

## SCHEDULE 2 – THE SERVICES

### A. Service Specifications

<b>Service Specification No:</b>	<b>170050S</b>
<b>Service</b>	<b>Primary Ciliary Dyskinesia Management Service (adults)</b>
<b>Commissioner Lead</b>	
<b>Provider Lead</b>	

#### 1. Scope

##### 1.1 Prescribed specialised service

This service specification covers the provision of Primary Ciliary Dyskinesia Management Services for individuals from the age of 16 years. NHS England also commissions a Diagnostic Service for Primary Ciliary Dyskinesia [service specification E13/S (HSS)/d] and a Primary Ciliary Dyskinesia Management Services for children including outreach when delivered as part of a provider network [service specification E13/S (HSS)/g].

##### 1.2 Description

Primary ciliary dyskinesia (PCD) is a genetic condition in which the microscopic cells in the respiratory system called cilia do not function normally. Ciliary dysfunction prevents the clearance of mucous from the lungs, paranasal sinuses and ears. Recurring respiratory infections can lead to an irreversible scarring and obstruction in the bronchi (bronchiectasis) and severe lung damage. Cilia are also present in the ventricles of the brain and in the reproductive system so ciliary dysfunction can also affect other parts of the body.

Primary ciliary dyskinesia management services include services provided by Highly Specialist Primary Ciliary Dyskinesia Management centres including outreach when delivered as part of a provider network. This provision applies to adults. Patients will transition into the service from the paediatric HSS PCD service.

##### 1.3 How the service is differentiated from services falling within the responsibilities of other commissioners

NHS England commissions management services for adults with PCD from Highly Specialist Primary Ciliary Dyskinesia Management centres, including services delivered

on an outreach basis delivered as part of a provider network.

Clinical Commissioning Groups (CCGs) commission inpatient care for adults with PCD.

Activity is identified via local data flows, which apply to Highly Specialised Primary Ciliary Dyskinesia Management centres only.

## **2. Care pathway and clinical dependencies**

### **2.1 Care pathway**

Access to treatment is guided by any applicable NHS England national clinical commissioning policies.

The national adult PCD service works closely with the paediatric PCD services and provides a geographically accessible service for patients offering seamless transition for patients from the paediatric PCD centres. The paediatric HSS manages patients up to the age of 16 years, thereafter patients will be managed in the national adult PCD service. Each service additionally provides an outreach service to ensure national equity of access.

The centrally commissioned specialised service provides all patients with an annual review, access to the multidisciplinary team (MDT) and a detailed management plan. Further outpatient appointments at the specialist centre are dictated by clinical need and stability. Some PCD patients require intravenous antibiotics either at home or as inpatients. Inpatient care, with appropriate specialised physiotherapy and MDT input, is provided either at the specialist centre or in local services with supervision by the specialist centre.

#### **2.1.1 Entry to the service**

Patients entering the service are likely to be:

- paediatric transition patients with a confirmed diagnosis of PCD
- adult patients with a confirmed diagnosis of PCD made by the PCD Diagnostic Service
- adult patients with clinically suspected PCD but with indeterminate results of investigations by the PCD Diagnostic Service.

#### **2.1.2 Routine Care (see Figure 1)**

##### **i. Annual review**

All patients have an annual review at the specialist centre. This involves a day in the specialist centre and includes the following in all patients:

- Formal lung function testing
- Physiotherapist review
- ENT specialist review
- PCD nurse specialist review

- PCD consultant review
- Sputum culture sent for bacterial, mycobacterial and fungal culture
- Blood tests:
  - Full Blood Count (FBC)
  - Urea and Electrolytes (U&Es)
  - Liver Function Test (LFTs)
  - C-reactive Protein (CRP)
  - Erythrocyte Sedimentation Rate (ESR)
  - Allergic bronchopulmonary aspergillosis (ABPA) markers
  - High-resolution computed tomography (HRCT) scan
- Chest X-Ray
- Audiology assessment
- Written annual plan for patient, primary and secondary care.

Additionally the following are available for all annual review patients as needed:

- Dietician review
- Psychology review
- Further blood tests including immunoglobulins, pneumococcal antibodies, aspergillus, ImmunoCAP for antigen-specific antibodies of immunoglobulin class G (ICAP IgG), drug monitoring
- HRCT scan
- Echocardiogram
- Fertility advice (limited dates / year).

## **ii. Additional clinic review**

It is anticipated that some patients have six-monthly, or for the most severe or progressive patients, three-monthly appointments at the specialist centre. The following are performed at each additional routine clinic visit for all patients:

- Spirometry
- Physiotherapist review
- PCD nurse specialist review
- PCD doctor review
- Sputum culture sent for bacterial, mycobacterial and fungal culture.

Additionally the following are available as needed:

- ENT specialist review
- Dietician review
- Psychology review
- Blood tests - FBC, U&Es, LFTs, CRP, ESR, ABPA markers
- Chest X-Ray
- HRCT scan.

## **iii. Transition clinic**

Transition to the adult service is co-ordinated by the paediatric management centres involving an integrated care pathway. It culminates in the transition clinic which is a joint clinic performed with a Paediatric PCD centre with the members of the paediatric MDT and attended by the adult PCD consultant and the adult PCD specialist nurse. At this clinic appointment, in addition to the standard clinic review, all aspects of the patient care are handed over and the adult team describes the practicalities of the adult service and answers questions from the patient (and family). The same adult PCD consultant (where possible) and PCD nurse see the patient at the first adult clinic following on from transition.

#### **iv. Relationship with non-specialist centres / Outreach clinics**

All PCD patients are reviewed in the specialist centre at least annually with the provision for more regular follow-up appointments as above. Each specialist centre provides outreach clinics with a PCD consultant, nurse specialist and physiotherapist to cater for those patients who require more than an annual review in the specialist centre, but find the specialist service difficult to access.

#### **v. Urgent clinic review**

Weekly urgent appointments / assessments are available at the specialist centres where patients are reviewed and the following provided:

- spirometry
- physiotherapist review
- PCD nurse specialist review
- PCD doctor review
- sputum culture sent for bacterial, mycobacterial and fungal culture.

Additionally the following are available as needed:

- Blood tests - FBC, U&Es, LFTs, CRP, ESR, ABPA markers
- Chest X-Ray
- HRCT scan.

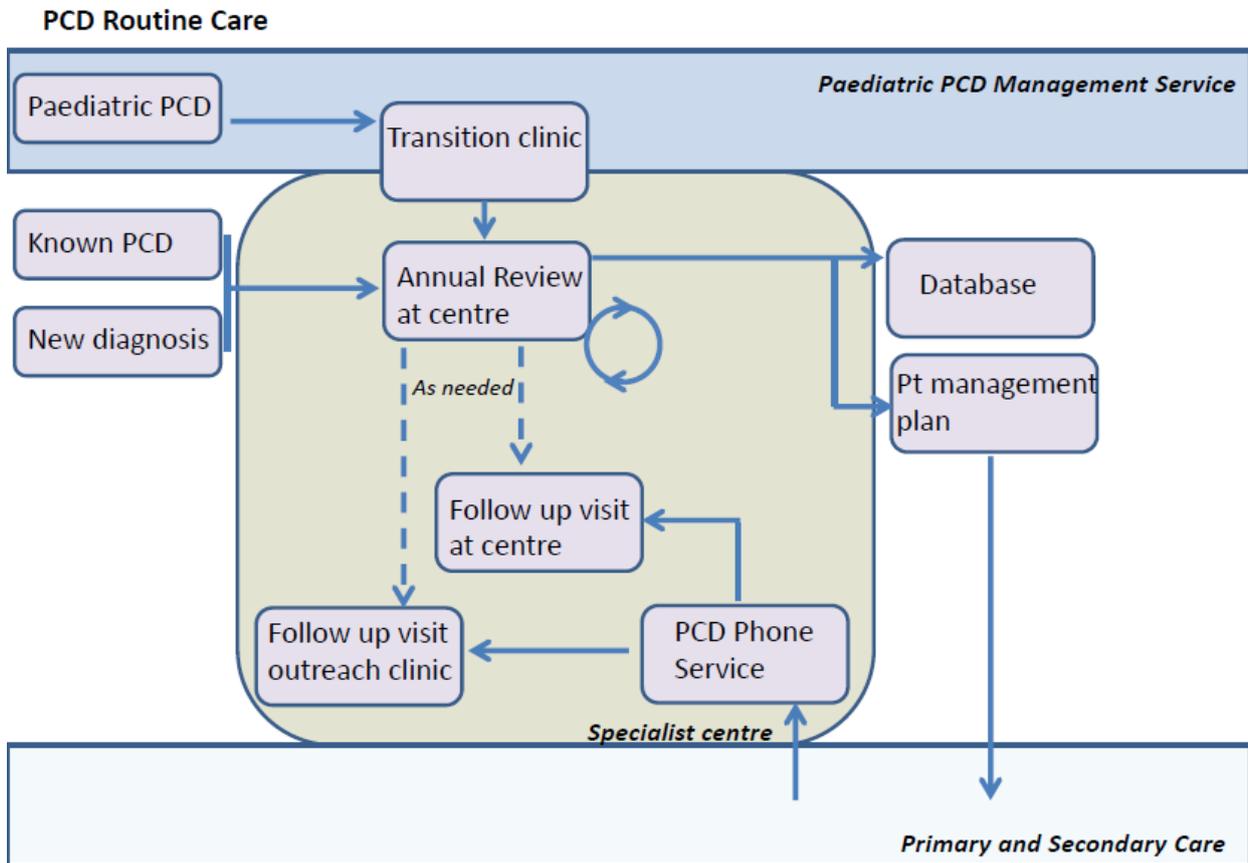
#### **vi. Phone advice**

Each centre provides a specialist nurse-staffed phone line during working hours with direct consultant input available for both patients and healthcare workers.

#### **vii. Other available services**

There is availability for specialist bronchoscopy for adult PCD patients as needed at the specialist centres. The service delivers evidence-based management developed through the best use of educational resources that are easily accessible by patients and professionals.

**Figure 1**



**2.1.3 Acute care (see Figure 2)**

All patients have a management plan for when they are unwell. Within working hours, further advice is available from the specialist nurse staffed phone line with direct consultant input. For acute problems outside working hours, patients can seek advice from their local hospital. There is provision for weekly urgent appointments /assessments at the specialist centres as above. Local hospitals that perform acute patient assessments are able to liaise with the specialist centres as needed.

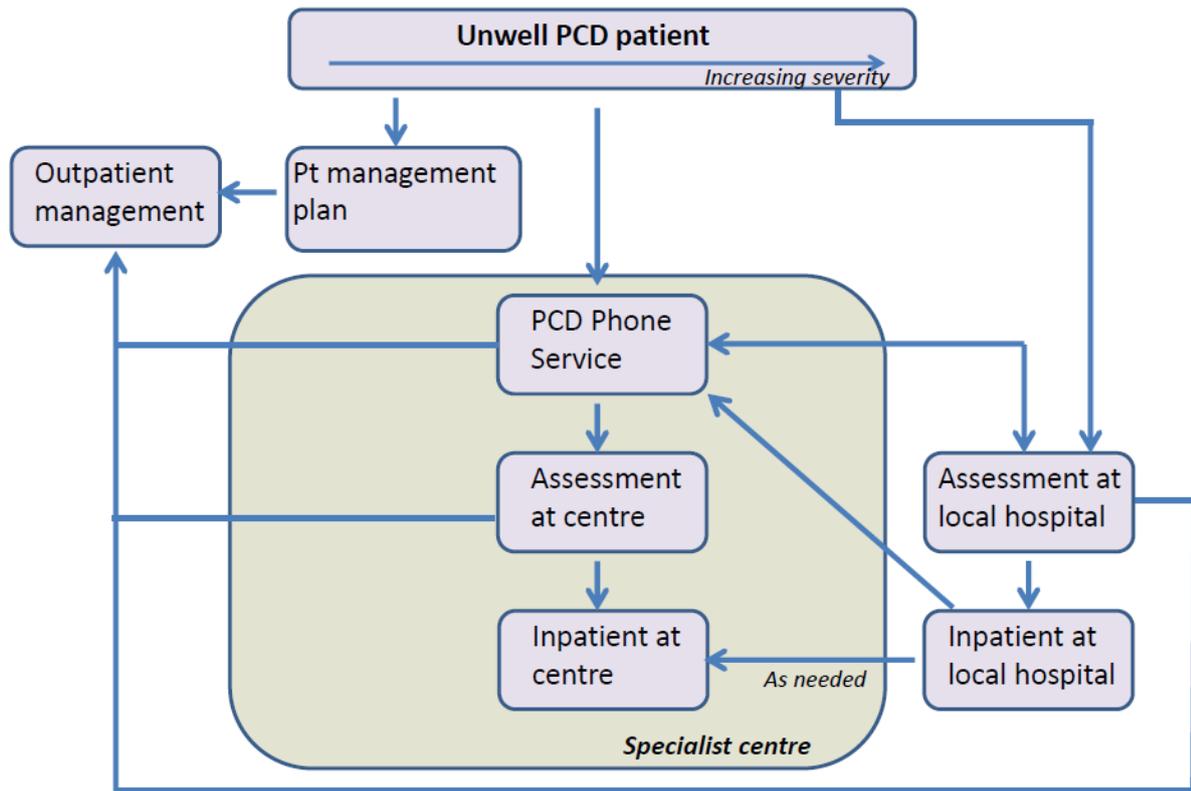
**2.1.4 Hospital admissions**

The flow chart below demonstrates the various entry points to inpatient admission. Assessments and inpatient admissions can be arranged directly in the specialist centres. Acute presentations are also be made to local hospitals based on severity and practicalities. Specialist centres liaise with the local centres to provide advice. The

inpatient spell at the local hospital is outside of the PCD service model and is outside of the scope of the service specification.

**Figure 2**

**PCD Acute Care**



**2.2 Interdependence with other services**

Figures 1 and 2 illustrate the interdependencies between the highly specialised centre, secondary and primary care. There is significant liaison with local hospitals that provides some acute assessments and inpatient care in liaison with the specialist centres.

There is a specialist nurse staffed phone line during working hours with direct consultant input to help provide this link.

Outreach clinics are provided jointly with local and specialised centres. Transition clinics are held jointly by the adult and paediatric PCD management services.

The service offers management advice to local providers where patients are admitted for IV antibiotics.

The PCD database spans both the adult and paediatric services. For patients with severe disease, there are interdependencies with ventilation services for patients that need assisted ventilation and interdependencies with transplant services for appropriate patients. There are also interdependencies with cardiac and other specialties for other rare conditions associated with PCD.

The service links closely with the Highly Specialised Diagnostic service for Primary Ciliary Dyskinesia.

### **3. Population covered and population needs**

#### **3.1 Population covered by this specification**

This service is for all patients with a diagnosis of PCD over the age of 16 years. This can be patients with a diagnosis made by the PCD Diagnostic Service, patients with a diagnosis of PCD made by a diagnostic service outside the UK, with diagnostic criteria compatible with those in England, or in rare cases, patients in whom a definitive diagnosis has not been made, but in whom the diagnosis is extremely likely (in the clinical opinion of the PCD Specialist). It is a lifelong service for these patients. The service can be accessed by any eligible patient with a diagnosis of PCD irrespective of gender, age, sex, disability, religious belief. Interpreters or use of a language line are provided for families for whom English is not their first language.

This service specification covers the population defined as the commissioning responsibility of NHS England. Commissioning arrangements for the devolved nations in relation to this service are as set out in *UK-wide Commissioning Arrangements of Highly Specialised Services*.

Trusts performing procedures on patients outside of S2 arrangements and aligned referral arrangements need to continue to make the financial arrangements directly with the governments involved, separately from their contract with NHS England.

Children (under 16 years of age) and patients found not to have PCD are not covered by this service. The paediatric HSS service manages patients up to the age of 16 years only.

#### **3.2 Population needs**

Primary Ciliary Dyskinesia (PCD) is a rare (estimated between 1:26000 - 1:40000) [<http://erj.ersjournals.com/content/36/6/1248.long>] hereditary disorder of ciliary dysfunction leading to multisystem abnormalities which include chronic respiratory, sinus and ear infections, cardiac abnormalities, infertility and ectopic pregnancies.

### **3.3 Expected significant future demographic changes**

None expected.

### **3.4 Evidence base**

Chronic infection of the upper and lower airway is the most common. Without appropriate specialised treatment, progressive chronic lung disease and bronchiectasis develop. If diagnosis is delayed or managed sub optimally, permanent bronchiectasis and deterioration of lung function occurs.

(<http://www.atsjournals.org/doi/pdf/10.1164/rccm.200303-365OC>)

Highly specialised management is needed to be provided for this condition by a multidisciplinary team of professionals trained in the multisystem manifestations of PCD. The importance of this is highlighted by a number of longitudinal studies demonstrating an improved patient course with diagnosis and the institution of appropriate management. (<http://erj.ersjournals.com/content/10/10/2376.long>) (<http://www.atsjournals.org/doi/pdf/10.1164/rccm.200811-1731OC>).

There is evidence that the lack of intervention results in up to 25% of adult patients developing respiratory failure, all of whom die early unless they have a lung transplant (<http://www.atsjournals.org/doi/pdf/10.1164/rccm.200303-365OC>). Poor management leads to a cycle of damage, increased risk of infection, further damage, leading to accelerated decline.

Specialised care is very important for this group of patients and has a direct impact on disease progression, morbidity and mortality, satisfying all of the NHS Framework domains. In addition to the substantial patient benefits, it reduces the pressures placed on resources by poorly managed patients with reduced hospitalisations, reduced need for advanced respiratory support and reduced socioeconomic costs of chronic ill health.

This service collaborates closely with the PCD paediatric service commissioned by NHS England. The service ensures excellence in transition to adulthood, a vulnerable period for patients. Adult patients continue to receive specialist input and the significant gains that have been made in the paediatric specialist service are maintained. The adult specialist service is crucial to build on the successes in the paediatric population and to directly care for the group of patients with more severe disease. In addition to the improvement in care and outcome that is provided by a specialised adult service, the service also ensures an improved patient experience compared to care outside of the specialist service.

## **4. Outcomes and applicable quality standards**

### **4.1 Quality statement: Aim and objective of service**

This service provides a specialised management service for adult patients with PCD.

The core objective of this service is to ensure that all patients with PCD have access to the optimal standards of care to enable a reduction in the morbidity and mortality associated with the condition.

The service aims to:

- ensure that all PCD adult patients, wherever they live, have access to and are managed according to optimal standards of care. This includes ensuring that:
  - all PCD adult patients have an annual review by a specialist PCD MDT consisting of a PCD specialist consultant, physiotherapist, nurse specialist and ENT specialist with access to a dietician and psychologist as needed.
  - all PCD adult patients have access to appropriate home and inpatient antibiotic services
  - care and management of patients with PCD is coordinated so that patients receive appropriate respiratory, ENT, cardiac, obstetric and physiotherapy care, fertility advice, palliative care and care for other conditions associated with PCD
  - patients / families and other health professionals are educated on the implications and management of PCD.
- raise awareness in the adult respiratory community about the diagnosis of PCD to ensure that these patients are transferred from bronchiectasis clinics to the PCD specialist service to monitor their condition and limit disease progression
- reduce the morbidity and mortality related to PCD, as well as the economic burden, associated with late diagnosis and poorly managed disease
- enter patients into a national database.

**NHS Outcomes Framework Domains**

<b>Domain 1</b>	<b>Preventing people from dying prematurely</b>	√
<b>Domain 2</b>	<b>Enhancing quality of life for people with long-term conditions</b>	√
<b>Domain 3</b>	<b>Helping people to recover from episodes of ill-health or following injury</b>	√
<b>Domain 4</b>	<b>Ensuring people have a positive experience of care</b>	√
<b>Domain 5</b>	<b>Treating and caring for people in safe environment and protecting them from avoidable harm</b>	√

## 4.2 Indicators Include:

Number	Indicator	Data Source	Outcome Framework Domain	CQC Key question
<b>Clinical Outcomes</b>				
101	% of patients in the PCD management service offered an annual review appointment. The annual review will consist of the processes listed in the service specification	National database	1, 2, 3, 4, 5	Safe, effective, caring, responsive
102	% of patients seen by a physiotherapist at annual review	National database	1, 2, 3, 4	effective, caring, responsive
103	% of patients seen by a nurse specialist at annual review	National database	1, 2, 3, 4	effective, caring, responsive
104	% of patients seen by a ENT specialist at annual review	National database	1, 2, 3, 4	effective, caring, responsive
<b>Patient Experience</b>				
201	Patient feedback	Self declaration	4	caring, responsive
202	Patient Information	Self declaration	4	caring, responsive
<b>Structure and Process</b>				
301	Team leadership	Self declaration	4	well-led
302	MDT Membership	Self declaration	1, 3, 4	well-led, effective, caring
303	Availability of advice	Self declaration	1, 2, 3	effective, caring, responsive
304	Transition clinics	Self declaration	1, 2, 3, 4	effective, caring, responsive
305	Patient pathways	Self declaration	1, 2, 3, 4	safe, effective, caring, responsive
306	Clinical Guidelines	Self declaration	1, 2, 3, 4	safe, effective, caring, responsive
307	Participation in clinical audit	Self declaration	1, 2, 3, 4	safe, effective, caring
308	Data collection	Self declaration	1, 2, 3	safe, effective

For recording outcomes, data from annual review measurements is collected in a national database which allows documentation of variables including number of patients in each centre, referrals to each centre, annual reviews performed, outpatient episodes per patient, inpatient episodes per patient.

**Detailed definitions of indicators, setting out how they are measured, are included in schedule 6.**

#### **4.3 Commissioned providers are required to participate in annual quality assurance and collect and submit data to support the assessment of compliance with the service specification as set out in Schedule 4A-C**

There is a requirement to hold national audit meetings involving all designated centres on an annual basis.

Each centre must assure that:

- all practitioners participate in continuous professional development and networking
- patient outcome data is recorded and audited across the service
- they participate in the national audit commissioned by NHS England. Audit meetings should address:
  - clinical performance and outcome
  - process related indicators e.g. efficiency of the assessment process, prescribing policy, bed provision and occupancy, outpatient follow-up etc
  - stakeholder satisfaction, including feedback from patients, their families, referring clinician and GPs.

#### **4.4 Applicable CQUIN goals are set out in Schedule 4D**

To be agreed with the Commissioner.

### **5. Applicable service standards**

#### **5.1 Applicable obligatory national standards**

Best practice guidelines from specialist centres were collated to produce a European consensus statement in children with PCD (Barbato et al Eur Respir J 2009; 34:1264-76) which recommends regular follow up in specialist centres with an MDT approach as described in this service specification. The British Thoracic Society guidelines on bronchiectasis were produced in 2010 (Pasteur et al Thorax 2010 65S). These provide general bronchiectasis advice but do not provide comprehensive PCD specific guidance.

#### **5.2 Other applicable national standards to be met by commissioned providers**

Nothing additional.

### **5.3 Other applicable local standards**

Not applicable.

### **6. Designated providers (if applicable)**

No more than four centres are proposed, which will establish links with the four paediatric PCD management services.

### **7. Abbreviation and acronyms explained**

The following abbreviations and acronyms have been used in this document:

CCGs - Clinical Commissioning Groups

ESR - Erythrocyte Sedimentation Rate

PCD - Primary Ciliary Dyskinesia

HRCT - High Resolution Computed Tomography

ICAP IgG - ImmunoCAP for antigen-specific antibodies of immunoglobulin class G

IV - Intra Venous

FBC - Full Blood Count

U&Es - Urea and Electrolytes

LFTs - Liver Function Test

CRP - C-reactive Protein

ABPA markers - Allergic Bronchopulmonary Aspergillosis Markers

HRCT scan - High-resolution Computed Tomography