8 Dietitian services

1 Introduction

An experienced dietitian plays an important role in the multi-disciplinary core team for inherited metabolic diseases (IMDs). The work is highly specialised and complex. Good dietary management is crucial to the outcome in many of these conditions.

The specialist dietitian leads and is responsible for the individualised dietary management of children and adults with IMD. This involves formulating the diet and teaching patients, parents, carers and other relevant lay persons about the patient’s dietary treatment. The dietitian provides support and collaborates with smaller specialist units and district general hospitals, providing expert advice and education to medical, nursing and allied health professionals. In addition, the dietitian will provide advice and support on the dietary management of patients with IMDs to other professionals in health, social care and education. Dietitians are also involved in research, development of protocols and education.

A review of the dietitian’s roles 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 by: a survey of main centres providing specialist services; a survey of dietitians through the BIMDG dietitians subgroup; and a focus group of three dietitians from centres in London and Manchester.

2 Review of the dietitian workforce in IMD services in the UK

2.1 Numbers and distribution

Services were asked to provide information on the number of dietitians involved in the provision of specialist IMD services, including details on whole time equivalents (WTEs) devoted to IMD work. This included information from 24 service providers. In addition, questionnaires were sent to 27 dietitians identified through the BIMDG dietitians subgroup, asking for further information about areas of work undertaken, education, training and experience, and perceptions of unmet need and service shortfall. Seventeen replies were received. The results are shown in Figure 8.1.

A total of 35 dietitians working across the UK were identified by service providers, between them contributing 22 WTE. This accounted for 63 per cent of the potential WTE available if all were full-time on IMDs. In two centres providing core IMD services there was no dietitian, and in a further nine the dietitian was single handed. In only nine services was there more than one WTE dietitian.
It needs to be recognised that at Manchester and London GOSH 0.75 WTE and 1.0 WTE (2 individuals) respectively are training posts, not specialist posts.
In Newcastle one dietitian is split between child and adult services.
There are dietitians from other services managing IMD patients who work and liaise quite closely with the main centres – for example, in the context of outreach clinics. These dietitians might not have been included in the study.

Figure 8.1 Dietitian workforce in the UK

<table>
<thead>
<tr>
<th>Service</th>
<th>Patients/wte</th>
</tr>
</thead>
<tbody>
<tr>
<td>Manchester</td>
<td>460:1</td>
</tr>
<tr>
<td>Liverpool</td>
<td>438:1</td>
</tr>
<tr>
<td>Birmingham</td>
<td>366:1</td>
</tr>
<tr>
<td>London Guys</td>
<td>120:1</td>
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<tr>
<td>London GOSH</td>
<td>227:1</td>
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<tr>
<td>London UCH</td>
<td>265:1</td>
</tr>
<tr>
<td>Glasgow</td>
<td>115:1</td>
</tr>
<tr>
<td>Belfast</td>
<td>136:1</td>
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</tbody>
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2.2 Age range

Dietitians’ age ranges were 25–34 (20%), 35–44 (33%), 45–54 (40%) and 55+ (7%). Respondents were asked about duration in their current post. Almost 20 per cent have been in post for less than one year and fewer than 50 per cent have been in post for more than five years (see Figure 8.2).
3 Roles of specialist metabolic dietitians

3.1 Clinical roles

The clinical roles include the following:

- Formulation and calculation of IMD feeding regimens for patients when they are well or unwell.
- Prescribing specialised metabolic dietary products to be used in treatments.
- Intensive monitoring (biochemical, growth and tolerance) of response to dietary treatment with necessary dietary adjustment.
- Long-term follow-up and assessment, adjusting dietary regimen according to growth, biochemical findings and clinical progress.
- Assessment, monitoring and adjustment of dietary regimens during pregnancy.
- Education for patients, their families and carers using a range of communication skills.
- Providing counselling and emotional support to patients, their families and carers on the practical implementation and management of highly complex dietary regimens.
- Key worker, particularly for patients where diet is the sole or major treatment; thus, providing a link between the family, the multi-disciplinary team and community professionals.
- Education, support and liaison with health professionals and others involved in the care of the patient (including professionals from education and social services), both in the specialist centre and in the local community.
- Referral to other professionals as appropriate, such as to speech and language therapy, clinical psychology (e.g. for help in management of adverse feeding behaviour), feeding and swallowing problems, psychological support for managing dietary treatment.
### 3.2 Provision of therapeutic dietary treatment

Dietary therapy is the mainstay of clinical management in three main groups of IMD: disorders of protein metabolism (amino acid disorders, organic acidaemias, urea cycle disorders), disorders of carbohydrate metabolism, and disorders of lipid metabolism (see Figure 8.3).

**Figure 8.3 Inherited metabolic diseases requiring therapeutic dietary management (box for lists of main disorders).**

#### A Protein
- **Amino acid disorders**
  - PKU
  - MSUD*  
  - Tyrosinaemia
  - Homocystinuria
  - Hyperornithinaemia
  - (gyrate atrophy of the choroid & retina)
- **Organic acidaemias***
  - Propionic acidaemia
  - Methylmalonic acidaemia
  - Isovaleric acidaemia
  - Glutaric aciduria type I
- **Urea cycle disorders***
  - OTC deficiency
  - CPS deficiency
  - Citrullinaemia
  - Argininoosuccinic aciduria
  - Arginase deficiency

#### B Disorders of carbohydrate metabolism
- Galactosaemia
- Hereditary fructose intolerance
- Fructose 1, 6 bisphosphatase deficiency*
- Glycogen storage diseases* mainly types I, III, IX

#### C Disorders of lipid metabolism
- Type I hyperlipidaemia
- Familial hypercholesterolaemia
- Fatty acid oxidation disorders*
  - VLCAD
  - LCHAD
  - MCADD
  - Carnitine transport disorders
- Peroxisomal disorders
  - Refsums disease
  - Infantile Refsums
  - Adrenoleukodystrophy

* denotes disorders requiring emergency dietary regimen during illness

**Example 1 Management of an amino acid disorder: phenylketonuria (PKU)**

PKU is due to a deficiency of the enzyme phenylalanine hydroxyylase. This results in the accumulation in plasma of the essential amino acid phenylanine (phe) which is neurotoxic. Without dietary treatment this accumulation would cause severe learning difficulties. Phenylalanine is normally converted to tyrosine, so tyrosine becomes deficient. To prevent phe toxicity, the diet needs to be restricted in phe intake. Tyrosine is supplemented to ensure adequate intake to prevent deficiency of the essential products of tyrosine degradation. Dietary treatment continues throughout childhood. Some adults remain on the diet, and diet is essential during during pre-conception and pregnancy to prevent damage to the baby, such as microcephaly and congenital malformations.

The dietitian will formulate, manage and continuously monitor a low-phe diet to support normal growth and development as follows:

1. Restrict natural protein intake to maintain plasma phe level within desirable reference range for age
2. Prescribe phe-free amino acid supplements to achieve an adequate protein intake
3. Ensure adequate intake of tyrosine and prescribe tyrosine supplements in maternal PKU
4. Prescribe vitamin and mineral supplements
5. Maintain normal energy intake using special low-protein prescribable foods
6. Manage the diet based on plasma phe results.

**Example 2 Management of a disorder of carbohydrate metabolism – galactosaemia**

Patients with classic galactosaemia are unable to catabolise dietary galactose (milk sugar) owing to a deficiency of the enzyme galactose-1-phosphate uridyl transferase. The presenting clinical symptoms seen in infancy – such as jaundice, hepatomegaly, liver failure and cataracts – will resolve with dietary treatment of strict avoidance of dietary galactose. The dietitian will advise on the exclusion of dietary galactose and monitor the diet to ensure an adequate intake of all nutrients, especially calcium. Dietary treatment needs to be lifelong.

### 3.3 Emergency dietary regimens

For some inborn errors of intermediary metabolism, intercurrent illness combined with a poor oral intake and fasting will cause severe metabolic decompensation. Such children may rapidly become extremely ill, sometimes encephalopathic, and require admission to intensive care units. To prevent this, an emergency diet is given which varies depending upon the disorder. The dietitian is instrumental in the provision of this, either at home by frequent telephone contact or in hospital.

### 3.4 Nutritional support

Patients with other IMDs – including mitochondrial disorders, LSDs and some peroxisomal disorders – do not require specific therapeutic dietary management as a primary treatment. However, individuals with these disorders may have more general nutritional problems, including swallowing problems which require dietetic intervention to provide nutritional support.

### 3.5 Case histories

Two case histories illustrate the intense and specialist clinical role of the IMD dietitian.

**Case history (a) The acutely ill child with an amino acid disorder**

Rajesh was born to first-cousin Asian parents. At 11 days old he became very unwell, with very poor feeding and lethargy, and was admitted to his local district general hospital. He had poor muscle tone and was encephalopathic. Initial investigations revealed raised plasma branch chain amino acids (BCAAs) and a diagnosis of Maple Syrup Urine disease (MSUD) was made.

He was transferred to the ICU at the tertiary metabolic unit, where he was ventilated and commenced on dialysis to reduce the plasma leucine level (leucine is neurotoxic at high levels). The mainstay of treatment for MSUD is diet (limiting intake of BCAAs). Dietary treatment was begun immediately on Rajesh’s arrival in ICU. A modular feed free of BCAAs was given. This feed had to contain sufficient BCAA-free amino acids and energy to promote the conversion of leucine to body protein, thereby reducing plasma leucine level. He was fed via a nasogastric tube.

Over the next few days, the dietitian reviewed his feeds daily. These were adjusted according to the plasma BCAA levels, weight, fluid allowance and feed tolerance. Branch chain amino acids were monitored daily during the acute phase.

BCAAs are essential amino acids. Once the high plasma leucine level had decreased, a measured amount of natural protein as infant formula was added to the modular feed to provide a source of BCAA. The amount of infant formula was altered to maintain plasma leucine levels within safe limits. Due to low levels of the other two BCAAs, valine and isoleucine, supplements of these were given to prevent them from being rate limiting for protein synthesis. The amounts prescribed were altered according to plasma levels.
The dietitian worked closely with the ICU nurses and intensivists as they had a very limited knowledge of MSUD and did not fully understand the importance of dietary management to the outcome. There was also close liaison with other members of the metabolic team (medical, nursing, biochemists), pharmacy and the special feeds unit.

After a few days Rajesh was well enough to be transferred to the ward, where he was gradually moved from nasogastric to oral feeds. During the next two weeks he was stabilised onto a feeding regimen which included a measured amount of normal infant formula, a BCAA-free formula, valine, isoleucine and calorie supplements. An emergency regimen would also be required for use during periods of intercurrent illness.

During this initial admission, one of the important roles for the dietitian was education of the family. This was complicated by the fact that the mother had limited English and any teaching about diet had to be undertaken in the context of an Indian family diet. The dietitian undertook teaching in several stages, using an interpreter, and always reinforcing verbal information with written information. The family needed to understand:

- the principles of dietary management
- what an MSUD diet consists of
- how to make up the special feeds very accurately
- how and when to put in place the emergency regimen for Rajesh if he should become ill
- what products can and should be used in the diet and how to avoid the risk of using the wrong products
- the importance of monitoring plasma BCAA levels and how the dietitian interprets these and makes dietary changes.

Sometimes teaching was in several sessions daily and included the extended family.

Before discharge, the dietitian made contact with health professionals who would be involved in caring for Rajesh when he returned home into his local community area, to explain his dietary treatment when well and unwell. This included consultant, local dietetic services, local pharmacy, health visitor and GP.

Rajesh was discharged home at 5 weeks of age.

After discharge he continued to have weekly blood tests to monitor BCAA. The results were interpreted by the dietitian, who phoned the family every week to discuss progress, weight, blood results and any necessary feed changes. A further period of intensive dietetic advice and support was needed at the time of weaning. The dietitian was also called by the family for advice when Rajesh was unwell, and discussed whether to institute the emergency regimen, when to contact the doctor or bring him to the hospital. She was also involved in giving advice regarding other medication and immunisations. Support to the family for Rajesh was also provided by involvement in community meetings to discuss what his dietary requirements in nursery and school were.

Case history (b) Management of maternal PKU

Barbara has phenylketonuria (PKU), which was diagnosed on routine neonatal screening. She was started on a phenylalanine-restricted diet at 2 weeks of age which was relaxed at 8 years of age and then further in her teenage year, but she still had great problems in adhering to the diet. When she was 27 she was allowed to come off the restricted diet completely and eat an unrestricted diet including high-protein foods.

At the age of 30 Barbara attended a clinic appointment with her partner to discuss that they were getting married in a few months and would like to plan a pregnancy straight away. The doctor and dietitian both confirmed the importance of Barbara going back onto a phenylalanine-restricted diet before she became pregnant and to continue this throughout pregnancy, to ensure that her unborn baby would not harmed by
high phenylalanine levels. They explained what would be involved in terms of re-education of the diet for pre-conception and during pregnancy, the frequency of blood tests, and the frequency of clinic appointments.

A date for re-education was set. Barbara attended the clinic and saw the dietitian and dietetic assistant (DA), who is involved in the practical dietary teaching/cooking with the patient. In total she attended for four days of intensive dietary re-education, and her husband attended the first day to learn more about PKU and what is involved with diet for pregnancy.

During the four days of re-education the following were covered by members of the multi-disciplinary team (individualised to Barbara’s needs) and coordinated by the dietitian:

- amino acid supplement tasting (RD)
- explanation of diet with written diet sheets (RD)
- tasting low-protein prescription products (DA)
- learning to do a finger prick test and send in a blood spot card (CNS)
- following recipes using low-protein prescription products (DA)
- calculating and weighing protein (phenylalanine) exchanges (DA and RD)
- supermarket visit (DA)
- planning meals and menus using a certain number of protein (phenylalanine) exchanges (DA).

DA = metabolic dietetic assistant, supervised by the dietitian
RD = metabolic dietitian
CNS = metabolic nurse specialist

The pre-conception dietary management aims for phenylalanine levels of 100–250 mmol/L. During this time Barbara had twice weekly finger prick phenylalanine blood tests. Blood results were reported by telephone and any changes to the diet discussed. Barbara became pregnant in April 2004. During pregnancy she had more frequent blood tests, three times a week. The diet became more complicated as she experienced nausea and vomiting and also had to reduce her protein intake still further to control phenylalanine levels.

During this difficult time Barbara required lots of support to help manage her diet — at least three telephone contacts per week. In addition to a phe-free amino acid supplement and a vitamin/mineral supplement, tyrosine supplementation was started at 16 weeks’ gestation. From 20 weeks’ gestation, protein (phenylalanine) tolerance starts to increase due to the growing fetus size and the fetal liver starting to produce the phenylalanine hydroxylase enzyme. During this time of increased phe tolerance, Barbara required further education on ways of incorporating the additional protein in her diet.

At 38 weeks’ gestation, Barbara developed gestational diabetes and her labour was induced due to very high blood glucose levels.

After the birth the dietitian ensured the baby’s Guthrie result (plasma phe level) was normal and reported this to Barbara. She also checked records of the baby’s head circumference, weight and length. The baby was reviewed by the paediatrician at 6 weeks of age and will be assessed further by a clinical psychologist at 1 year, 4 years, 8 years and 14 years of age to check developmental progress. After delivery Barbara was encouraged to return to a normal unrestricted diet and to ensure that she had a balanced diet to support breast feeding.
3.6 Provision of dietitian-led clinics

Dietitian-led clinics are increasingly being established. They may be held in collaboration with doctors and clinical nurse specialists or solely by dietitians. These clinics may be particularly useful for management of patients whose treatment is mainly dietary (e.g. PKU, MCADD).

Example of a dietitian/CNS-led clinic

At London GOSH a dietitian/CNS-led group clinic for PKU has been established. Groups of similarly aged patients with PKU and their families attend together to undertake education on different topics. A pre-clinic questionnaire is completed by the dietetic assistant on the telephone to identify dietary and other problems, allowing more time for discussion at the clinic.

3.7 Education and training for health professionals

This is a major aspect of the work and includes the following:

- Development of educational materials such as diet sheets, information leaflets specific to particular disorders, and visual aids in order to support other professionals looking after IMD patients.
- Formal and informal training on IMDs to dietitians and other health professionals (e.g. medical, nursing, pharmacy, allied health professionals).
- Mentorship and clinical supervision for non-specialist dietitians.

3.8 Audit, research and consultancy work

This includes the following:

- Development and evaluation of new dietetic treatment regimens, treatment guidelines and protocols.
- Audit and research.
- Provision of professional advice to voluntary organisations (parents’ support groups for IMDs, e.g. NSPKU, CLIMB).
- Provision of advice on IEM to the manufacturers of specialised metabolic dietetic products (e.g. advising on development of new products).

4 Education and professional development

4.1 Education and training reported by UK dietitians from survey

Nearly all of the respondents reported that their professional qualifications included a primary degree, and most had a postgraduate diploma in dietetics. Nearly half of respondents had or were in the process of getting a higher degree. The formal training in IMDs included:

- Experience gained via clinical practice, including specific rotations in metabolic dietetics
- Advanced Course in Paediatric Dietetics – Module 4
- SHS study days and meetings
- BIMDG study days and BIMDG dietitians subgroup study days
- SSIEIM (Society for the Study of Inborn Errors of Metabolism) meetings including international dietitians’ sessions.

Most dietitians came to work in IMDs from other paediatric dietetic posts, both general and in other specialist areas such as liver disease.
The survey showed their formal training in IMDs was limited. A few have participated in training rotations at the specialist centres, but most received training and gained experience on the job, which they supplemented as far as they could by training days such as those provided by the BIMDG. Dietetic posts offering training in IMDs are very limited. Currently only two specialist centres are known to have rotation training posts for their own staff (London GOSH and Manchester).

An educational opportunity which was highly valued by survey respondents was the current three-day Module 4 of the Advanced Course in Paediatric Dietetics, which is run by the Paediatric Group of the British Dietetic Association. Module 4 focuses on the practical dietary management of IMDs. The Paediatric Dietetics course team is currently working with the University of Plymouth to develop this module further to Master’s level, so that it forms part of a Master’s degree. This course would have extended content building on the current Module 4, and would include a total of about 200 hours study, with face-to-face teaching and some preparatory and follow-up study in the practitioner’s service location. This venture provides an exciting opportunity for further developing and formalising the work of IMD dietitians.

The BIMDG dietitians subgroup also plays an important role in networking and providing professional support. This is mainly done by group email and study days.

5 Recommendations

1 Staffing

1. The role of dietitians is highly specialised and integral to the IMD service. There should be a specialist dietitian post in every centre, with more than one in the main centres. The numbers of specialist dietitians required will depend on the patient case mix. The staffing profile should consider using an appropriate mix of staff such as dietetic assistants, senior dietitians, specialist dietitians, advanced practitioners and consultant dietitians.

2. There are opportunities for dietitians to take on substantial areas of the service. However, their extensive roles need to be properly recognised and time made available for them to be undertaken properly.

3. Specialist metabolic dietitians also need to be adequately resourced to undertake organisational work in developing networks and protocols with peripheral services, education, audit, research and CPD.

4. It would be advantageous to train more dietitians to undertake these roles — but as a prerequisite for this, education should be developed further and formalised.

2 Education

1. There should be a working party including workforce development members to develop and implement educational programmes and training based on required competencies and predicted workforce requirements. This should include consideration of the following elements:

   • 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.
   
   • Additional specialist dietitian posts in this area need to be created so those in training can have an expectation of a post being available.
   
   • Dietitians 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.
• The new Master’s course should be supported.
• 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 specialist dietitian with a lead role for education in IMDs.
• Specialist dietitians must have the necessary time and resources to undertake teaching.