

## **SPECIALISED SERVICES NATIONAL DEFINITIONS SET (3<sup>rd</sup> Edition)**

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### **Specialised Metabolic Disorders Services (all ages) - Definition No. 36**

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#### **Preface**

This definition is part of the third edition of the Specialised Services National Definitions Set (SSNDS) published in 2010. The second edition of the SSNDS (containing 35 definitions) was published in 2002.

The third edition of the SSNDS contains 34 definitions (4 definitions have been dropped and 3 new definitions have been added). Each definition has been updated or created by an inclusive process involving providers (clinicians, hospital managers, and information and coding staff), commissioners and patients' groups. Every effort has been made to ensure that each definition contains the all the relevant medical condition and treatment classification codes as well as referencing key policy and standards documents. The final version of each definition has been approved by the National Specialised Commissioning Group (NSCG) and endorsed by the relevant professional organisations. The third edition definitions are available from the NSCG website: [www.specialisedcommissioning.nhs.uk](http://www.specialisedcommissioning.nhs.uk)

The 10 Specialised Commissioning Groups, acting on behalf of their member PCTs, are responsible for the commissioning arrangements for specialised services. The purpose of a definition is to identify the activity that should be regarded as specialised services. A service is specialised if the planning population (i.e. catchment area) for that service is greater than one million people. This means that a specialised service would not be provided by every hospital in England; generally, it would be provided by less than 50 hospitals.

The definitions are not prescribed service models nor do they set service standards. Where national standards for a specialised service already exist, these may be referred to in the definition. Inclusion of a treatment or intervention in a definition should not be taken to mean that there is established evidence of clinical or cost effectiveness.

The content of individual definitions in the SSNDS will inevitably change over time as new healthcare services which are specialised are introduced into the NHS and other services, which were previously specialised, become commonplace and cease to be considered specialised. The SSNDS will be regularly reviewed and further editions will be produced in the future. Definitions may also be updated on an individual basis if that is appropriate.

Future editions of the SSNDS will become more refined as the classifications systems develop and become better able to categorise specialised service activity. The current classification systems used in the third edition are the International Classification of Diseases, version 10, and the OPCS Classification of Interventions and Procedures, version 4.5.

Queries and suggestions for possible improvements should be sent to the NSCG's email address at: [enquiries@nsscg.nhs.uk](mailto:enquiries@nsscg.nhs.uk)

## **1. Introduction**

Specialised metabolic disorders services cover a diverse range of primarily genetic conditions which, although varying widely in their presentation and management according to which body systems are affected, are all caused by a disruption in normal biochemical processes. They lead to severe disturbance of metabolic processes in the body, resulting in either a deficiency of products essential for health or an accumulation of unwanted or toxic products. This can cause disease or damage in many organ systems, leading to severe learning or physical disability and death at an early age.

Although the individual metabolic disorders are rare, collectively they represent a significant patient population. Approximately 10,000 prevalent cases attend specialist metabolic disorders centres for the management of their condition, but it is estimated that a further 6,000 children and 3,000 adults are lost to follow-up or never attend. There are estimated to be over 600 new cases per year, i.e. 1 in 1,000 live births.

This definition covers the core specialised diagnostic and treatment services for inherited metabolic disorders, and the provision of advice to family members. This activity is provided by specialist centres, supported by metabolic biochemistry laboratory services. Outreach clinics provided by specialised metabolic disorders centres in local hospitals improve accessibility and make best use of scarce expertise. Many patients are treated in local hospitals on a shared care basis with advice from the specialist centre's multi-disciplinary team on the condition itself and its impact on other illnesses.

The definition does not include services for non-specialised conditions such as familial hypercholesterolaemia, diabetes, etc, which are provided by local hospitals.

## **2. Rationale for the service being included in the Specialised Services National Definitions Set**

- Care is delivered by multi-disciplinary teams in a limited number of centres; there are currently less than 20 specialist metabolic disorder centres in England providing care for children and/or adults.
- The diverse range of conditions and wide variety of presentations for a relatively small number of patients necessitates a concentration of diagnostic and management expertise.
- Disorders are multi-system and involve input from other specialised services co-located in the same specialist centre.
- Treatments may be complex, expensive and rare e.g. enzyme replacement therapies.

## **3. Links to other services in the Specialised Services National Definitions Set**

Metabolic disorders can affect many organs and therefore specialised services for metabolic disorders require close liaison with a number of other specialised services as listed below. The specialty has particularly close professional relationships with the medical genetics and endocrinology specialties.

Definition No.2, Specialised Blood and Marrow Transplantation Services (all ages)  
Definition No.4, Specialised Services for Women's Health (adult)

Definition No.5, Assessment and Provision of Equipment for People with Complex Physical Disabilities (all ages)

Definition No.6, Specialised Spinal Services (all ages)

Definition No.7, Specialised Rehabilitation Services for Brain Injury and Complex Disability (adult)

Definition No.8, Specialised Neurosciences Services (adult)

Definition No.11, Specialised Renal Services (adult)

Definition No.13, Specialised Cardiology and Cardiac Surgery Services (adult)

Definition No.19, Specialised Services for Liver, Biliary and Pancreatic Medicine and Surgery (adult)

Definition No.20, Medical Genetics Services (all ages)

Definition No.22, Specialised Mental Health Services (all ages)

Definition No.23, Specialised Services for Children

Definition No.26, Specialised Rheumatology Services (all ages)

Definition No.27, Specialised Endocrinology Services (adult)

Definition No.29, Specialised Respiratory Services (adult)

Definition No.34, Specialised Orthopaedic Services (adult)

Definition No.37, Specialised Ophthalmology Services (adult)

#### **4. Detailed description of specialised activity**

The twin aims of specialised services for metabolic disorders are to diagnose and support affected patients so they may live as normal a life as possible and to provide genetic advice to families. The broad components of the specialised service for metabolic disorders are set out below.

##### **4.1 Diagnosis**

Patients with the metabolic disorders may initially present to primary or secondary care services with varied and often non-specific signs and symptoms. The specialist metabolic disorders centre provides accurate specialist clinical and laboratory diagnosis for patients presenting with clinical symptoms compatible with suspected metabolic disorders. In some cases acutely ill neonates, children and adults will require an urgent diagnosis. The specialist centre also provides confirmatory tests for babies identified through the newborn screening programme and for other family members at high risk.

- urine amino acids
- plasma amino acids
- urine organic acids
- blood carnitine and carnitine species
- other metabolites including very long chain fatty acids, homocysteine acids, intermediary metabolites, orotic acids, bile acids, 7-hydrocholesterol metabolites, glycosaminoglycans, purines and pyrimidines, and creatine synthesis metabolites
- abnormal proteins e.g. carbohydrate deficient glycoproteins
- cerebro-spinal fluid neurotransmitters
- enzymes (there are around 200 different enzymes) including enzymes in leucocytes, fibroblasts, tissue (liver/muscle), red blood cells and plasma.

Molecular (DNA) tests are increasingly used to aid diagnosis and can be undertaken in either specialist genetic laboratories or in specialist metabolic laboratories.

## **4.2 Patient management**

Patient management is provided by a multi-disciplinary team at the specialist metabolic disorders centre. This will require both clinical and laboratory elements (e.g. for dietary support monitoring).

Most care is provided on an out-patient or shared-care basis with some of this being led by the specialist nurse or dietician member of the specialist metabolic disorders centre team. Rare and expensive drugs are sometimes used in patient care, with infusions commonly taking place in the patient's home.

Specialist metabolic nurses support families with metabolic disease by providing an interface between the hospital, clinic and the home and tailoring advice on medical care to the practical setting of the individual family.

Specialised metabolic dieticians play an important part in patient management. They draw up dietary regimens for well patients and for patients in crisis; give prescribing advice on special metabolic dietary products; monitor (biochemical / growth / tolerance) responses to dietary treatment with subsequent dietary adjustments and feedback to patients / professionals / carers; and manage patient pregnancies e.g. patients with phenylketonuria where the foetus is at risk.

In-patient care includes planned admissions for investigations, initiation of and ongoing treatment, and routine assessments as well as emergency admissions for patients with acute metabolic decompensation.

For most children shared care is provided by the local hospital acting in conjunction with the specialist metabolic disorders centre, but a small number of patients will need to be admitted for most or all their care to the specialist metabolic disorders centre. It is usual for children to require continuing supervision at the centre throughout their childhood. Although many disorders are life limiting, newer treatments have resulted in improved survival into adulthood, necessitating an expansion of adult metabolic disorders facilities and appropriate transfer arrangements.

The specialist metabolic disorders centre's multi-disciplinary team liaise with and support other health professionals, including primary and secondary care colleagues, regarding the ongoing care of patients with metabolic disorders. This includes access to appropriate facilities, including respite, and the planning of appropriate end-of-life care.

## **4.3 Lysosomal storage disorders service for adults and children**

Lysosomal storage disorders (LSDs) are a group of rare genetic conditions, characterised by specific lysosomal enzyme deficiencies. The main LSDs are: Gaucher's disease, Anderson-Fabry disease, Mucopolysaccharidosis Type I (Hurler's disease), Mucopolysaccharidosis Type II (Hunter's disease), Mucopolysaccharidosis Type VI (Maroteaux Lamy's disease) and Pompe's disease.

The service has been commissioned since April 2005 by the National Commissioning Group on behalf of English residents. There are four nationally designated centres for children and four nationally designated centres for adults. The nationally commissioned service includes:

- diagnosis
- assessment
- treatment including enzyme replacement therapies and substrate reduction therapies (the infusions are often given at home).

Much of the care of patients with LSDs remains locally provided and funded, for example, bone marrow transplantation services for patients with Hurler's disease.

#### **4.4 Rare mitochondrial disease service for adults and children**

Mitochondria are small organelles, present in every cell in the body, whose function is to process the cell's energy. Mitochondrial disease can therefore affect any cell process. It commonly presents as muscle weakness but any system (including liver, pancreas and kidney) can be affected, and the range of neurological symptoms is very wide.

This service has been commissioned since April 2007 by the National Commissioning Group on behalf of English residents. There are three nationally designated centres. The nationally commissioned service includes:

- a diagnostic service for children and adults with suspected respiratory chain disease in whom a diagnosis cannot be established using standard genetic tests available at regional genetics centres
- a clinical service consisting of genetic counselling and clinical management (out-patients and in-patients) for adults and children with mitochondrial respiratory chain disease
- clinical management advice for clinicians involved with patients with mitochondrial respiratory chain disease.

#### **4.5 Barth syndrome service for adults and children**

Barth syndrome is an x-linked disorder of lipid metabolism presenting as cardiac/skeletal myopathy, neutropenia and growth retardation with a high infant mortality rate. Patients with Barth syndrome present with frequent cardiac problems and, in two-thirds of patients, neutropenia (reduced white blood cell count leading to susceptibility to infection).

A service for Barth syndrome has been nationally commissioned by the National Commissioning Group since April 2010 on behalf of English residents. There is a single nationally designated centre for children and adults. The nationally commissioned out-patient service includes:

- diagnosis
- management advice
- annual monitoring
- lifelong follow-up.

It does not include the treatment of associated disease.

#### **4.6 Population screening programmes**

The specialised metabolic disorders service supports the national newborn screening programme for metabolic disorders; currently tests are carried out on all newborns for phenylketonuria (PKU) and medium chain acyl-coA dehydrogenase deficiency (MCADD). All newborns with positive initial screening tests need urgent clinical advice and support and rapid follow-up with diagnostic tests carried out by a metabolic laboratory. The patient and

family are then seen by the clinical team at a specialist metabolic disorders centre, or by a general paediatrician with advice from the specialist centre.

## **5. Identifying and costing activity**

### **5.1 Possible currencies:**

- out-patient attendances
- out-patient procedures
- non face to face out-patient attendances
- day cases
- in-patients
- HRG codes.

### **5.2 Existing classification systems**

- ICD-10 codes

See 'Metabolic ICD' worksheet in 'SSNDS Definition No. 36 Specialised Metabolic Disorders (all ages) website codes' spreadsheet.

Note: The ICD-10 diagnostic codes listed in this worksheet attempt to identify metabolic conditions requiring a specialised metabolic disorder service. However where shared care is being provided by a local hospital in conjunction with the specialist centre, both the specialist centre and the local hospital may use the same diagnostic codes. Hence the codes listed in the worksheet should not be solely used to identify *specialised* metabolic disorders service activity.

Note: see also the first bullet pointing in 5.4 below.

- OPCS-4.5 intervention codes

Note: There appear to be no OPCS-4.5 intervention codes which specifically identify specialised metabolic disorders service activity.

### **5.3 Costing activity**

*Please refer to the latest Department of Health Guidance on Payment by Results for up to date information on national tariffs and activity included / excluded from tariff.*

*Please note that not all the Payment by Results inclusions and exclusions listed below are specialised activity, but they are included here for completeness.*

(i) Is in scope of 2010/2011 Payment by Results and has a national tariff:

- endocrinology first/follow-up and single/multi-professional:multi-disciplinary out-patients attendances (Treatment Function Codes: 252 and 302) - MANDATORY tariff
- non face to face out-patient attendances (for TFCs that have a mandatory tariff for face-to-face out-patient attendances) - NON MANDATORY tariff
- out-patient procedures - MANDATORY tariff for 49 procedures only
- admitted patient care -
  - o MANDATORY combined tariff for day case and ordinary elective in-patient spells
  - o MANDATORY separate tariff for 17 day case and for 18 elective in-patient spells
  - o MANDATORY tariff for ordinary non-elective spells

(ii) Is excluded from 2010/2011 Payment by Results and therefore requires a locally negotiated tariff:

- services -
  - o nationally commissioned services
- out-patient attendances -
  - o paediatric metabolic disease out-patient attendances (Treatment Function Code: 261)
- admitted patient care - see list of specific exclusions
- drugs - see list of specific exclusions

#### **5.4 Outstanding issues raised regarding currencies and classifications systems**

It should be noted that ICD-10 is now wholly inadequate to classify metabolic disorders. Many metabolic conditions, including those requiring specialist care, do not have an individual ICD-10 code and the structure of the current ICD-10 classification system is too simple for current use. A more comprehensive classification system (Microsoft Access file) is available on [http://www.ssiem.org/webresources\\_inborn.asp](http://www.ssiem.org/webresources_inborn.asp)

There are no OPCS-4.5 intervention codes.

### **6. National standards and guidelines**

#### **Available from the Department of Health - [www.dh.gov.uk](http://www.dh.gov.uk)**

- Department of Health (2003) 'Our inheritance, our future: realising the potential of genetics in the NHS'

#### **Available from the National Screening Committee - [www.newbornbloodspot.screening.nhs.uk](http://www.newbornbloodspot.screening.nhs.uk)**

- National Screening Committee (2008) 'Standards and guidelines for newborn bloodspot screening'

#### **Available from PHG Foundation - [www.phgfoundation.org](http://www.phgfoundation.org)**

- Burton H (2005) 'Metabolic pathways, networks of care' Public Health Genetics Unit

#### **Available from the National Metabolic Biochemistry Network - [www.metbio.net](http://www.metbio.net)**

- various diagnostic guidelines

#### **Available from the British Inherited Metabolic Diseases Group – [www.bimdg.org.uk](http://www.bimdg.org.uk)**

- various management guidelines

**Clinical Pathology Accreditation** for clinical chemistry and neonatal screening laboratories currently covers the metabolic biochemistry laboratories as part of the clinical chemistry inspection process.

**Quality assurance** is provided by the European provider ERNDIM with some schemes covered by NEQAS SAC and arranged through Metbionet.

**Endorsement**

British Inherited Metabolic Diseases Group  
Genetics Commissioning Advisory Group (GenCAG)  
Genetics Interest Group