Health Inequalities – Early Years

Rare Disease UK

Introduction:
Rare Disease UK is the national alliance for people with rare diseases and all who support them. It was established by Genetic Alliance UK, the national charity of over 160 patient organisations supporting all those affected by genetic conditions, in conjunction with other key stakeholders in November 2008 following the European Commission’s Communication on Rare Diseases: Europe’s Challenges. Rare Disease UK was developed in response to the unmet healthcare needs of the millions of people living with a rare disease and who currently struggle to get access to integrated care and support from the NHS.

A rare disease is defined by the European Union as one that effects less than 5 in 10,000 of the general population and will affect approximately 1 in every 17 people at some point in their lives. There are between 6,000 and 8,000 known rare diseases and these can include rare cancers such as childhood cancers, and some other well known conditions such as cystic fibrosis and Huntington’s disease. Often rare diseases are chronic and life-threatening and can be single-gene, multifactorial, chromosomal or non-genetic.

Rare diseases will affect approximately 300,000 people in Scotland. 75% of rare diseases will affect children and 30% of rare disease patients will die before reaching their fifth birthday.

Diagnosis of a relatively common, chronic condition can be devastating, but coming to terms with a rare disease can be made much harder by difficulties in getting an accurate diagnosis, accessing support and information and securing appropriate treatment. Rare Disease UK has campaigned for the development and implementation of an effective strategy for rare diseases in the UK. Rare Disease UK has also campaigned for rare disease patients to have equal access to diagnosis, services, information and support as those affected by more common conditions and conducted research to identify the gaps that currently exist in healthcare provision for rare disease patients.

This document will highlight the experiences of people in Scotland living with a rare condition, the importance of early diagnosis and early intervention for rare disease patients and draw the Committee’s attention to the current work being undertaken by the Scottish Government to produce a plan for rare diseases in Scotland.

Experiences of Rare Diseases in Scotland and the importance of early diagnosis and intervention:

Between October and November 2012, Rare Disease UK carried out a survey of patients and families living in Scotland affected by rare diseases. The aim of this survey was to find out more about the experiences of people in
Scotland living with a rare condition, and to identify some of the common issues and problems they frequently face. In February 2013, Rare Disease UK published the findings in the report, “Experiences of Rare Diseases: Patients and Families in Scotland”1.

The report indicated that, whilst many families received high quality services and support, this was not universal and too many families affected by rare diseases in Scotland struggle to receive a speedy diagnosis and access high quality information. A summary of the key findings are provided below.

Diagnosis:

Early diagnosis and intervention of a rare disease can improve a patient’s life expectancy and quality of life and can also provide important information to guide further reproductive choices for the family. However, there is a lack of awareness and identification of rare diseases amongst health professionals which often results in a delay in diagnosis or misdiagnosis of rare disease patients. The consequence is a delay in accessing appropriate treatment, therapy or effective management of the condition, as well as an inefficient use of NHS resources due to multiple avoidable appointments, incorrect diagnostic tests and treatments.

Rare Disease UK’s report highlights that 44% of patients with a rare disease in Scotland had to wait over one year for an accurate diagnosis, with 10% waiting over five years and a staggering 4% waiting in excess of twenty years for a diagnosis.

Furthermore, an unacceptably high proportion of patients and families affected by rare diseases receive at least one incorrect diagnosis before their final diagnosis is confirmed. 36% of respondents to RDUK’s survey received more than 3 incorrect diagnoses with 13% receiving in excess of five incorrect diagnoses before their final diagnosis was confirmed. Not only is misdiagnosis distressing for the patient and their family, it can lead to a deterioration of the condition as effective treatment, therapy and management of the condition is delayed.

Information:

When a diagnosis is given, too often patients do not receive appropriate information and are left to research their condition on their own. This can result in patients finding unsuitable, often alarming information and not being able to discuss this with anyone who understands the condition. A patient’s condition may be being managed effectively by the NHS according to best current knowledge of that condition, yet the lack of appropriate people and/or tools to communicate that information and translate into care, together with a patient’s feeling of insecurity and isolation, can lead to a perception of inadequate care. Information is a crucial element of the service a patient receives and should not be viewed in isolation or as something which is

optional. Empowering patients through information is a relatively low – cost way of ensuring better management of a condition as well as increasing a patient’s satisfaction with the service that they are receiving\(^2\). As information on rare diseases and their management is often scarce and difficult to find in comparison to information on common diseases, it is of even greater importance that patients with rare diseases are supported to obtain the information that they need.

Rare Disease UK’s report highlights that 41% of patients and families do not have someone who they can approach to answer questions about their condition, care and/or treatment. Our survey showed, many patients and families are forced to search online for information on their conditions. At the time of diagnosis, almost half (45%) of respondents did not receive sufficient information on the condition, care or treatment and a staggering 22% were forced to search for this information by themselves.

Access to Services:

Rare Disease UK’s survey shows that patients and families in Scotland experience difficulties accessing non-medical services.

- Only 13% of respondents found it easy to access respite care.
- 30% of respondents reported being able to access psychological support easily.
- Only 26% of patients and families were able to obtain equipment (for example, wheelchairs) with ease
- Just 24% were able to obtain home adaptations easily.
- 22% of respondents were able to access support for special education needs.

The survey also highlights that many patients and families are not receiving sufficient information on the welfare benefits available and are experiencing difficulties when trying to obtain financial support.

30% of respondents to the survey received no information on welfare benefits, a further 36% of patients and families felt that more information was needed. 42% of patients and families reported experiencing difficulties in obtaining financial support and/or benefits.

Benefits may be available to financially assist patients with rare diseases and their families, but too frequently patients are not aware of or informed about these, and are not given the help they may require to apply for them. Other patients have reported having to battle to access benefits to which they are entitled. This may stem for the relevant authorities lacking awareness of rare diseases and the challenges patients face with these conditions not fitting into boxes easily.

Access to appropriate therapies and treatment can be difficult for rare disease patients in Scotland. The findings of our survey show that many patients are experiencing difficulties accessing drugs for rare diseases in Scotland. 26% of respondents highlighted that they had experienced difficulties, or had been unable, to access medications for their diseases. For many rare diseases, there are no effective treatments available. For patients who do have an effective treatment available to them, their lives, and those of their family, can be transformed. It is for this reason that experiencing difficulties when accessing drugs for rare diseases can be distressing for the patient and their family. RDUK commends the Health and Sport Committee for their work on Access to Medicines, which has led to recommendations for improved processes for approving new medicines for rare diseases and the IPTR process.

Coordination of Care:

Most rare diseases affect multiple parts of the body and many different professionals are often involved in the care and treatment, as such there must be good coordination and communication between them. However, for many rare disease patients care is often poorly coordinated and fragmented and care is not always person-centred and does not always fully take into account a patients individual needs and preferences.

Rare Disease UK’s report found that many rare disease patients have multiple health professionals involved in their care and treatment and this is often administered in multiple locations. Almost two thirds of respondents have at least three health professionals across a range of disciplines, involved in their routine care and treatment and almost half (47%) of all respondents reported that their care or treatment was provided in more than three locations. Attending multiple clinics can result in large disruption to a patient’s and carer’s daily life, and can make regular attendance at school or work very difficult and can have a large financial impact on the patient or the family due to the cost of travel.

It is encouraging that most patients access their care or treatment within Scotland, with just 11% having to travel outside of Scotland.

Very few patients in Scotland have access to a Care Coordinator, however, most patients and families believe that having a trained professional whose role is to ensure that a care plan is in place and acted upon, would be beneficial. Rare Disease UK’s research shows that just 24% of respondents had a dedicated care coordinator / care advisor and that 48% of patients and families who currently do not, believed that having one would be beneficial.

Research & Registries:

Research into rare diseases is vital to enable the development of new therapies, diagnostic tests and preventative measures for patients affected by these conditions. However, only 16% of patients and families in Scotland felt that sufficient research was being conducted into the rare disease that they
are affected by. In addition, only 8% of respondents believe that there is enough opportunity to participate in research.

Encouragingly, 61% of respondents have participated in research, with participation in a clinical trial being the most common method of involvement. If made aware of ways in which to aid research, patients are generally willing to participate. Research represents hope for many patients and families and as such, patients and families need to be kept up to date on research developments into their condition and need to be made aware of research which may be relevant for them to participate in.

Disease registries can provide essential information for clinical care, planning and service delivery, and are a valuable tool for the initial collection of data on rare disease patients. The research shows that the majority of patients support the use of registries and indicate that most would be willing to join one if it existed and they were made aware of it. The research shows that only 24% of respondents were aware of a registry that exists for their rare disease.

A Strategy for Rare Diseases:

Since 2009, Rare Disease UK has campaigned for the development and implementation of an effective strategy for rare diseases in the UK that is comprehensive and fully reflects the needs of the rare disease community.

In November 2013, the UK Strategy for Rare Diseases was published containing 51 commitments with the aim of ensuring no one gets left behind just because they have a rare disease. Key elements of the strategy include:

- Personal care plans for patients, bringing together health and care services, with more support for patients and their families;
- Support for specialist clinical centres offering better care and support;
- Better education and training for health professionals to help ensure earlier diagnosis and access to treatment;
- Promotion of the UK as a world leader in research and development in this field.

The four countries of the United Kingdom have committed to producing their own implementation plans and the Scottish Government is currently developing the plan for Scotland. The Scottish Plan is intended to “reflect the

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excellent work already carried out across the NHS, universities, enterprise, industry, social care and the third sector. It will acknowledge the importance of timely and accurate diagnosis, allowing appropriate treatment to start and giving people with rare conditions and families access to support services.\(^6\)

**Conclusion:**

There are between 6,000 and 8,000 rare diseases affecting 3.5 million people in the UK – collectively, rare diseases are not rare. Rare disease patients should expect to have equal access to diagnosis, services, information and support as those affected by more common conditions. Rare Disease UK urges the Health and Sport Committee to consider the difficulties faced by rare disease patients, particularly in relation to early diagnosis, during their inquiry into early years. 75% of rare diseases affect children with 30% of rare disease patients dying before their fifth birthday – delays in diagnosis can result in missed opportunities for intervention, allowing conditions to become progressively worse and more difficult, sometimes impossible, to treat.

With the publication of the UK Strategy for Rare Diseases, patients with rare diseases in Scotland can have a clear expectation of what to expect from the NHS in Scotland. Rare Disease UK asks the Health and Sport Committee to support and encourage the development of a rare disease plan for Scotland and to monitor its implantation.

**Rare Disease UK**
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**Useful Links:**

**Strategy for Rare Diseases** –

**Rare Disease UK** - http://www.raredisease.org.uk/index.htm

**Experiences of Rare Diseases: Patient and Families in Scotland** –


\(^6\) http://www.scotland.gov.uk/Topics/Health/Services/RareDiseases