All health professionals need to be aware of rare disease and ensure patients receive prompt diagnosis, information and ongoing support

RARE DISEASES: PART 1 OF 6

Rare diseases: what do you need to know?

In this article...
▷ A definition of rare disease and the extent of the problem
▷ Why health professionals need to know about rare diseases
▷ How to improve the care of patients with rare diseases

Keywords: Rare diseases/ Diagnosis/ Individualised care
● This article has been double-blind peer reviewed

5 key points
1. Rare diseases often present at birth or occur during infancy and childhood, and over 75% affect children
2. Early diagnosis improves the quality of life for both patients and families
3. One in five people wait longer than five years for a correct diagnosis
4. One-stop clinics providing multidisciplinary specialist care have transformed the management of some rare diseases
5. Health professionals need to know how to find information about rare diseases so they can support patients

Over 75% of rare diseases affect children

A ll nurses, midwives and health visitors are likely to encounter patients with highly complex, rare diseases during their careers. These conditions are multisystem, debilitating, often life threatening and frequently inherited. The challenge for health professionals is to be aware of, and recognise them. Fully coordinated specialised care can only begin when someone puts the pieces together to make a diagnosis. This involves looking beyond a single system – such as cardiac, liver or kidney disease – and connecting the symptoms that may indicate an inherited condition.

The Department of Health (2012) consultation on rare disease suggests these conditions affect fewer than five in 10,000 people. Some are rarer still, affecting only a handful of people in the world. The Council of the European Union estimates that 5,000-8,000 distinct rare diseases exist, affecting 6-8% of the population at some point during their lives; this equates to one in 17 people worldwide (CEU, 2009).

A survey showed only 27% of patients with rare diseases are cared for in specialist centres (Rare Disease UK, 2013a). The remainder are likely to be seen in general hospitals or the community. As these patients tend to need frequent tests and ongoing medical care, it is important that everyone working in these settings is at least aware of rare diseases (DH, 2012).

Presentation
Frequently, rare diseases present at birth or in infancy/childhood, and over 75% affect children (RDUK, 2013a). These include:
» Rare cancers, such as neuroblastoma affecting nerve tissue alongside the spine, chest, abdomen and pelvis;
» Bone diseases, such as osteogenesis imperfecta (brittle bones); and
» Life-threatening birth defects, such as CHARGE syndrome, which is linked to heart and breathing problems.

This means that midwives, neonatal staff, paediatric nurses, district nurses and health visitors are likely to be involved in the care and support of these patients and their families. However, as more children with rare diseases survive into adulthood, greater recognition and treatment of adults is now needed.

Some rare diseases mainly manifest in adult life with progressive symptoms of chronic debilitating disease, often resulting in admissions to general wards. Huntington’s disease with mood and cognition problems in addition to physical symptoms, or acromegaly with arthritis,
Fabry disease (FD) is a multisystem rare disease, affecting 1:40-60,000 men. It affects women less frequently. Ideally, patients are cared for by a cardiologist, renal physician, dermatologist and a specialist in inherited diseases.

FD is an inherited X-linked lysosomal storage disease. The lysosome uses enzymes to act as a cell recycling centre and if one of the enzymes, Alpha Galactosidase A, is missing, waste products build up in the body. This results in accumulation of glycosphingolipids, leading to symptoms affecting the heart, kidneys, eyes, nervous system and bowel. Patients often develop a rash around the torso and are at high risk of stroke.

Transmission of X-linked recessive gene is shown in Fig 1. It often takes years to be diagnosed and for treatment to start (Mehta and Ramaswami, 2012).

The patient
Mr Elton’s symptoms started when he was five years old and had unexplained neuropathic pain. When he was 20, he developed a rash and a skin biopsy did not confirm a diagnosis. This was later identified as a Fabry rash called angiokeratoma. Between 30 and 35 years of age he was diagnosed with renal dysfunction and at 38 he developed left ventricular hypertrophy. In view of these symptoms, his cardiologist referred him for FD testing. Once diagnosed, other symptoms of FD were discovered, including: reduced hearing; white-matter brain changes; and signs of irritable bowel syndrome.

Mr Elton’s mother was found to be clinically affected with FD. Usually X-linked conditions do not affect women as they only carry the gene causing the disease but, in FD, women may develop symptoms. Mr Elton’s mother had three stents inserted for coronary heart disease and has long-standing kidney problems.

Mr Elton also has a young daughter and a son. With X-linked conditions, men cannot pass the condition onto sons as the affected GLA gene is not carried on the Y chromosome. His daughter, however, is a carrier for FD and is having annual paediatric surveillance screening. The family tree is shown in Fig 2.

FD has a wide clinical spectrum and some patients will need to have a kidney transplant. In Mr Elton’s family, his mother has coped well with symptoms for many years and now is relieved to have a diagnosis. However, Mr Elton and his wife are struggling to come to terms with the life-long illness, shortened life span and uncertain future for their daughter.

*Names have been changed.

Some rare diseases are variants of conditions, including breast cancer, diabetes, osteoporosis, and renal and heart disease. Some are more common in certain parts of the world; for example cystic fibrosis is rare in Asia but more common in Europe. Many are progressive and incurable.

Approximately 80% of all rare diseases have a contributing genetic factor, including single gene, multifactorial or chromosomal defects (RDUK, 2013b). This makes it important to involve clinical geneticists or genetic counsellors and genetic testing in care packages. Compiling a family history is vital as relatives of patients with a known genetic mutation...
Nursing Practice Discussion

may need to undergo predictive testing (cascade testing), outpatient preventive clinical screening and treatment. The emotional and psychological impact of inherited diseases must be considered when caring for these patients and their relatives.

Early diagnosis improves the quality of life for both patients and their family and may allow therapeutic interventions to be started sooner (DH, 2012). However, many of these diseases are difficult to diagnose as they share symptoms with other common disorders (Wraith and Beck, 2012). Many patients do not receive a diagnosis or are misdiagnosed for much of their life. An RDUK (2010) report highlights that:

- 30% of patients received more than three incorrect diagnoses;
- One in five waited longer than five years for a correct diagnosis;
- 52% felt they did not receive enough information on their condition after being diagnosed.

The case study in Box 1 describes the patient and family journey from presentation of symptoms to diagnosis; it illustrates the complexity of these conditions.

Supporting patients

Burke et al (2007) found patients with rare diseases wanted health professionals to:

- Have greater awareness of their condition;
- Have greater willingness to consider the possibility of a genetic condition;
- Refer patients appropriately; and
- Take their concerns seriously.

An RDUK (2011) report identified the obstacles patients and families come up against when seeking individualised care, and makes a series of recommendations. It highlights that patients with rare diseases can present in any clinic, ward or GP surgery and deserve high-quality care from all health professionals. It is vital each centre knows its network of local hospitals, which should have systems in place to put patients on a pathway to the expert centre that will offer help, support, advice and assistance.

Patients with rare diseases have identified fragmented care and a lack of communication as major concerns; most turn to support groups for information (RDUK, 2011). Health professionals must provide clear, individualised information, recognising the emotional impact it may have, and be prepared to engage and communicate with other health professionals involved in complex care pathways.

It is also important not to see patients solely in terms of their rare disease. Caring successfully for them demands the ability to think laterally, especially in accident and emergency where they may present acutely with an unrelated condition (for example Addison’s disease with sudden hypotension and collapse due to lack of steroids). Health professionals must consider how the presenting disorder could affect the existing rare disease, or how the rare disease may have altered presentation of a common condition. Either scenario may adversely affect the patient journey (DH, 2012). Articles in this series will provide information about specific conditions.

“One-stop clinics” providing multidisciplinary care in a centre of excellence can optimise the care pathway of patients with rare diseases, but communication between them and local centres can be patchy. Time off work, travel costs and distance to a specialist clinic can also be burdensome so collaboration with local care providers is vital. It is also crucial that local health professionals know how to enlist specialist centre advice and support. Patient support and information groups play an important part in this dialogue (RDUK, 2013). Useful sources of information are listed in Box 2.

Conclusion

More than 5,000 rare diseases have been identified. They can affect anyone at any age, physical health, mental health or both. Patients and their families need care from a centre of excellence and wide-ranging support from other medical, nursing, diagnostics and social care professionals.

The UK Strategy for Rare Diseases is aiming to raise awareness of, and tackle the challenges posed by, rare diseases. Similar plans are in progress throughout Europe. Improved clinical pathways, transitional care and communication, collaboration with patient support groups, extended nursing and medical training, creation of national disease registers and heightened research awareness will all benefit patients with rare diseases.

We are increasing general awareness of rare diseases by hosting the first national Conference for Rare Disease in the UK on 28 February 2014 (open to all health professionals). Visit tinyurl.com/RD-studyday for more details. This is also World Rare Disease Day (Join Together for Better Care) and will be celebrated internationally.

References


Rare Disease UK (2013a) Centres of Excellence for Rare Diseases. tinyurl.com/RDUK-excellence

Rare Disease UK (2013b) About Rare Diseases. tinyurl.com/RDUK-facts

Rare Disease UK (2011) Improving Lives, Optimising Resources: A vision for UK Rare Disease Strategy. tinyurl.com/RDUK-strategy


Box 2. USEFUL SOURCES OF INFORMATION

- Specialist centres (mainly located in teaching hospitals) should have a telephone service for families and health professional enquiries. Contact details for our clinical nurse specialists will follow in individual articles
- Clinical genetics units – an invaluable source of advice for rare diseases
- Orphanet (European portal for rare disease) www.orpha.net
- Useful contact numbers for specialist centres, disease/location specific, publishes journal dedicated to information on rare diseases
- Rare Disease UK www.raredisease.org.uk
- National alliance for people with rare diseases and their families
- Unique www.rarechromo.co.uk
- Charity supporting patients and families with rare chromosome disorders
- Genetic Alliance UK www.geneticanalience.org.uk
- National charity for those affected by genetic disorders
- Contact a Family www.cafamily.org.uk
- For parents of disabled children in the UK
- Individual disease support groups
- (more details in following series)
- In Birmingham, we are coordinating a Rare Disease Centre for children, adolescents and adults within our Institute of Translational Medicine. We aim for this to become a centre of excellence for patients with rare diseases by providing a unit where health professionals can optimise medical and nursing care of these patients, increase knowledge of disease processes and instigate clinical research trials.

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