7 Nursing services

1 Introduction

An experienced nurse plays an important role in the multi-disciplinary team for IMDs. The work is highly specialised and involves complex aspects of care for individual patients, as well as working on familial aspects of disease with the extended family. In addition to direct clinical work with families, specialist nurses take on roles within the organisation such as leading clinics, providing first-line telephone information and advice, undertaking teaching, coordinating services, researching and becoming involved in commissioning.

A review of the nursing role and current service input was undertaken as part of a larger needs assessment and review of IMD services in the UK in 2005. Information was gained from a survey of main centres providing specialist services, a survey of specialist nurses in the nurse network of the British IMD Group, and a focus group of nine specialist nurses from centres around the country.

2 Review of the specialist nursing workforce within the UK

2.1 Overview

The main survey revealed a total of 29 nurses working in specialist IMDs, based in 14 centres across the UK. Ten metabolic service providers did not have a nurse specialist; these included all of the services in Scotland and Northern Ireland. In a further four centres the nurse specialist was single handed. The total number of WTE devoted to the specialist IMD services was 22.6, representing 78 per cent of potentially available nursing time.

Through the BIMDG nurses group, a questionnaire was sent to 23 nurses working in the field of IMDs. Fourteen replies were received (61%).

Figure 7.1 gives a summary of the nursing workforce in the various services across the UK.
2.2  **Job titles**

There appears to be a range of job titles for nurses working in the various services providing care for those affected with IHDS. The most common title was Clinical Nurse Specialist. Others include:

- Clinical Research Nurse
- Liaison Sister
- Metabolic Research Nurse
- Nurse Specialist
- Practice Educator
- Sister.

The age ranges of respondents was 25–34 (25%), 35–44 (42%) and 45–54 (33%).

2.3  **Professional qualifications, training and experience in inherited metabolic disorders**

Nearly all respondents indicated that their professional qualifications included RGN or RN, and two thirds had a primary degree. Three of the respondents (20%) had or were in the process of getting a higher degree.

The formal training reported included study days, conferences and training on the job.

The duration in the current posts of respondents is shown in Figure 7.1. Almost 75 per cent had been in post for one to five years, 21 per cent more than six years, and 7 per cent less than one year.

**Figure 7.2 Duration in current specialist post**

The pathway to the present post of respondents was varied. The commonest route was from a general nursing post involving some experience of the care of those with IMDs.

3  **Specialist nursing roles in inherited metabolic diseases**

Following a discussion of special expertise in clinical cases with a group of nine nurse specialists in IMDs, clinical specialist roles were agreed to include:

- providing specialist information, practical and emotional support to patient and family in acute phase of illness and over long periods
• providing specialist expertise to other clinical teams
• managing acute crises
• multi-disciplinary work
• managing familial aspects of disease, including dealing with potential dilemmas of family screening
• managing clinical trials expectations
• managing misdiagnosis from other services.

These are illustrated in accompanying case histories.

Other general roles include:

• education
• coordination
• telephone advice
• providing a nurse-led service
• setting up, monitoring and tendering for homecare systems
• administrative responsibilities in relation to NSCAG.

3.1 Case histories to support specialist clinical and other roles

The following eight case histories illustrate the specialist nursing roles in IMDs in the context of clinical care.

(a) Supporting parents with an acute presentation in a newborn baby

Judith was a newborn baby girl to parents of a consanguineous marriage. They previously had a male child who died as a neonate and the brother of one and sister of the other (who married each other) had a male infant who had died just two weeks previously. The nurse specialist was asked to see the mother in the maternity unit. The baby at this time was in the intensive care unit undergoing ventilation and haemodialysis with a diagnosis of OCT deficiency.

Her main role at first was to help the mother and family understand the condition by explaining it in simple terms. They also needed to start to understand the familial aspects and come to terms with the perceived guilt of having passed on a genetic condition.

As Judith got through the acute phase of the illness, the nurse became involved in many arrangements for the transfer home, liaising and providing education for community staff and local dietitians and arranging post-discharge follow-up. She made frequent home visits and provided support through several admissions for metabolic decompensation when Judith became ill again.

(b) Providing specialist information, practical and emotional support to patient and family over long periods

Richard was a 45-year-old man with Fabry disease. He was referred to the specialist unit, having been diagnosed the previous year with Fabry disease when he was seeing the cardiologists for left ventricular hypertrophy.

Richard attended the specialist clinic with his wife and was seen first by the clinical nurse specialist, who gave them some information about Fabry disease. The nurse also told them about some of the other investigations that would be recommended, and allowed the family plenty of time to ask questions prior to their appointment with the consultant. The nurse went on to organise the necessary investigations. It was found that his renal function was relatively poor.
Richard fitted the criteria for ERT and the nurse saw him on a fortnightly basis to administer this.

During the subsequent year, Richard’s renal function deteriorated, causing both him and his family a great deal of concern. The nurse spent a lot of time on the telephone with them, trying to respond to their anxieties. The family had felt that by his starting ERT, his renal function would be preserved. It was reiterated that whilst ERT might slow the progression to chronic renal failure, Richard’s kidneys were quite damaged by the time ERT was started.

Eventually, his renal function reached the point where he required peritoneal dialysis, and he was also put on a waiting list for renal transplant. At this stage the nurses provided support to their colleagues on the renal unit, giving information about Fabry disease, and in particular guidance on such aspects of management as ERT. The importance of continuing with ERT was also emphasised to the family as this would help protect Richard’s heart and brain from further damage due to Fabry disease.

(c) Managing an acute crisis

Derek, aged 38, was a patient with Gaucher disease who attended a national specialist centre for treatment and monitoring. He was admitted to his own local hospital with severe back pain, numbness in the left leg and evening pyrexia. With his consent, his wife contacted the specialist nurse at the centre and, as there was a shared care protocol in place between the centre and the local hospital, the specialist nurse was able to become directly involved in his care. She was able to give advice straightaway on pain management, advising the local staff to refer to the pain management team to provide an effective regime. She was also able to advise medical staff on the appropriate investigations, and results were then evaluated by both local and centre staff. Differential diagnosis was important to exclude the possibility that Derek might have serious spinal complications. This was subsequently ruled out following X-ray and MRI scanning, which was reviewed by the specialist centre. During this time the clinical nurse specialist made a number of visits to the local hospital, provided much information and emotional support to the patient and family – whom she knew well – and teaching and training to the ward staff to help them to manage this case effectively.

(d) Multi-disciplinary work to manage the angry adolescent

Mary was a 17-year-old symptomatic patient with Fabry disease and exhibiting low enzyme levels. She had a family history of the disease, in that her father was quite severely affected and died aged 46 having had several strokes and having developed renal failure requiring kidney dialysis and experienced a failed kidney transplant. No mutation was found. She had a sister and two nephews affected with the disease.

Mary was recently recruited to an ERT trial and was determined to ‘find help’ for her nephews. However, she was still grieving for her father, who had died only 12 months previously, and was going through quite a traumatic grieving process, exhibiting anger, denial, heartbreak, disbelief, self-abuse with alcohol and drugs and being sexually promiscuous. Surprisingly, she was very compliant with those providing care within the trial – having skin, kidney and cardiac biopsies – but very irrational and badly behaved outside this. To the nurse she admitted to drinking a bottle of vodka a day and smoking cannabis.

Just before the second renal biopsy, she told the nurse that she wished to kill herself as she could not deal with the death of her father and the extreme pains in her hands and feet that she suffered as a result of the disease.

The nurse recognised this as a crisis, and in responding enlisted the help of the mental health crisis team, ongoing counselling and the drug and alcohol rehabilitation unit – as well as working with the IMD physician, Mary’s parents and Mary herself. This illustrates the multi-disciplinary approach to patient management instigated by the specialist nurse, including liaison with various other disciplines for variable time periods.
To the uninformed, Mary looked well. The special relationship with the nurse enabled this crisis to be managed safely with a positive outcome and ongoing care.

(e) Managing familial aspects of the condition

John was a 43-year-old, apparently healthy, man who was diagnosed with Fabry disease following a routine medical in association with new employment. The discovery of proteinuria led to referral to a renal physician, and a scan of the kidneys showed shrunken bilateral kidneys. Further investigation led to a diagnosis of Fabry disease and it was found that he also had cardiac involvement. A genetic analysis identified a gene mutation. There was no known family history.

John was referred to a specialist centre, where it was decided that family members at risk should be traced and testing offered. The nurse specialist made contact initially with John and began to construct a family tree. John’s mother had died of renal failure; this was now thought to be Fabry disease. John had three sisters who were counselled and all were screened. All three sisters were found to be gene carriers and two, although asymptomatic, had some renal damage.

While screening the family members considered at genetic risk, John informed the centre of his nephew, Stephen, presenting at his local DGH with a stroke at 41 years. The nurse specialist visited Stephen at the DGH. He had presented with a right-sided hemiplegia, no speech, and post-stroke seizures. The family had no knowledge of the existing diagnosis in the uncle’s case. On further exploration of Stephen’s medical history, signs and symptoms of Fabry disease were found to have been present in childhood. He had felt pain in his hands and feet as a boy, and was found to have angiokeratomas (benign vascular skin lesions characteristic of Fabry disease). Stephen was invited to the centre, and following an assessment commenced on ERT.

Stephen and his wife had four children. Two of the daughters confirmed that the eldest daughter was adopted at birth and they had no contact with her. They now realised that she would have inherited the Fabry gene and they wanted her to have this information. The nurse specialist ensured that information was forwarded to the adoption agency. They contacted the girl, resulting in her referral to the specialist centre.

(f) Dealing with potential dilemmas in family screening

Susan, aged 29, with three young children aged 12, 8 and 4, using an internet website Genes Reunited discovers her half-sister and makes contact. She had been brought up in a care home and this is her first contact with blood relatives.

The newly found sister informs her relations that there is an IMD in the family — Anderson-Fabry disease. She puts her half-sister in contact with a specialist centre where she can receive counselling and advice. Using the internet, she has already gained some lay-person knowledge of the condition, but some of it is misleading and frightening. The team at the specialist centre is able to give helpful and reassuring advice and education that enables the mother to make a choice about screening her children and herself.

Genetic screening for Anderson-Fabry disease is undertaken, and the results show that the mother, her two sons and daughter test positive for the family DNA mutation. Further testing of cardiac and renal function enables them to make informed choices about treatment.

With legislation that enables adopted children to seek out their parentage and the prevalence of electronic means of doing this, such situations will become more commonplace. The role of the metabolic team is to prevent these dilemmas from becoming a family crisis.

(g) Managing misdiagnosis

This case history highlights the problems of non-specialist centres giving information without up-
to-date information, and the importance of parents having access to someone who is really knowledgeable about a condition at the time of diagnosis and during management, and who has networks to provide parent support.

Indira was a newborn baby girl who presented with severe neonatal jaundice and organomegaly. Initially it was thought that she had a severe infection and she was treated with antibiotics. However, there was no resolution and the liver disease persisted over the next few weeks, when it was thought that she had biliary atresia. Her parents were told that the liver disease was so severe she would die. A scan, followed by surgery, showed that this was not the case, and over the next six months she had further four liver biopsies. Eventually storage cells were found in the biopsies and a full metabolic work-up was performed, which eventually showed that she had Niemann Pick disease.

At this point, the parents were informed that Indira would not develop normally, would never walk or talk, and would die before the age of 2 years. By the time she was referred to the specialist team, the parents had already been referred to the local palliative care team.

The first task for the specialist nurse was to pick up the pieces as the parents were in despair. She was able to tell the parents that the natural history of the disease was variable, and they were introduced in the clinic to parents of children up to the age of 5 who had no neurological problems, so that they could see that the outlook was not as bad as they had originally been told. They were also told that there would be a possibility of genetic counselling for them, and that there might be other options for them if they wished to have more children.

When Indira was seen in clinic, at age 9 months, she was still very jaundiced. However, the last report on her at age 17 months was that she was developing well, had learned to walk and was speaking about 18 recognisable words.

(h) Managing clinical trials expectations

Neuropathic Gaucher disease is a life-limiting illness with very poor genotype-phenotype correlation. ERT has changed the prognosis of this disease, maintaining children’s visceral disease relatively well. Family fears of neurological deterioration and premature death are high.

Clinical trials exploring a smaller molecule to halt neurological progression started in July 2003. Parental expectations were extremely high. This was seen as the new ‘cure’. The delay in commencement of the trial led to anxiety, anger and frustration.

Managing parental expectations in a realistic way is very problematic. Parents hear what they want to hear. They cling on to the information they want. The research nurse must maintain a realistic view of the issue. The risk of their child ‘failing’ screening prior to admission to the trial due to poor cooperation, and especially level of disease progression, adds significant tension and stress. When one sibling ‘succeeds’ in screening and starts treatment while the other sibling fails (often being the more severely affected sibling as well), this opens up an array of emotions that are very difficult to support and manage. Randomising siblings (even if the trial is blinded) causes similar emotions and situations.

The research nurse managing the trial must also inform and educate other professionals and departments about the impact and implications of the trial, and treatment on clinical management and resources and so on, and future prediction of services.

Metabolic treatments being developed and introduced into clinical care are an increasing aspect of our workload. As well as understanding the disease itself and the various treatments, the nurse needs to be fully conversant with ethical considerations in clinical trials, research governance, liaising with other professionals, working with the pharmaceutical sponsor, and the reporting and presentation of results.
3.2 Organisational roles undertaken by the clinical nurse specialist

**Education**

Specialist nurses in IMDs have a key role in the provision of education of health professionals. This is undertaken in the context of care for the individual patient and more widely, including nursing students and any other allied health professionals, medical students, general practitioners and those training and undergoing CPD in paediatrics.

One further large element of education has arisen recently, in which nurses are involved in supporting the neonatal screening programmes and will be particularly involved in the roll-out of the MCADD programme with teaching required for midwives, health visitors, community paediatricians and local hospital paediatricians. This has been implemented on the whole by specialist nurses for IMDs.

**Providing telephone advice**

The specialist nurse is often the first port of call for GPs or geneticists who have a patient that they suspect or know has an IMD. The nurse will usually give information on the disorder, how to refer and what other tests to do to confirm the diagnosis.

Nurses are also called by patients who have seen, read or researched a disorder and believe they might be at risk. The nurse will provide information about the disorder and tell them if it sounds likely and how to get referral.

**Coordination**

Patients and families with severe chronic disease need a key worker to link the multi-disciplinary team and the wider group of health and other professionals and voluntary organisations concerned with the care of the family. The clinical nurse specialist often fulfils this role. She may initiate and then subsequently attend a multi-disciplinary team meeting. As the family’s needs increase, she may introduce other specialists and she may have a particular role here in coordinating appointments for the family who have long distances to travel.

**Provision of a nurse-led service to provide patient care**

This role is not yet officially recognised, but does occur in the IMD service. Experience of those undertaking such a service, notably at the Manchester Fabry Centre, is that this is very positive and a good way of dealing with diagnosis and future care monitoring.

**Setting up, and monitoring and also tendering for homecare systems**

Department of Health regulated, but privately run, homecare companies are often used in IMDs – for example in the administration of ERT infusions at home. It is increasingly the clinical nurse specialist’s role to be involved in commissioning and tendering for these services. Once set up, these systems need to be monitored continuously for quality by the nurse, using information gained from patient audit. The metabolic nurses are the first point of call for homecare nurses for crisis management.

**Administrative responsibilities in connection with NSCAG**

The CNS role with NSCAG includes administrative responsibilities such as providing activity data, quality reports and ERT financial monitoring. This also includes future service planning.
4 Education and professional development for specialist nurses

4.1 Education and training reported by UK nurses from survey

Respondents to the nursing survey commented on the present state of training for nurses and what could be improved. The main point was that there is no formal nurse training in IMDs. Most practitioners have simply learned on the job, sometimes with an initial bit of training as part of their induction. They then go on to have further training through ward rounds and attendance at various educational meetings. Some support to nurse education is provided through the nurses’ network of the BIMDG.

Educational needs

Formal and accredited education for nurses in IMDs is urgently needed, and should be at basic level as an introduction and at a more specialist level. It should cover clinical aspects, pathophysiology, genetics, biochemistry, dietetics, social and psychological aspects, research and development aspects, as well as the nursing role in providing support for patient and family.

As a preliminary to this, key competencies should be set out on a national basis and courses should be recognised and accredited.

As an adjunct, professional development for nurses should be supported through grants to enable attendance at national and international meetings, seminars and workshops. One example is that the annual Gaucher course at the Royal Free Hospital in London, currently offered to physicians from other countries, could be offered to nurses from Europe too.

5 Recommendations

Development of the specialist nursing role

1. The role of the nurse in the IMD team is highly specialist and integral to the service. There should be clinical nurse specialist posts in every major centre.

2. There are opportunities for clinical nurse specialists to take on substantial areas of the service. However, their extensive roles need to be properly recognised and adequate resources in terms of session time and support made available for them to be undertaken properly. This would allow services to:

   • provide high-quality nursing care
   • develop the wider health services to be able to provide competent care and thus support the role of the specialist service
   • care for more patients by freeing consultant medical time through the provision of nurse-led clinics and services
   • develop good networks of specialist care between centres and districts with shared care protocols.

Education

1. Education should be developed further and formalised as follows:

   • There should be development of a formal course at Master’s level, which should cover clinical aspects, pathophysiology, genetics, biochemistry, dietetics, social and
psychological aspects, and research and development aspects, as well as the nursing role in providing support for patient and family.

- There should be an agreed formal training programme that includes theoretical, practical and professional aspects of the work. This should be developed to include documentation of education by the trainee.
- Clinical nurse specialist posts in this area need to be created so that those in training can have an expectation of a post being available.
- Clinical nurse specialists in training and others already in post must be supported to undertake more formal education – recognising that this is highly specialised and cannot be undertaken simply within the home service.
- There should be more formal training rotations that take in a period of training at one of the main specialist centres. These centres should be formally recompensed for the time they spend teaching.
- Each region / specialist centre should have a clinical nurse specialist with a lead role for education in IMD.
- Clinical nurse specialists must have the necessary time and resources to undertake teaching.