

E13/S(HSS)/d

**2013/14 NHS STANDARD CONTRACT
FOR DIAGNOSTIC SERVICE FOR PRIMARY DYSKINESIA
(ALL AGES)**

SECTION B PART 1 - SERVICE SPECIFICATIONS

Service Specification No.	E13/S(HSS)/d
Service	Diagnostic service for primary dyskinesia (All Ages)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

1.1 National/local context and evidence base

Background:

Primary ciliary dyskinesia (PCD) is inherited as an autosomal recessive condition. The prevalence is unknown but it is probably between 1:26000 and 1:40,000 in the caucasian population (Kuehni ERJ 2010) Its incidence is very much higher in the Asian population (O’Callaghan Arc Dis Child 2009). It is caused by one of a number of different ciliary ultrastructural defects each of which may result in ineffective ciliary movement (Schidlow, 1994). In primary ciliary dyskinesia impaired mucociliary clearance is associated with recurrent chest infections, leading to bronchiectasis. It is also associated with serous otitis media (glue ear) chronic rhinitis and sinusitis. Symptoms frequently start in the neonatal period. Significant hearing problems occur in approximately 50% of children. Fifty percent will have situs inversus. The standards of care and indications for biopsy have been published (Barbato et al, 2009 and Bush, 2007).

Medical consequences of late or incorrect diagnosis:

Patients are frequently diagnosed after many years of chronic respiratory symptoms with established, and frequently very severe, lung disease (Ellerman & Bisgaard 1997; Coren et al, 2002). They have often undergone a battery of invasive diagnostic procedures in an attempt to delineate their underlying problem (Bush 2002), but have failed to be investigated for PCD.

Delayed diagnosis is primarily due to lack of appropriate diagnostic facilities to test for PCD and inaccessibility to tertiary diagnostic centres.

Early and accurate diagnosis of PCD is necessary for the following reasons:

- The vast majority of those with PCD have symptoms from birth (Greenstone et al, 1988) or early infancy. However, the diagnosis is often made at a very late stage, when significant, bronchiectasis and permanent lung damage has already occurred (Ellerman & Bisgaard, 1997). The life span of these patients will be reduced
- Early diagnosis and interventions can delay or prevent irreversible lung damage, offset developmental delay by appropriate management of hearing impairment and allow a better quality of life and potential to normal life span
- The diagnosis of primary ciliary dyskinesia is difficult. Although false positive diagnoses occur, the greatest problems are due to late diagnoses (Turner et al, 1981) and false negative diagnoses.

2. Scope

2.1 Aims and objectives of service

PCD is an abnormality of cilia on the surface of cells in the lung, nose and ear. The diagnostic service depends on full-time highly skilled technicians and scientists who take samples from nasal or bronchial brush biopsies. These epithelial cells are then analysed using light microscopy with high speed video technology to study the movement of the cilia. The samples are then looked at using electron microscopy to determine the ultrastructure of each individual cilia and scientists who analyse ciliary function from patient samples, perform detailed electron microscopy and cultured ciliated epithelium. They also perform nasal nitric oxide measurements. The diagnostic service depends on a team of health care professionals, scientists and technicians to assess the patients, and examine ciliary function and structure. Nasal and bronchial biopsies are analysed by high speed video microscopy and electron microscopy. Specialist cell culture techniques are used to differentiate primary and secondary ciliary defects. Only three centres in England (University Hospitals of Leicester NHS Trust, Royal Brompton & Harefield NHS Foundation Trust and Southampton University Hospital NHS Foundation Trust) can diagnose primary ciliary dyskinesia.

The condition causes chronic inflammation and infection in the lung leading to bronchiectasis. Patients also suffer from recurrent rhinitis, nasal obstruction and sinusitis and more than half have associated hearing problems requiring active management. Early diagnosis leads to appropriate treatment that will give a better quality of life." ear and sinus infection. This can lead to a progressive and irreversible form of lung damage; result in developmental delay due to hearing impairment, and significant debility from chronic sinusitis.

The aim of this service is:

- To provide a national referral service for the diagnosis of PCD. This will include a nasal, and where necessary a bronchoscopy, biopsy service at the referral

centres and full diagnostic assessment

- A courier service, in London to the Royal Brompton & Harefield NHS Foundation Trust and from major UK centres (e.g. Cambridge, Leeds, Liverpool, Bristol, Oxford and Newcastle) to University Hospitals of Leicester NHS Trust and Southampton University Hospital NHS Foundation Trust to support national coverage
- Outreach clinics to facilitate collection of samples, e.g. from Leicester to Manchester, Birmingham and Cambridge
- To establish standards of care for children and adults with primary ciliary dyskinesia and to provide education nationally on the diagnosis and management of PCD
- To use the national database to collect relevant diagnostic information (clinical & laboratory).

Objectives of the service:

- Improve awareness, diagnosis and management of PCD
- Provide a diagnostic service for PCD for adults and children throughout England
- Improve awareness, diagnosis and management of PCD within the UK by education and provision of highly specific and sensitive diagnostic testing alongside liaison with and education of local healthcare providers.

The purpose of the service will be to:

- Develop a unified diagnostic service across the three centres. To standardise methodologies for functional and structural assessment of cilia
- To undertake specialist cell culture techniques as developed by the three UK centres
- Develop a courier service to allow national coverage. Where required to establish outreach diagnostic clinics to improve diagnostic samples and allow counselling of patients of the disease by a specialist in PCD
- Ensure difficult cases are given provision for consultant PCD specialists to perform biopsies at referral hospitals and patients to travel to a referral centre for assessment. In a small number of cases bronchoscopic sampling is required
- Ensure referring teams are given support and advice on how to manage patients once back in their local setting
- Improve awareness, diagnosis and management of PCD within the UK by education and provision of highly specific and sensitive diagnostic testing
- Offer patient centred assessment and advice regarding the many organ specific problems associated with PCD
- Minimise impact on the patient and their family life, education and work practice
- Liaise with, and advise, healthcare workers in all relevant disciplines
- To enter data into the national PCD database with regards to clinical presenting symptoms and diagnostic testing.

2.2 Service description/care pathway

Discharge criteria & planning including any transition arrangements to adulthood

Patients are discharged following their final diagnosis appointment at which results are given and explained. Detailed letters are written to the referring doctor and to the patient these will include information on their advised ongoing management (to be provided locally)

Days/hours of operation

The diagnostic service operates non-emergency NHS hours, i.e. Monday to Friday 0900 – 1700 hours. It does not operate on bank holidays.

2.3 Population covered

This service covers patients registered with an English or Scottish GP, resident in the European Union and eligible for treatment in the NHS under reciprocal arrangements. Patients from Wales and Northern Ireland are not part of this commissioned service and the trust must have separate arrangements

2.4 Any acceptance and exclusion criteria

Referral criteria, sources and routes

Referrals are primarily from other specialist centres such as respiratory units, ear, nose and throat surgeons, cardiologists and neonatal units. Increasingly physicians and paediatricians will make referrals as will GPs. All of these are screened by the specialist team in each centre and appointments made to either have the patient come to the centre or for the PCD consultant to visit another centre to see and assess patients and undertake brushings where appropriate.

All referrals will have the same level of assessment, screening tests [if at the specialist centre where Nitric Oxide is available] and uniformity of experience in the diagnostic process. This is possible because all referrals are triaged by the consultant lead from one of the three specialist centres.

Exclusion criteria

The service has no excluding factors for clinical assessment. Nasal NO is the screening test used by all three centres. It involves patient having a sample of gas from their nose analysed during a breath hold. The test can also be done during normal tidal breathing where patients are too young or unable to hold their breath.

This latter technique is less sensitive, but can be a useful indicator to assist the team

where brushings have been difficult to obtain.

Response time & detail and prioritisation

All new referrals at will seen well within the 18 week waiting list time. Any urgent requests are dealt with immediately. This may involve patients on neonatal intensive care unit (NICU) where a diagnosis is deemed very important. Cardiac patients who are requiring surgery and a co-existent diagnosis of PCD may be crucial to their management and outcomes. A Nitric Oxide and brushings services is also provided for other units the provider has links with.

2.5 Interdependencies with other services

Internally the PCD teams will link into multiple clinical and administrative teams as a result of the composition of the broad multi-disciplinary team (MDT).

External to this the nationally designated PCD providers are the leaders in the NHS for patient care in this area. They provide a direct source of advice and support when other clinicians refer patients into the nationally designated providers.

The national providers will form a relationship with local health and social care providers to help optimise any care for PCD provided locally for the patient. This may include liaison with consultants, GPs, community nurses or social workers etc.

Accessibility/acceptability

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

The service is commissioned by NHS England for all eligible patients from England. The clinic can be accessed by any eligible patient who is suspected to have PCD irrespective of gender, age, sex, disability, religious belief. Interpreters or use of a language line will be provided for families for whom English is not their first language.

The service is expected to demonstrate equitable geographical access across the country and take actions to address gaps in access.

The provider will provide information to patients on public transport access and accommodation for patients and relatives as needed.

An equality assessment impact has been completed for the establishment of this service.

Service description

Please note that unless specifically mentioned NHS England commissions all the listed services/functions.

NHS England does not commission services/functions at any non-designated trust. Any procedures carried out, any appointments attended or any diagnostic tests performed at non-designated trusts involved in the management of possible and confirmed PCD patients is not funded by NHS England. The NHS England assumption is that this activity will continue to fall within those trusts' service level agreements.

Care and referral pathways

Patients with suspected PCD are usually managed by a secondary or tertiary clinician involved in respiratory medicine. This could be a paediatrician with an interest in respiratory medicine or a specialist respiratory consultant within a specialist respiratory unit.

Where the patient is being managed will usually dictate the pathway for accessing the NHS England funded service.

If the patient is managed by one of the specialist respiratory clinicians who have been trained in taking the nasal brush biopsies then this will be performed by that clinician at their non-designated trust.

If the patient is not managed by one of these clinicians the patient will be referred to the designated centre. If the results of the biopsy are uncertain following the initial biopsy or the biopsy suggests PCD then a confirmatory biopsy is usually taken at one of the nationally designated centres. Where there are diagnostic concerns or where the initial biopsy suggests a positive result, patients, at the discretion of the centre director, maybe invited for repeat testing.

Samples already prepared for the electron microscope are occasionally sent through to a centre.

Each centre will develop a network of respiratory clinicians who will be trained in the nasal brush biopsy technique. University Hospitals Leicester NHS Trust has developed a biopsy service with Newcastle Hospitals NHS Foundation Trust, Leeds Teaching Hospitals NHS Trust, Royal Liverpool and Broadgreen Hospitals NHS Trust and Cambridge University Hospitals NHS Foundation Trust.

Royal Brompton and Harefield NHS Foundations Trust, which also runs a full post diagnostic care package, with regular multidisciplinary PCD clinics sees the majority of referrals at the centre. Biopsies are also sent in from other respiratory specialist units with which there is a close working relationship and where trained consultants are competent to undertake nasal brushings.

These include Great Ormond Street for Children NHS Foundation Trust, Barts Health NHS Trust and Kings College Hospital NHS Foundation Trust.

Southampton University Hospital NHS Foundation Trust has developed links with University Hospitals Bristol, Oxford University Hospitals NHS Trust and Royal Devon and Exeter NHS Foundation Trust.

Service configuration

Outpatient service

The start of the 'diagnostic' service will be a day attendance at either the centre or a linked trust.

At the linked trusts this attendance will usually consist of:

- History taken
- Clinical examination
- May need to perform a computerised tomography (CT) or ventilation scan. This is not covered under this service specification.
- Spirometry measured (if age-appropriate)
- Nasal brush biopsy
- Completion of clinical proforma to be returned with biopsy
- The biopsy is couriered to the designated centre's laboratory.

On rare occasions there might be a bronchoscopy sample provided by the referring trust.

The linked trust should always co-ordinate with the centre to make sure the laboratory staff are expecting the biopsy and that the appropriate brushings kit is sent out. This includes special media and cytology brushes plus instructions on courier conditions to ensure samples arrive in the best condition.

When the first attendance is at the centre the following additional services are performed:

- Nasal biopsy and/or bronchial biopsy where previous nasal biopsy failed
- Bronchoscopy where nasal biopsy not possible
- The biopsy is confirmed as adequate and the patient can go home. If not adequate, a second biopsy might be taken
- Measurement of nasal and exhaled nitric oxide (if age appropriate)
- Where patient has progressive respiratory disease there may be a need to perform a (CT) or ventilation scan
- Physiotherapist to take respiratory samples (cough swab, sputum) for culture, and assess airway clearance techniques.

The secretary will co-ordinate the attendances, the laboratories and dissemination of results to all professionals involved in the patients care.

In difficult cases, the lead PCD consultant will perform assessments and biopsies at the referring trust.

The service will provide nursing sessions to provide support at the centres. This will be when the patients attend for the biopsies and bronchoscopies.

Laboratory diagnostic service

The service will undertake the following diagnostic tests on the sample:

- Ciliary beat frequency measurement
- Ciliary beat pattern analysis
- Electron microscopy of ciliary ultrastructure
- Measurement of ciliary disorientation
- In cases of suspected primary ciliary dyskinesia especially cases of ciliary aplasia, ciliary disorientation, and unusual ciliary phenotype and in cases where secondary tissue damage renders diagnosis impossible cell culture with re-growth of the ciliated epithelium is required.

The electron microscopist, senior laboratory assistant and technician will review tissue taken and process all samples for functional analysis and electron microscopy. They will establish cell culture techniques for ciliary differentiation.

The technician will run the Nitric Oxide measurement service and perform spirometry, when the patients attend the centres.

In difficult cases and to facilitate the collection of high quality samples, the lead consultant will perform assessments and biopsies at the referring trust.

The senior laboratory scientist, electron microscopist and technician will review tissue taken and process all samples for functional analysis and electron microscopy.

The measurement of ciliary beat frequency and ciliary beat pattern will be standardised between the three diagnostic centres. Slow motion footage of cilia from each patient will be archived to allow audit and review by other centres.

Electron microscopy (EM) will be undertaken in each centre. Similarly, interpretation and reporting will be standardised between centres. EM samples from each centre will be archived to allow audit and review by other centres. Measurement of nasal and exhaled NO will be standardised between the three diagnostic centres. Members of the PCD diagnostic team will run the NO measurement service and perform spirometry when patients attend the centres. (This maybe the nurse or the physiotherapist)

Specialist cell culture methodology allows differentiation/growth of cilia from biopsies. This is essential in cases of suspected ciliary aplasia, disorientation, central microtubular agenesis, unusual phenotypes and in patients with persistent secondary damage to their ciliated epithelium. Methodology will be standardised between the three centres.

Measurement of nasal and exhaled Nitric Oxide will be as standardised as possible

by the diagnostic centre.

History, examination, ciliary functional analysis and detailed electron microscopy will be reviewed by the multi-disciplinary team led by the lead PCD consultant and a summary letter generated following this meeting. A letter containing the team's interpretation of the results is sent to the referring consultant. The centre will also provide published guidance on the management of the diagnosed condition.

Once the diagnosis of primary ciliary dyskinesia is established the following are essential:

- Complete care or care shared with an expert in paediatric respiratory medicine
- Access to an experienced respiratory physiotherapist
- Access to consultant ear nose and throat (ENT) surgeons preferably with a specialist interest in the management of ear problems in patients with PCD.

Access to these three services is not commissioned within this contract. It is assumed that this activity will fall within the relevant trust's current service level agreements (SLA).

All of these services will have access to tertiary advice from the relevant PCD specialist.

Teaching, training and liaison

The other diagnostic centres will support training of new personnel.

Following training the electron microscopist, senior laboratory scientist and technician will be able to cover each other for leave/sickness.

The lead PCD consultants will be responsible for education of other medical staff in the UK regarding the diagnosis and treatment of patients with PCD. This will include protocols guiding which children should be referred, and the development of an evidence base for treatment.

The lead PCD consultants will liaise with the UK PCD patient support group.

The electron microscopist, senior laboratory scientist and technician will have time allocated for teaching, audit, research and personal development.

Audit

All centres are expected to collaborate to perform audits.

The electron microscopist, senior laboratory scientist and technician will organise the auditing of results between centres.

Review of electron microscopy (EM) results by another diagnostic centre on a regular basis is essential to maintain standards. All EM blocks and grids should also be retained for audit between specialised centres. Digital pictures from each sample

showing a defect or normality will also be archived.

A video collection and EM specimens are available to demonstrate experience in the diagnosis of all of the phenotypes of PCD.

Storage of ciliary beat pattern and frequency of all patients, in slow motion, for future audit. All functional assessments will be stored on digital tape or DVD to allow audit of results at any time in the future.

Centres should also be able to grow ciliated respiratory epithelium in cell culture.

Follow up of patients:

- A national database will be established of patients with PCD. This will not be funded by NHS England . The establishment of the database will allow sufficient patients to be approached to take part in clinical trials of therapy
- Lung function measurements will be collected prospectively, following diagnosis, from referring centres
- Contact with ear, nose and throat and respiratory/general consultants caring for patients with PCD will be established.

Risk management

Care delivered by the PCD service must be of a nature and quality to meet the care standards, specification and agreement for the service. It is the trust's responsibility to notify the commissioner on an exceptional basis should there be any breaches of the care standards. Where there are breaches any consequences will be deemed as being the trust's responsibility.

Patients must be managed in line with the specification and care standards. Any deviation from these which has not been approved by NHSE is at the trust's risk both clinically and financially. It is the trust's responsibility to inform the commissioners of any such non-approved deviations on an exceptional basis.

Where a patient's presentation challenges the assumptions that underpin the specification, service standards and contractual arrangements, it is the trust's responsibility to inform the commissioners on an exceptional basis, prior to any treatment (except for emergency treatment) so that the implications of the patient's requirements can be considered. This does not affect situations where the individual funding application process applies.

3. Applicable Service Standards

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

Providers will carry out a mandatory patient note and consent audits in accordance with the trust audit requirements.

All audits and the patient satisfaction survey must be registered with the trusts audit department.

Providers must comply with the trusts requirements for safe haven status when transferring patient information between NHS organisations.

Also see NHS England service standards for PCD.

Continual service improvement plan

The PCD service 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 will agree a service development improvement plan with commissioners and demonstrate progress at joint service review meetings.

Areas of service improvement may be stimulated through the following areas:

- Complaints
- Monitoring information
- Learning from other services (if appropriate)
- Service user feedback / patient involvement;
- Research
- Policy / guidance on best practice e.g. NHS Institute for Innovation and Improvement
- Other communication with stakeholders.

4. Key Service Outcomes

Percentage of patients tested positive.

5. Location of Provider Premises

The service is delivered in three locations:

Royal Brompton & Harefield NHS Foundation Trust
University Hospitals of Leicester NHS Trust
Southampton University Hospital NHS Foundation Trust

Sub-contractors

None

Adopted

Primary Cilliary Dyskinesia – Patient Pathway

