

E06/S(HSS)/d

**2013/14 NHS STANDARD CONTRACT  
FOR MCARDLE'S DISEASE SERVICE (CHILDREN)**

**PARTICULARS, SCHEDULE 2 – THE SERVICES, A - SERVICE  
SPECIFICATIONS**

<b>Service Specification No.</b>	E06/S(HSS)/d
<b>Service</b>	McArdle's disease service (Children)
<b>Commissioner Lead</b>	
<b>Provider Lead</b>	
<b>Period</b>	12 months
<b>Date of Review</b>	

**1. Population Needs**

**1.1 National/local context and evidence base**

Data on the epidemiology of McArdle disease are limited, with few published epidemiological studies. A study from the Netherlands looked at the frequency amongst newborns of the most common genetic mutation associated with McArdle disease in northern Europeans (van Elfen et al, 2002), estimating the prevalence of McArdle disease to be 3 per million. Extrapolating this to the population of England produces a figure of 150 cases nationally.

A considerably higher prevalence was estimated by Haller in Dallas, USA, at 1 per 100,000 population (Haller, 2000), which would equate to an English prevalence of 500.

An additional prevalence estimate comes from an international workshop held in 2006, at which 290 cases of McArdle disease were identified as being known to services in Dallas USA, UK, France, Spain, Italy, Germany and Denmark (Quinlivan & Vissing, 2007).

**References:**

- Haller R.G., 2000. Treatment of McArdle disease. *Archives of Neurology*, 57, pp.923-924.
- Quinlivan R., Vissing J., 2007. The 144<sup>th</sup> ENMC workshop: Outcome measures in McArdle Disease. *Neuromuscular Disorders*, 17, pp.494-498.

- Van Elfen N., de Bie H.J.J., Wevers R.A., Arenas J., van Engelen B.G.M., 2002. The prevalence and genetic characteristics of McArdle's disease in The Netherlands. *Neuromuscular Disorders*, 12, p. 752.

McArdle disease is a rare condition with only a few interested physicians worldwide seeing significant numbers of patients. The largest cohort of patients to participate in a randomised controlled trial is 19, and, as such, the evidence base for effective treatments is limited.

A Cochrane systematic review of the literature (Quinlivan et al, 2008) reviewed the outcomes of randomised controlled trials of D-Ribose, Glucagon, Verapamil, Vitamin B6, high protein diet, oral branched chain amino acid supplementation and Dantrolene, none of which demonstrated benefit.

Creatinine supplementation has been shown to benefit exercise capacity of healthy individuals undergoing resistive training (Vandenberghe et al, 1997) and to increase strength in mitochondrial myopathies (Tarnopolsky et al, 1997). A randomised placebo controlled cross-over trial of creatine 60mg/kg/day versus placebo for five weeks in nine McArdle subjects demonstrated subjective improvement in five out of nine subjects (Vorgerd et al, 2000). In a follow up study, however, symptoms in 19 McArdle patients worsened when 150mg/kg of creatine was given daily (Vorgerd et al, 2002).

Haller and Vissing (Haller & Vissing, 2002) demonstrated a 20% improvement in oxidative capacity when intravenous glucose was given during exercise. Oral sucrose 75g has been compared with placebo 30-40 minutes before fixed intensity exercise on a cycle ergometer (Vissing & Haller, 2003), measuring heart rate, work load and RPE (rating of perceived exertion), together with biochemical measures such as glucose, lactate, pyruvate, ammonia, insulin and free fatty acids. Oral sucrose was significantly better than placebo in improving exercise performance. In a study by Vissing (Quinlivan & Vissing, 2007) a high carbohydrate diet given over three days improved exercise performance when compared with a high protein diet given for the same duration.

Aerobic training was studied in eight McArdle subjects (Haller et al, 2006). Each was requested to train for 30-40 minutes to 60-70% maximal heart rate four times each week. The study demonstrated that aerobic training, at less than 70% maximum heart rate, is safe in McArdle disease and increases work capacity without worsening symptoms of fatigue, cramping and pain.

Portero (In Quinlivan & Vissing, 2007) studied four patients and five controls during eight weeks of regular training undertaken three times a week for 30-45 minutes. The results demonstrated a slight decrease in creatine kinase (CK), reduced body fat and an increased feeling of well being in the subjects. Aerobic training has been shown to increase the amplitude of the second wind (Quinlivan & Vissing, 2007).

Data from repeated 12 minute walking assessments of patients has demonstrated an improvement in muscle performance following instruction in appropriate lifestyle changes and exercise, with the greatest benefit experienced by the most severely affected patients (Quinlivan et al, 2007).

## **2. Scope**

### **2.1 Aims and objectives of service**

#### **Definition of Service:**

A diagnostic and management service for people with McArdle disease and rare glycogenolytic disorders which will be predominantly outpatient based and led by a multi-disciplinary team.

#### **Aims**

The National Diagnostic and Management Centre for McArdle Disease aims to provide equitable access to diagnostic and multi-disciplinary management services for patients with McArdle disease, and related disorders which are extremely rare conditions and include: Phosphofructokinase deficiency, the muscle form of phosphorylase B kinase deficiency, Phosphoglycerate mutase deficiency, phosphoglycerate kinase deficiency, enolase deficiency and lactate dehydrogenase deficiency. The largest group of disorders seen will be people with McArdle disease. The service will provide high quality diagnostics and care for this small patient group, to enable effective control of symptoms and improved quality of life.

#### **General overview**

McArdle disease is a rare, genetic, neuromuscular disorder associated with muscle cramps and injury, and myoglobinuria induced by sudden, vigorous exercise. The National Diagnostic and Management Centre for McArdle Disease provides clinical assessment and diagnostics for patients with, or suspected of having, this disease, in addition to some closely related disorders, and provides on-going management for patients with confirmed diagnoses.

Diagnosis of McArdle disease has been problematic. Patients with McArdle disease often experience considerable delay, not infrequently up to 20 years, in obtaining an accurate diagnosis. Misdiagnosis is also reported, and it is estimated 18% of patients previously receiving a diagnosis of McArdle disease have been misdiagnosed, subsequent diagnoses including Becker muscular dystrophy, mitochondrial myopathy, congenital myopathy and chronic fatigue syndrome. The National Diagnostic and Management Centre for McArdle Disease aims to address these problems.

Ensuring correct diagnosis of McArdle disease similarly enables appropriate genetic

counselling and health monitoring and management for these patients. Prevention of life-threatening complications such as acute rhabdomyolysis, through timely diagnosis and appropriate management of the disease, has obvious health benefits, but also wider benefits to the healthcare economy, reducing the need for critical care admissions. Appropriate management can reduce obesity and its complications, improve fitness levels with the inherent health benefits, improve quality of life and increase participation in society, especially in terms of employment by teaching patients strategies to manage their condition and reduce the rate of associated disability. Early diagnosis may additionally improve psychosocial outcomes for patients.

Inappropriate diagnoses of McArdle disease can also prevent appropriate management, such as failure to monitor for serious cardiac complications associated with Becker muscular dystrophy but not seen in McArdle disease.

### **Objectives:**

- To provide a timely and accurate diagnosis for patients presenting with symptoms suggestive of McArdle disease;
- to assess new patients with an established diagnosis of McArdle disease to confirm the diagnosis;
- to ensure appropriate utilisation of muscle biochemistry and gene sequencing, which are relatively expensive and accessed directly by clinicians outside the national service for diagnosis of McArdle disease;
- to provide instruction for patients to enable effective self-care and disease management;
- to manage associated disorders and complications experienced by patients including chronic fatigue syndrome;
- to provide high quality written information for patients about McArdle disease and its treatment;
- to monitor patients with a confirmed diagnosis of McArdle disease clinically, biochemically and functionally to ensure effectiveness of treatment;
- to provide advice to patients' local healthcare providers to enable local management of patients where appropriate;
- to involve patients and patient groups in service planning to ensure a high quality, appropriate and responsive service;
- to develop the knowledge and skills of the multi-disciplinary team (MDT) to ensure sustainable provision of a high quality service;
- to increase awareness of McArdle disease and the national service amongst healthcare professionals and the public.

## **2.2 Service description/care pathway**

McArdle disease, also known as glycogen storage disorder type V, is a rare metabolic muscle disorder characterised by a deficiency of the enzyme muscle phosphorylase and resulting from an autosomal recessively inherited genetic mutation.

During anaerobic exercise, muscle phosphorylase converts muscle glycogen stores

to glucose which acts as a source of energy for muscle activity. The deficiency of muscle phosphorylase in McArdle disease results in an inability to release this energy, producing the typical symptoms of skeletal muscle pain and fatigue, breathlessness, palpitations, dizziness and fatigue. Continued exercise without resting can result in the development of muscle contractures, with severe, painful, rigid paralysis lasting for hours and associated with muscle swelling and damage.

This muscle damage may result in myoglobinuria, producing a coca cola discolouration of the urine, and can be associated with rhabdomyolysis, a potentially life-threatening condition associated with collapse and acute renal failure, in more severe cases necessitating admission to intensive care units and a period of renal dialysis. Premature death is reported in individuals with McArdle disease as a result of accidents resulting from muscle weakness, such as whilst swimming or climbing.

The more long-term effects of the muscle contractures associated with McArdle disease include permanent muscle weakness, and adoption of a sedentary lifestyle which may result in incapacitation with chronic, non-exercise related pain and fatigue, overweight and obesity (affecting 72% of McArdle patients), and a poor quality of life with poor participation in society. Excess body weight increases the tendency for anaerobic exercise and exacerbates exercise intolerance.

Key to the management of McArdle disease is the concept of the 'second wind'. If patients rest or reduce the intensity of exercise after the initial painful, cramping period, alternative energy sources can be utilised by muscles resulting in reduced pain and fatigue, enabling continued exercise. There is a wide variation in each individual's capacity to experience this second wind, the efficiency of which determines the severity of the condition. With appropriate clinical management, however, quality of life, exercise intolerance and muscle strength can be optimised. In addition to the physical effects of McArdle disease, anxiety and depression are common; 31% of people attending the McArdle clinic had previously been seen in NHS psychiatric services, compared with the figure of 25%, quoted by Mind, for the general population seen in NHS psychiatric services. Potentially incapacitating chronic fatigue syndrome is also common, estimated to affect 40% of McArdle patients, and resulting in severely restricted physical activity. The cause is likely to be multi-factorial and may include a secondary impairment of fatty acid oxidation (exacerbated by a sedentary life-style), psychosocial factors (delayed diagnosis and previous episodes of collapse secondary to rhabdomyolysis) and poor pain coping strategies.

Other disorders caused by glyco(genolytic) enzyme deficiencies are extremely rare, and include phosphofructokinase deficiency and the pure muscle form of phosphorylase B kinase deficiency. The presentation and management of these conditions are similar to McArdle disease.

The service comprises three levels:

1. Direct access to diagnostic investigations, including metabolic analysis of muscle tissue and/or deoxyribonucleic acid (DNA) testing.
2. Clinical diagnostic assessment and investigation of patients presenting with symptoms that could be caused by McArdle disease or a related disorder.
3. Management of patients with a confirmed diagnosis of McArdle disease, phosphofructokinase deficiency and the pure muscle form of phosphorylase B kinase deficiency.

### **Diagnostic service**

Diagnostic investigations provided for new patients referred to the service include (dependent upon clinical need):

- routine biochemistry including serum CK (creatine kinase), urate, carnitine and acyl carnitine;
- screening DNA blood test to look for hot spot mutations p.Arg50X and p.Gly205Ser;
  - It is important to confirm the diagnosis by DNA analysis to ensure accuracy of diagnosis, enabling appropriate genetic counselling and disease management.
  - This analysis is offered by The Metabolic Laboratory Birmingham Children's Hospital (BCH) and by Sheffield Children's Foundation Trust.
- electromyography (EMG) for patients for whom the diagnosis is not confirmed to rule out other disorders
- muscle biopsy is performed for histochemistry and quantitative biochemical analysis to exclude other rare but similar metabolic myopathies affecting the glycol(geno)lytic pathway (such as phosphorylase B kinase deficiency and Tarui's disease) and other rare muscle disorders especially Becker muscular dystrophy, if the diagnosis of McArdle disease is not confirmed. Skin biopsy is performed at the same time and cryopreserved or cultured in case the diagnosis of McArdle disease is subsequently not confirmed:

### **Types of Diagnostic tests**

Microscopy and other histochemical studies are performed on muscle biopsy tissue for histochemical stains for glycogen, phosphorylase and phosphofructokinase and to rule out other muscle disorders. This analysis will be performed at The National Hospital, Queen Square, UCLH.

In selected cases, muscle biopsy tissue is analysed biochemically for most of the enzymes in the glycogenolytic and glycolytic pathways, and a quantitative assay of muscle stored glycogen and investigation of glycogen structure is undertaken. This analysis is performed by the enzyme laboratory Great Ormond Street Hospital for Children NHS Trust (GOSH).

A forearm exercise test for selected patients; performed if a false positive diagnosis of McArdle disease is suspected prior to subjecting the patient to a repeat muscle biopsy or if a novel glyo(geno)lytic disorder affecting another enzyme is suspected (a rare example might be phosphoglycerate kinase deficiency)

full gene sequencing where appropriate if the diagnosis of McArdle disease is suggested by muscle histochemistry and/or biochemistry, but a hot spot mutation in the myophosphorylase gene (*PYGM*) is not identified or present only on one of the two alleles, full gene sequencing is performed to identify the mutation this will be undertaken by Sheffield Molecular Genetics Service (SMGS), Sheffield Children's NHS Foundation Trust, Sheffield all laboratories undertaking these diagnostic tests are CPA accredited the specialist diagnostic investigations provided by the service are directly accessible in isolation or as part of a complete package of specialist multi-disciplinary assessment and management.

The lead physician provides a gate-keeping mechanism for direct access to muscle biochemistry and gene sequencing testing by means of review of patient history. This ensures appropriate utilisation of tests and resources. This service is available to clinicians in England and by agreement with the rest of the United Kingdom.

### **Clinical service**

The clinical service is provided at the National Hospital for Neurology and Neurosurgery at Queens Square and Great Ormond Street Hospital. The multi-disciplinary clinic team includes consultant, sports scientist, physiotherapist, dietician, clinical psychologist and nurse specialist.

Patients referred with symptoms suggestive of McArdle disease or patients with a confirmed diagnosis of McArdle disease and related disorders are referred to the clinical service, consisting of two weekly clinics at the National Hospital for Neurology and Neurosurgery.

The consultant will participating in a weekly clinic at GOSH, new referrals with a suspected diagnosis will be seen in the clinic and investigations will be arranged. Children with a confirmed diagnosis requiring follow-up and management will be seen in a designated follow-up clinic at GOSH, the number of designated clinics will be dependent upon the number of confirmed patients. Initially, this may be one or two clinics each year to begin with, however, this is expected to increase over time as we hope that a greater awareness of the condition will lead to earlier referral patterns.

One weekly clinic focuses on diagnosis, seeing new, as yet undiagnosed, patients. Dependent upon disease type and the complexity of investigation required patients require between one and three diagnostic clinic attendances. It is anticipated that the numbers of new referrals to the clinic will increase over the first three years of the service being in place as awareness of the service increases nationally.

Patients attending the diagnostic clinic undergo clinical assessment and assessment of exercise capacity through a 12 minute walk test adapted for people with McArdle disease. This test gives a good indication of the ability of patients to cope with ADLs (activities of daily living). Cycle ergometry with oxygen consumption to assess oxygen consumption ( $VO_2$  Max), is also undertaken to assess aerobic capacity and

can be used to prescribe a personalised exercise plan. The previously described diagnostic investigations are initiated as required. If muscle biopsy is required this will be performed as a day case admission.

The second clinic focuses on disease management, and sees follow-up or previously diagnosed patients. There is a wide range of clinical severity amongst people suffering with McArdle disease, and the frequency of follow-up is tailored to clinical need, varying between six monthly for the most severely affected (achieving less than 250m during a 12 minute walking test) and 1-2 yearly for less severely affected (able to walk between 250 and 800m in a 12 minute walk test). Patients may be seen for monitoring by the physiotherapist in between appointments. Patients with episodes of rhabdomyolysis requiring renal dialysis or intensive care will be seen more frequently as required in order to try and prevent recurrence.

Attendance at disease management clinics enables review of care and progress. Biochemical monitoring includes serum CK to provide an early warning with regard to lifestyle. For example, if on one occasion the CK level is uncharacteristically high a review of recent activities may provide useful feedback to the patient which might reduce subsequent episodes of rhabdomyolysis. Exercise assessment enables monitoring of functional improvement. An association between McArdle disease and chronic fatigue syndrome has been demonstrated and for affected patients advice is provided to improve sleep hygiene and pacing, in addition to cognitive behavioural therapy. Psychology sessions are offered on the same day as the follow-up clinic appointments.

Management of patients with phosphofructokinase deficiency and the pure muscle form of phosphorylase B kinase deficiency is shared with the metabolic diseases clinic.

Patients who improve significantly are discharged back to their GP or local muscle clinic once they are able manage every day activities with little or no disruption. Some of these patients may require referral back to the service if symptoms worsen, usually due to stressful life events that cause the individual to lose motivation for the life-style changes necessary for improvement.

In addition to clinics, the service includes regular multi-disciplinary team meetings and the provision of on-going telephone and written advice and support for patients and clinicians.

## **Roles of individual multi-disciplinary team members**

### **Lead Physician**

The role of the lead physician includes:

- consultation of suspected and newly diagnosed patients in the diagnostic and management clinics;
- liaison with patients' local services;
- gate-keeping role for direct access to muscle biochemistry and gene sequencing



testing to ensure appropriate use of resources through review of patient history;

- review of results undertaken on patients seen by the service, including joint review of muscle biopsies with the pathologist;
- where clinicians have sent only tissue samples for diagnosis, writing to them with results and offering appropriate advice;
- liaison with patient support group;
- liaison and collaboration with the collaborating centres in Birmingham and Sheffield;
- overall co-ordination of the service;
- leading multi-disciplinary team meetings and audit;
- ensuring continuing professional development.

### **Specialist Physiotherapist**

The role of the physiotherapist includes:

- attendance at diagnostic and management clinics;
- assessment of patients including measurement and documentation of muscle strength, half day assessments to assess patient's aerobic capacity using a standardised cycle ergometry test;
- provision of advice to patients on the relative aspects of management of McArdle disease, including on posture;
- liaison with patients' local physiotherapy teams;
- provision of regular contact with patients by telephone or e-mail to reinforce their management plan and ensure compliance;
- attendance at multi-disciplinary team meetings;
- participation in audit and continuing professional development.

### **Dietician**

The role of the dietician includes:

- attendance at management, but not diagnostic, clinics;
- provision of dietary advice to patients including guidance on weight management, adequate protein intake to maintain muscle bulk (i.e. prevent catabolism), the potential benefits of a carbohydrate rich diet (Vissing, in Quinlivan and Vissing 2007), trial of low dose creatine supplementation (Vorgerd 2000) and the benefit of sucrose prior to planned exercise (Vissing 2003).

### **Sports Scientist**

The Sports Scientist's roles in include:

- attendance at management clinics;
- supervision of functional exercise assessments undertaken by the physiotherapist in diagnostic clinics;
- provision of advice to patients on exercise capability based on results of cycle ergometry assessments.

## **Nurse specialist**

The specialist nurse role in the service includes:

- attendance at both diagnostic and management clinics;
- provision of information and support to patients;
- provision of on-going support to patients, especially those new patients suffering with acute anxiety following an episode of rhabdomyolysis;
- reinforcement of information and advice regarding energy pacing and sleep hygiene programme for chronic fatigue syndrome (CFS/ME) symptoms;
- liaison with the patient support group, including to provide a support group meeting within the clinic session;
- liaison with schools in the case of paediatric patients to provide advice and support.

## **Clinical Psychologist**

The role of the clinical psychologist in the service includes:

- screening for psychological issues such as anxiety, depression and chronic fatigue syndrome;
- providing cognitive behavioural therapy for those patients suffering from anxiety and depression, chronic pain and chronic fatigue syndrome;
- referral to, and liaison with, local psychology services for on-going treatment if required;
- leading on assessment of quality of life.

## **Service Co-ordinator**

The service co-ordinator's role in the service includes:

- provision of secretarial and organisational support for the service;
- maintenance of records of all samples and patients referred on a secure database.

## **Clinical Scientist, Biochemistry**

The clinical scientist, biochemistry role in the service includes involvement in:

- quantitative biochemical analysis of glyco(geno)lytic enzymes;
- analysis of glycogen structure and content of muscle tissue.

## **Clinical Scientist, Muscle Pathology 0.10 WTE**

The clinical scientist, muscle pathology role includes interpretation of muscle biopsy samples and slides from muscle biopsies undertaken elsewhere.

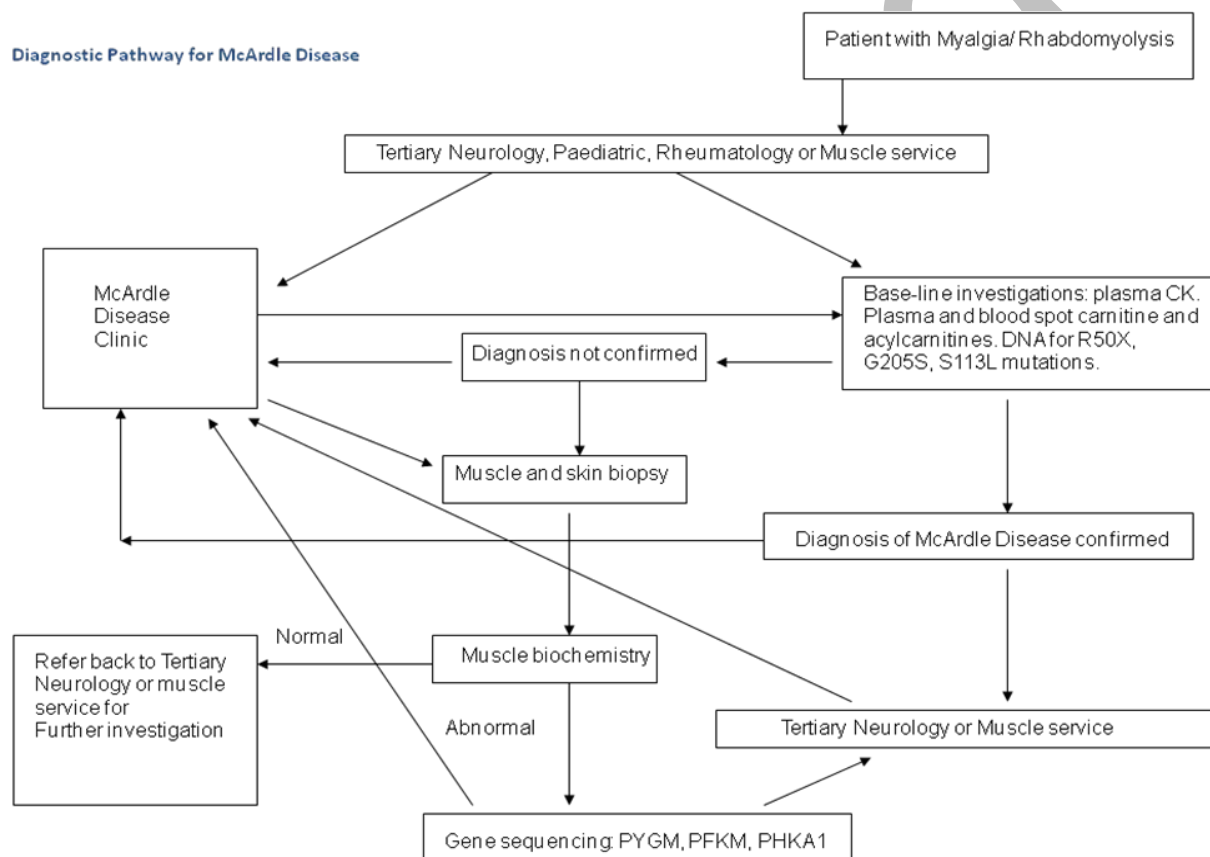
## **Service model and care pathways**

As described, the service comprises of three levels:

- Direct access to diagnostic investigations;
- clinical diagnostic assessment and investigation of patients presenting with symptoms that could be caused by McArdle disease or a related disorder;
- management and advice for patients with a confirmed diagnosis of McArdle disease, phosphofructokinase deficiency and the pure muscle form of phosphorylase B kinase deficiency.

The service will operate during office hours.

The diagnostic pathway and routes into and out of the service are described in the flow diagram below:



## 2.3 Population covered

The service is commissioned by NHS England for the residents of England and Scotland. Patients from the member states of the European Union can access the service via reciprocal agreements. Patients from Wales and Northern Ireland are not part of this commissioned service and the trust must have separate arrangements in place for patients from these and other non EU referrers

## **2.4 Any acceptance and exclusion criteria**

The McArdle service is commissioned on behalf of the NHS in England and Scotland and so the service will accept referrals for all residents of England and Scotland. The service will be made available to all patients by ensuring awareness of the national service amongst healthcare professionals and informing patients about the service through the Association for Glycogen Storage Disease (AGSD) website.

Patients from Wales and Northern Ireland are not part of this commissioned service and the trust must have separate arrangements in place for patients from these and other non EU referrers.

The provider has a duty to co-operate with the commissioner in undertaking Equality Impact Assessments as a requirement of race, gender, sexual orientation, religion and disability equality legislation.

### **Referral criteria, sources and routes**

The service sees and investigates patients who have been diagnosed with, or who are suspected of having, McArdle disease, phosphofructokinase deficiency or the pure muscle form of phosphorylase B kinase deficiency. Referrals are accepted from tertiary neurology, paediatric, rheumatology or muscle services.

All referrals will be reviewed by the lead clinician. The lead physician in the service will also provide a gatekeeping role for some diagnostic services; requests for muscle biochemistry and gene sequencing testing for McArdle disease out with the national service will be reviewed by the lead clinician in conjunction with review of patient history to ensure the appropriateness of testing.

### **Exclusion criteria**

Patients are excluded from the service if:

- they are referred for the treatment or investigation of neuromuscular conditions other than McArdle disease, phosphofructokinase deficiency and the pure muscle form of phosphorylase B kinase deficiency. Patients with Pompe disease and other Glycogen Storage disorders are the remit of metabolic services and provision for these patients is not included in the service;
- their referral suggests that a diagnosis of McArdle disease is unlikely.

Patients who, following investigation, are found not to have McArdle disease are discharged back to the care of the referring service.

Those patients who are diagnosed with McArdle disease but who achieve more than or equal to 800m on a 12 minute walking test and who are not experiencing episodes of myoglobinuria are also discharged from the service.

Most patients accessing the service are adults (approximately 90%). The small proportion of children accessing the service are seen at GOSH by the same clinical team. Children requiring on-going monitoring and management by the service are transferred to the main adult service at the age of 16 years. Transition arrangements will be in place such that the transfer of patients from the paediatric to adult service will be done smoothly with the patient at the centre of the care provided.

## **2.5 Interdependencies with other services**

Internally the multi-disciplinary team (MDT) links into multiple clinical and administrative teams as a result of the broad composition of the team. Strong links are also required between the clinical and diagnostic teams involved in the service.

The provider will link with patients' local healthcare providers to ensure provision of high quality, integrated care, in addition to liaison with patients' schools, employers etc. to provide support and advice.

The service promotes increased awareness of McArdle disease and the national service amongst healthcare professionals and also the public. This includes close working with Association for Glycogen Storage Disease (AGSD).

There are no national/clinical networks/expert patient programmes and screening programmes applicable to the service

### **Sub-contractors**

Enzyme Laboratory, Great Ormond Street Hospital, London  
Sheffield Molecular Genetics Service, Sheffield Children's NHS Foundation Trust, Sheffield  
Department of Clinical Biochemistry, Sheffield Children's NHS Foundation Trust, Sheffield  
Dubowitz Neuromuscular Centre, Great Ormond Street Hospital, London

## **3. Applicable Service Standards**

### **3.1 Applicable national standards e.g. NICE, Royal College**

The service sits within the trust's clinical governance and risk management arrangements.

Governance arrangements will include:

- a weekly MDT meeting with the clinic team;
- a monthly muscle biopsy MDT;
- a six monthly MDT meeting with the DNA laboratory in Birmingham;
- a monthly metabolic muscle MDT meeting;

- a six monthly meeting with the DNA laboratory in Sheffield;
- annual service audit;
- membership of key team members of the British Society of Myology and attendance at national and international muscle conferences;
- incident reporting and annual appraisal procedures and policies.

Risk management procedures including:

- monitoring of patients for muscle cramps and myoglobinuria
- provision of a standardised letter for patients to advise local services on how to manage rhabdomyolysis
- open access to the nearest nephrology unit for any patient presenting to the clinic with myoglobinuria.

Education, training and development of team members.

The provider must inform NHS England of any formal complaints made by patients that are processed within the Trust's complaints procedure.

The provider must inform NHS England of any serious untoward incidents (SUIs) that occur as part of the operation of the McArdle Service. The provider must also make

available to NHS England any evidence, reports etc. produced in the course of investigating the incident.

Both the commissioner and provider have a commitment to working together to continually improve the service and react to innovative and dynamic ideas. The provider is required to demonstrate continual improvement in patient care and service delivery. This process will be informed by clinical and service audit, patient and public engagement and awareness of national and international clinical and policy developments that could inform service development.

The service provides patients with written information about McArdle disease, which is also available at <http://www.muscular-dystrophy.org>.

Patients are signposted to additional sources of information and support, including the GSDnet, an e-mail mailing list system enabling communication between people with all types of glycogen storage disease, and the McArdle disease support group at the AGSD.

With patient permission the service liaises with schools, universities and employers to provide information and support as required.

A representative from the AGSD attends clinics to facilitate a support network for patients. At the end of each clinic a brief meeting is held with the lead clinician and the AGSD representative to discuss service development and to feedback any issues regarding the service. This feedback process has already resulted in improvements in the service, including enhanced communication with patients and an improved system for co-ordinating multi-disciplinary assessments to reduce waiting times.

#### 4. Key Service Outcomes

The providers will provide agreed performance monitoring data on a yearly basis in the form of an annual report. Where any elements of this deviate from the agreed plan, the service will provide a brief explanation accompanying the submission of the report.

The commissioner may wish to follow this up and request further information to inform any necessary actions that will be agreed between the service and commissioners in the context of the terms and conditions of the agreement.

Three groups of key outcome measures are used to evaluate the quality of the service:

**Clinical outcomes** measured for patients attending the multi-disciplinary clinic:

- functional capacity, assessed with a 12 minute walking test;
- muscle strength
- biochemical parameters, specifically serum creatine kinase (CK), urinary myoglobin and plasma urate
- frequency of rhabdomyolysis, ascertained through patient questioning
- patient perception of improvement as a consequence of attending the clinical service, assessed by means of a self-completed questionnaire
- a quality of life measure such as short form (36) health survey (SF36).

These clinical outcomes are measured at the time of clinical visits.

**Patient satisfaction**- audited through patient questionnaires.

#### **Diagnostic services**

- satisfaction with the diagnostic services will be assessed through a feedback questionnaire sent to users of the muscle biochemistry, hot spot mutation analysis and full gene sequencing services
- 80% of results sent for full gene sequencing for phosphorylase, glycogen, muscle (PYGM) expected to demonstrate a positive diagnosis.

#### **Bench marks for McArdle service**

Frequency of symptoms in 80 new patients seen for the first time:

1. muscle pain with exercise 100%
2. second wind 85%
3. acute renal failure (unplanned admission 10%)
4. muscle weakness 20%
5. overweight or obese 72%
6. non-exercise chronic pain and fatigue 42%

7. depression and anxiety 37%
8. 12g
9. diagnosis for 80% McArdle patients.

### **Increasing professional awareness of the service**

Professional and public awareness of the service will be promoted through communications via the UK Muscle Interest Group and the British Myology Society and through websites and magazines produced by the Association for Glycogen Storage Disease (AGSD) UK and the Muscular Dystrophy Campaign. In addition, national and international journal publications and conference presentations and lectures will increase professional awareness of the disease and the service.

The provider will work with NHS England to ensure sufficient considerations are given to communications.

### **5. Location of Provider Premises**

The National Hospital for Neurology and Neurosurgery  
University College London Hospitals' NHS Foundation Trust  
*Dubowitz Neuromuscular Centre, Great Ormond Street Hospital*

Adopted