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EDITORIALS

Developing a strategy for the management of rare diseases

Needs central coordination and input from patients

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Three million people in the United Kingdom have a rare disease, defined in both Europe and the United States as a disease that affects fewer than one in 2000 people. It is well recognised that those with rare diseases face intrinsic inequalities in healthcare, and in response to a 2010 recommendation by the European Commission, the UK government, like other member states, agreed to produce a strategy for rare diseases by 2013.

Liam Donaldson, when chief medical officer for England in 2009, paved the way for this in "Rare is common," a crucial chapter in his annual report,[1] which recalled that patients with rare diseases tend to be diagnosed late, find it hard to get information about their condition, are slow to benefit from therapeutic advances, and struggle to adapt to the day to day demands of what are often chronic conditions with a genetic cause. Subsequently, Rare Disease UK (RDUK), an alliance of many organisations, undertook an extensive analysis of what is needed and determined that a successful national strategy would integrate research, prevention, diagnosis, treatment, care and support, information, and commissioning.[2]

The Department of Health's rare diseases consultation process that will end on 25 May 2012 acknowledges the need for a coordinated response that empowers patients.[3] However, the selection of preformed questions embedded in the consultation suggested several blind spots.

The consultation suggests that commissioning to provide complex networks of care for patients with rare diseases will be the responsibility of the NHS Commissioning Board. If so, the board will need to become highly responsive to many patients' voices and powerful enough to harness the benefits of current research, which are currently widely distributed across trusts and universities.

A strategy that approves centres of excellence for rare diseases, similar to the French approach, would be easy to administer but would probably serve only a fraction of the 6000 or so recognised rare disorders. Nationally commissioned services in the UK currently deal with only 60 diseases. For some diseases, however, a geographically placed centre of excellence would be inadequate. For example, someone with a particular rare disease may at different times require urgent management in a local hospital with advice from an external expert source; local monitoring in primary care while stable; or a one-off complex procedure that is provided only in one or two institutions in the UK. It is unclear how an integrated service would be commissioned and funded. The concept of a "centre of expertise" may need to be functional or virtual rather than geographical, and it would need to overcome the organisational restrictions that now block working across different trusts, primary care, and social care.

Might any existing strategies inform the national consultation? A strategy currently being rolled out by the Renal Association is attractive.[4] With very light governance from the association, expert groups that include researchers, patients, and clinicians from across relevant disciplines work on a particular disease or disease grouping. Thus, the plan devolves to experts at a grass roots level where possible and centralises efforts when appropriate. Patient empowerment and engagement, and national liaison, are key aspects. Patients, either as individuals or more often through a patient support organisation, influence the thinking and direction of expert groups. Literature is produced for professionals and the public. Patients and researchers can contact each other. The expert group is required to advise commissioners about care pathways that affect patient experience and the patient journey. Thanks in large measure to funding from the British Kidney Patients Association and Kidney Research UK, 11 groups have become operational in the past two years and the number is expected to double by 2015.

An important aspect of the Renal Association plan is a sustainable secure web based patient registry that has been commissioned for all expert groups to use (www.RareRenal.org). It is built on the success of both Renal Patient View and the UK renal registry, and it has clear governance and ethical approval. Uniquely, this system allows patients to see their own

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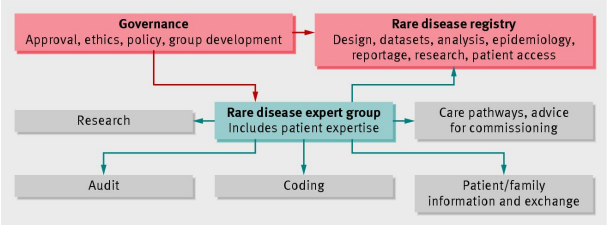
specialised clinical information and acts as an electronic health record. The registry has various levels of access, which makes it a safe platform for audit and research (figure). The consultation on rare diseases has recognised the importance of registries. However, beyond acknowledging that ICD-10 (international classification of diseases, 10th revision) is inadequate, it avoided discussing the closely related problem of diagnostic coding, for which a better system is clearly needed.



The pivotal role of an expert group: the Renal Association subsidiary model. Output tasks shown in grey are devolved to the group; centralised components are shown in orange

It is worrying that the consultation document makes no mention of a national coordinator, which was clearly recommended by Donaldson to identify patients who are not well served, to give patients a voice, and to be finally responsible for the delivery of the strategy as it unfolds—a rare diseases czar.[1] Any strategy for rare diseases will need to be able to respond flexibly to rapidly advancing technologies and treatments. Clinical processes and outcomes will change. A national coordinator is key to monitoring the impact of implemented strategies on health services and ensuring that changes are implemented quickly.

Unsurprisingly, the UK's rare diseases strategy is set to be cost neutral. Late or erroneous diagnoses are wasteful of NHS resources, as well as costly in personal terms for those who are affected, and earlier diagnoses will save resources. Advances in genetic testing promise reduced diagnostic costs. Exome, or whole genome sequencing (with appropriate interpretation), is predicted to make dramatic inroads into delayed and missed disease attribution and will inevitably result in more patients being managed using a personalised medicine approach—where clinical pathways can be tailored to suit the individual.[5] However, expert advice, some of it currently unavailable, will increasingly be needed. Although there are great opportunities, comprehensive management of those with rare diseases lies somewhat uneasily with both the market incentivised NHS envisioned in the Health and Social Care Act and territorial competition between foundation trusts.

The proposed national strategy for rare diseases has the potential to affect working practices in the NHS profoundly. Unlike more common diseases, evidence based outcomes may not currently be known, or even knowable, for rare diseases. The joint journey of clinician and patient requires trust and a flexible organisational structure. The strategy should support a new relationship between patient and doctor,[6] one of shared decision making in the context of highly detailed biomedical information made interpretable for both care provider and recipient. Its success will be measured by its ability to deliver coordinated and inclusive care to the 5% of the population who deserve better than they are currently getting.

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