3 Effectiveness and quality

Like care for other, more common chronic diseases, our specialist IMD care is directed at achieving a range of improved outcomes for patients and their families. Some outcomes – such as mortality – are more measurable than others, which relate to quality of life; but all are important. As the diseases are varied, the precise nature of the outcomes to be achieved will vary. In some cases outcomes have been investigated and audited for certain diseases and treatments (e.g. control of PKU during pregnancy and pregnancy outcome). However, we found no examples in the UK or worldwide where outcomes for specialist services directed at the whole range of IMDs have been described or investigated.

We set out below the important outcomes for specialist IMD services, which the stakeholder group discussed at various stages. These are illustrated with clinical examples derived from case histories. The aspects of the specialist management that facilitated these outcomes were also described – giving the possibility of setting some standards of structure or process that would be expected to lead to better outcomes.

The main categories of outcome are as follows:

- Decrease in mortality
- Decrease in morbidity
- Reduction in disability
- Prevention of harm to family members
- Prevention of damage to unborn child
- Reproductive choice
- Overall quality of life (reduction of handicap).

Decrease in mortality

A decrease in mortality can be measured as longer survival, older age at death, and a higher proportion of patients surviving into adulthood. Some data on mortality are given in the epidemiology chapter (Chapter 2), where it is reported that about 11 per cent of patients with IMD survive into adulthood. It is expected that more will survive into adulthood as a result of better recognition, improved management and new therapies.

Our case histories illustrate how mortality may be prevented through the following means:

- Better and more rapid initial diagnosis. For example, the child with Maple Syrup Urine Disease (MSUD) in the dietitians' case history (a) (Chapter 8) was encephalopathic and required ventilation and dialysis when transferred from the local DHG to the specialist unit where he was diagnosed and started on emergency dietary management.
- Management of acute crises. The same child required an emergency regime to prevent potentially fatal metabolic decompensation every time he developed intercurrent illness.
- Prevention of potentially fatal complications. In case history 4 (Appendix 3), specialists needed to recognise and manage the potentially fatal consequences of childbirth in a woman who appeared to be mildly affected by ornithine carbamyl transferase deficiency.

Decreased morbidity

This includes reduction in number and severity of complications arising from the disease and reduction in disability.
IMDs cause harm to many organ systems, both through deficiency of critical intermediate products or specific final products, the accumulation of products usually present in small quantities ‘upstream’ of the defect, or sometimes the toxic effects of products derived from alternative metabolic pathways.

PKU is a good example of a condition where harm is caused to various organs by the build-up of intermediate metabolites. Phenylalanine accumulates in the body owing to deficiency in the enzyme that breaks down this substance. High levels cause damage to the nervous system, with severe learning disability and neurological problems. If identified in the newborn, special diets can be instituted which prevent these problems.

Case history 1 (Appendix 3) gives an example of a child with methylmalonic acidemia (MMA), an organic acid disorder which can present with non-specific symptoms in the newborn. The local paediatrician needed to be aware of the possibility of an IMD, to have access to discuss the case with IMD specialists, and to arrange specialist investigations with the specialist biochemistry service. In this case some neurological damage occurred as the child had brain damage before the diagnosis was made.

Reduction of complications is another issue. The nursing case history (b) (Chapter 7) illustrates how a diagnosis of Fabry disease may lead to ERT, which may slow down the development of renal complications, heart and brain damage.

Reduction of disability

Case history 1 (Appendix 3) shows how, for a boy with MMA, a large number of services needed to be coordinated in order to help deal with the child’s disabilities and help his family to cope. Multi-agency support had to be provided in the community, including nursing (to manage gastrostomy and overnight pump feed), physiotherapy, occupational therapy, wheelchair services (for movement disorder and kyphoscoliosis), education and psychology. Coordination of a wide range of services must take place, the specialist team being able to optimise long-term medical care, predict and prevent complications as far as possible, and provide expert advice to the multi-agency team and family.

Prevention of harm to family members

Because these diseases are genetic in origin, other family members may be at risk of the same condition. They may be unaware of this as some have non-specific symptoms, or they may be only mildly affected, or onset might be in later life. However, for some conditions, such as Fabry disease, treatment can be offered with the chance of reducing longer-term organ damage.

In case history (e) (Chapter 7), specialist nurses described the case of a 43-year-old man diagnosed with Fabry disease following the finding of protein in his urine and subsequent discovery of renal damage and cardiac involvement. Three asymptomatic sisters required counselling and screening and two were found to be carriers, with potential risk to any offspring. With knowledge of the family history, a nephew who had presented with a stroke at age 41 was also found to have the disease and his family was also counselled.

Prevention of damage to unborn child

High levels of intermediate metabolites, such as may occur in PKU, cause damage to the unborn child, so it is important that women with IMD are warned of the risk of this and encouraged to seek advice before and during pregnancy.

Dietitians reported the case of a 30-year-old woman with PKU who was treated from birth with
diet (case history (b), Chapter 8). Her diet was relaxed during teenage years, but services had impressed on her the need to go back on a strict diet and ensure good control before and during any pregnancy to ensure that the unborn child was not harmed by the high phenylalanine levels. She attended a period of re-education prior to pregnancy, embarked on her new diet, and was monitored closely during pregnancy, with a successful outcome.

**Reproductive choice**

Again, because of the genetic nature of the conditions, there may be a risk that the offspring of a patient will also have the disease. Patients need to be counselled about this risk, and advised on their various options to have as healthy a child as possible.

Case history 4 (Appendix 3) presents the case of a woman with OCT deficiency. Although relatively mild in females, this can have very severe and fatal consequences in male offspring. This case shows how a woman was given information about this as a teenager, and was subsequently counselled and supported through a pregnancy, in which she chose to have prenatal diagnosis and eventually a termination owing to the finding of an affected male fetus.

**Quality of life**

Patients and their families seek to have as normal a life as possible, and not to be limited by disease in their fulfilment of educational and social potential. They want to achieve the maximum in educational attainment, having a job, integrating into society, participating in family life and generally experiencing psychological well-being.

The role that specialist services may play is described in the nursing case history (d) (Chapter 7). The specialist nurse looking after a teenage girl undergoing treatment for Fabry disease described multi-disciplinary work in caring for the 17-year-old, who became depressed and exhibited self-harming behaviours, including self-abuse with alcohol, drugs and sexual promiscuity. The nurse was instrumental in obtaining and coordinating support from the mental health crisis team, from counselling, and from drugs and alcohol rehabilitation teams in statutory and voluntary agencies.

The role of voluntary organisations is extremely important in helping families adapt and cope with an IMD and get the most out of life. Voluntary organisations can usually give personal support, advice and guidance to parents at the time of diagnosis; provide information about the condition; put parents in contact with other families affected by the condition; and help them to cope with complex therapies or difficult behaviours. Very importantly, they often also act as advocates for the family to help them to get services such as special schooling, mobility services, and even special health treatments. Specialist services, with their understanding and relationship with the voluntary organisations, are critical in helping patients and parents make contact with support groups and gain maximum benefit.

**What is needed for effective care?**

Effective care requires:

- making the correct diagnosis as quickly as possible
- instituting appropriate immediate therapy as soon as possible
- ensuring longer-term maintenance regimes are in place and being followed
- surveillance for possible complications and appropriate management
- prevention of complications which can cause death or further morbidity
- identification of family members at risk
- counselling and testing for family members
• identification of risks to pregnancy or fetus
• surveillance during pregnancy
• availability of counselling and antenatal genetic testing
• interdisciplinary work with and support from social services, education and voluntary organisations.

Effective services

We do not have systematic measures of outcome by which to judge the effectiveness of IMD services. However, some structural and process measures could act as proxies. Following discussion, we suggest that the following requirements would be important for providers to achieve effective services.

Structure

1. Availability of full specialist clinical and laboratory team to provide advice, support, and services for children, adolescents and adults
2. Clinicians and laboratory staff with appropriate level of specialist education and undertaking continuous professional development (CPD)
3. Adequate professional support
4. Professionals with adequate resources (including out-patient capacity) to undertake the necessary volume of work
5. Availability of full specialist clinical and laboratory team on a routine and emergency basis; and specialist services with formal relationships with the main feeder hospitals to provide diagnostic and management advice
6. Provision by specialist services of information on access to advice, testing and referral
7. Provision by specialist services of education to help non-specialists recognise IMDs and provide care appropriate to their level of expertise
8. Protocols for joint management and shared care to provide ongoing care and manage crises as far as possible
9. Arrangements for shared care to oversee long-term management and ensure referral back to specialist services at times of particular concern, such as during preparation for pregnancy and before surgical operations
10. Formal arrangements between specialist IMD team and genetic services
11. Formal arrangements between specialist team and a wide range of other specialties such as cardiology, renal, obstetrics and neurology
12. Full range of information to help patients make contact with relevant voluntary organisation
13. Active audit programme
14. Education and training programmes for specialists and other health professionals.

Process (activity)

15. Services should be undertaking the necessary volume of work to maintain sufficient experience across the breadth of IMDs.
Quality of care

Effectiveness is a vital element of high-quality services. However, there are other dimensions of quality that are important to professionals, patients and society as a whole. These include the following:

1. Efficiency. What is the most efficient way to deliver a specialist service? Is there some element of critical mass for efficient services?
2. Accessibility. Can patients get treatment when and where they need it? Are there barriers such as lack of information, distance, inability to pay, breakdown in service availability or waiting times?
3. Equity. Are there failings in equity? Are some groups of patients treated differently on the grounds of disease category, geography, age or ethnic group?
4. Relevance. Is the overall pattern and balance of services the best that could be achieved?
5. Acceptability. How is the service perceived by those who receive it (or might receive it)? Is it relevant, fair and responsive to demand? Is it what patients want and is it what professionals judge good practice?

These were key questions for us to address throughout the work, and they will be reported on further after the main reviews of services (see Chapter 11).