are consistently person-centred...for every person, all of the time'

RDUK's recommendations encourage this by, for example, the production of individual care plans, the designation of Care Coordinators and the development of information prescriptions for patients.

Aim: the 'most appropriate treatments, interventions and services will be provided at the right time to everyone who will benefit, and wasteful or harmful variation will be eradicated'

RDUK's recommendations facilitate this through methods to ensure equitable, timely access to effective diagnostic tests, drugs and therapies for all patients that need them, regardless of their location or the rarity of their condition. Recommendations to aid timely diagnosis would avoid unnecessary or harmful interventions and ensure early access to effective interventions and better management of the condition.

Aim: to 'build on NHS Scotland's integrated delivery arrangements'

Adopting RDUK's recommendations would encourage effective collaboration and communication between everyone involved in a patient's care; it would improve continuity of care at transition periods; it would help to ensure clear communication between patients and professionals, including information on their condition, management and treatments.

Aim: research to 'improve patient benefit and population health'

*Investing in Research: Improving Health*² outlined the Scottish Government's ambitions to build upon the exceptional research base in Scotland. There are a number of barriers that hinder research into rare diseases. RDUK suggest ways these can be overcome to ensure that Scotland can foster research into rare diseases, to the benefit of patients affected by both rare and common diseases and to Scotland's economy.

In June 2009, the UK Government adopted the Council of the European Union's Recommendation on an action in the field of rare diseases. This calls on member states to develop plans or strategies for rare diseases by 2013. The Scottish Government has committed to working collaboratively with the other home nations to develop a strategy for rare diseases. RDUK calls on the Scottish Government to take on board the recommendations outlined in *Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy* and implement an effective strategy for rare diseases.

1. 'The Healthcare Quality Strategy for NHS Scotland', The Scottish Government, May 2010

2. 'Investing in Research: Improving Health', The Scottish Government, 2009

Some excellent examples of good practice already exist in Scotland. It is vital that these are developed or used as models on which to base new services. Where expertise already exists, it should be shared and utilised to benefit others.

Rare disease Managed Clinical Networks: examples of good practice in Scotland

1. The **Scottish Muscle Network (SMN)** raises awareness of neuromuscular diseases (NMDs) and promotes the delivery of equitable, high quality care to all NMD patients in Scotland regardless of where they live. The network carries out a range of activities including educating professionals, increasing communication between everyone involved in a patient's care, and the development of information for patients and professionals.

Since it was established in 1998, the SMN has produced guidelines, leaflets and patient care cards and in its first three years alone it delivered over 9000 hours of CPD education for professionals. It has been used as a model by the Muscular Dystrophy Campaign for development of more networks in the UK.

A recent review of the SMN carried out by the National Services Division¹ showed that benefits of the network included increased life expectancy and quality of life for Duchenne muscular dystrophy patients, improved care for patients with other NMDs and delivery of significant benefits to the NHS in Scotland.

2. The **Uveitis National Managed Clinical Network** was established by an informal collaboration of clinicians and patient organisations with an interest in uveitis to best achieve high quality, equitable care for uveitis patients throughout Scotland. It has since developed a number of guidelines, including treatment guidelines aimed to help ophthalmologists in the prescription of immunosuppressive therapy for ocular inflammatory disease and to help them identify when to refer to a specialist. It has also produced care guidelines which outline the care and services that all uveitis patients in Scotland should receive; and management and screening guidelines for juvenile idiopathic arthritis associated uveitis.

¹. 'Review of nine National Managed Clinical Networks', National Services Division, February 2010

A collaborative approach to genetic testing: an example of good practice in Scotland

The **Scottish Molecular Genetics Consortium** is a collaboration of four genetic testing laboratories based in Aberdeen, Dundee, Edinburgh and Glasgow. They work together to provide tests for a range of inherited conditions for people in Scotland. Each laboratory has its own remit of tests. The patient's most local consortium laboratory provides them with information and takes the sample and dispatches it to the relevant laboratory for testing. This collaborative approach to testing ensures that laboratories only hold enzymes and probes relevant to their assigned tests which results in financial savings and development of expertise. It also means that the sample travels, not the patient, and it allows an integrated and flexible response to service need. It results in increased collaboration between laboratories which is advantageous in terms of education between professionals.

Caring for patients with complex needs: an example of good practice in Scotland

The Scottish Government Health Directorate recently (2008) invested in the redesign of the **Single Gene Complex Need (SGNC)** service which works with families throughout Scotland living in their local communities with rare diseases and complex needs as a consequence of genetic change.

The service has two distinct but related elements:

- To develop a strategic approach to service delivery
- To provide direct support to families and professionals in their local communities

The service consistently puts people at the centre of each care encounter. It works with people in an anticipatory way to improve their outcomes and increase their confidence to manage their long term condition(s). It effectively collaborates with patients and their support networks (non-paid carers and the multi-disciplinary teams) towards mutually providing more effective service delivery.

The project is in its infancy but initial findings support greatly improved outcomes for some living with complex needs as a consequence of genetic change but also highlights many of the challenges families and professionals face working towards improving outcomes for people with rare diseases.

Produced by Rare Disease UK

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