# Primary ciliary dyskinesia: a national expert consensus statement on standards of care

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**This national consensus statement sets out standards of care for the management of children and adults with primary ciliary dyskinesia to promote consistency for patient care and interprofessional collaboration and to enable benchmarking for future care** https://bit.ly/46BksAd

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## Abstract

Primary ciliary dyskinesia (PCD) is a genetically and clinically diverse disorder characterised by loss of normal ciliary function leading to chronic oto-sino pulmonary disease, situs abnormalities and subfertility in men and women. There is limited evidence to support robust guidelines on the management of children and adults with PCD; however, there is a clear clinical need to establish a framework of care for the follow-up of these patients. The European Respiratory Society (ERS) has published consensus statements on diagnostic and treatment approaches in children with PCD, and the BEAT-PCD (Better Experimental Approaches to Treat PCD) network provides guidance on infection prevention and control. This is a national consensus statement to outline a set of standards for the provision of specialist care for children and adults with PCD living in England. A national PCD expert panel made up of specialists working in both paediatric and adult UK highly specialist management services, was established to create a consensus statement on the minimum standards of care for PCD. Using a modified Delphi process, consensus to a statement required at least 80% agreement within the PCD expert panel group. Patient organisation representatives were involved in reviewing the statement and have produced an accompanying layperson summary. We present a consensus statement on 15 standards covering provision of pulmonary, ear, nose and throat, and fertility care, screening for situs abnormalities and transition from paediatric to adult care services. It is targeted at clinicians and allied health professionals managing paediatric and adult patients with PCD, patient organisations and patients and their families.

## Introduction

Primary ciliary dyskinesia (PCD) is a genetically heterogeneous multisystem disorder characterised by abnormal function (and/or ultrastructure) of the motile cilia. Clinically it presents with chronic upper and lower respiratory tract symptoms, bronchiectasis, recurrent sinopulmonary infections and subfertility in men and women [1, 2]. People with PCD also manifest laterality defects in about 50% of cases [2, 3] and congenital heart disease in 5–17% of the population [1–3].

The prevalence of PCD is likely to be underestimated and genomic analyses suggest a global prevalence of at least 1:7500 [4] with variation between ethnic groups, and higher prevalence in highly consanguineous populations [5].

There is a lack of clear evidence on the best approach to monitoring and managing patients with PCD, and much of the current practice is extrapolated from cystic fibrosis (CF) and non-CF bronchiectasis guidelines. However, despite certain similarities, these conditions differ in pathophysiology and outcomes. While overall long-term prognosis appears better in the majority of PCD phenotypes than in CF, children with PCD generally have lower lung function than those with CF [2, 6] and morbidity can be considerable if the condition is managed incorrectly.

Centralisation of care of rare conditions in referral centres ensures adequate expertise in diagnosis and management, appears to reduce the time to diagnosis and has the potential to improve outcomes [2, 7, 8]. For this reason, in England, the National Health Service (NHS) has established centrally commissioned highly specialised services (HSSs) across the country for the management and care of individuals with PCD. The services have been established to ensure time and funding for a multidisciplinary team (MDT), including respiratory and ear, nose and throat (ENT) physicians, respiratory physiotherapists and clinical nurse specialists, dedicated to the care and management of people with PCD. These HSS management services are located across the country and cover specific geographical areas to ensure adequate provision of service across the country (see supplementary material for description of centres and areas covered).

The aim of this project was to create a standards-of-care document to ensure that all patients with PCD in England have access to high-quality standardised care, to create a benchmark for auditing and assessing the HSS services, and to highlight areas where further research and development are required.

## Methods

A panel of physicians leading the four HSS adult and paediatric PCD management services in England was established to develop a consensus on standards of care. A modified Delphi consensus approach *via* online surveys (eDelphi) was used to collect expert opinion supported, when possible, by evidence, ideally in PCD or in CF and non-CF bronchiectasis.

An initial exploratory survey (available in the supplementary material) was sent out to the clinical leads of the four adult and paediatric management services in England to be completed with the support of and/or on behalf of their MDT. The survey investigated the current and aspirational practices of the four services with regards to access to care, management of routine and annual review appointments, monitoring and treatment for people with PCD and highlighted the topics for the statements. A focused literature search was then completed by three members of the panel (ER, AB and GS), to review the available evidence relevant to each statement topic. Search terms used included search words for PCD, CF and non-CF bronchiectasis, combined with search words relevant to each statement topic. Survey results and literature review were used to formulate the initial statements. Panel members were asked to comment on the statements using a Likert scale. Statements that reached 80% of agreement were considered final, those with <40% agreement were excluded. Statements where the agreement was between 40% and 80% were reviewed following a round of discussion. In addition, statements where specific comments were made by more than one panel member were also reworded, irrespective of the agreement level. A second round of eDelphi was conducted on the revised statements.

Two members of the UK patients and family group (PCD Support UK) were involved in planning this work and were invited to comment on the statements between the first and second round, and at completion of the Delphi process. In addition, patients and families from each paediatric and adult management centre were invited to comment on the proposed statements once the initial survey and literature review were completed, to ensure that the topics discussed were appropriate and relevant, and at least one representation from each centre was received.

## Results

A total of 15 panellists contributed to the development of the consensus. Panel members were a combination of paediatric (n=8) and adult (n=7) respiratory physicians, working in large tertiary centres with expertise in the management of PCD. Two members of PCD Support UK were also involved in the revision of the statements.

A total of 16 statements were initially developed to cover topics such as: access to care in the management service, provision of annual reviews, routine clinic follow-up and urgent reviews, disease monitoring and management with respect to microbiology, lung function, radiology and audiology. All panellists provided a response to both rounds of statements. All the statements had an agreement of at least 40%, with 13 reaching >80%. A total of four statements were reworded and all reached consensus. Statements 2 and 3 of the initial round reached a high level of agreement (>90%) but, in view of the comments received, were reworded into a single statement. The consensus led therefore to the development of a total of 15 statements.

Figure 1 shows a flowchart of the process and table 1 summarises the statements. Rejected and revised statements are in the supplementary materials.

## Discussion

In this consensus statement, a framework for management and monitoring of individuals with PCD is proposed, with the scope of standardising care for PCD, setting a benchmark to assess PCD services and accelerating clinical research to underpin the evolution of evidence-based best practice.

The panel agreed that prompt access to a PCD management service should be encouraged and become a priority (Statement 1). Adults and children with PCD often attend numerous clinical appointments prior to the consideration of a PCD diagnosis and receive inappropriate treatment for conditions they do not have (*e.g.* inhaled corticosteroids for presumed asthma [9]). Older age of diagnosis has been associated with more frequent pulmonary exacerbations requiring antibiotics [10] and previous surgical lung resection [11, 12], with surgery having occurred prior to diagnosis in half of [12] or all cases [11]. PCD diagnostic teams should be able to directly refer newly diagnosed individuals or those with a highly likely diagnosis to a management service to avoid further delays in commencing appropriate treatment and follow-up.

While limited evidence is available on the impact of regular monitoring on outcomes among people with PCD; the identification of reference centres for diagnosis and management has been shown to allow for an

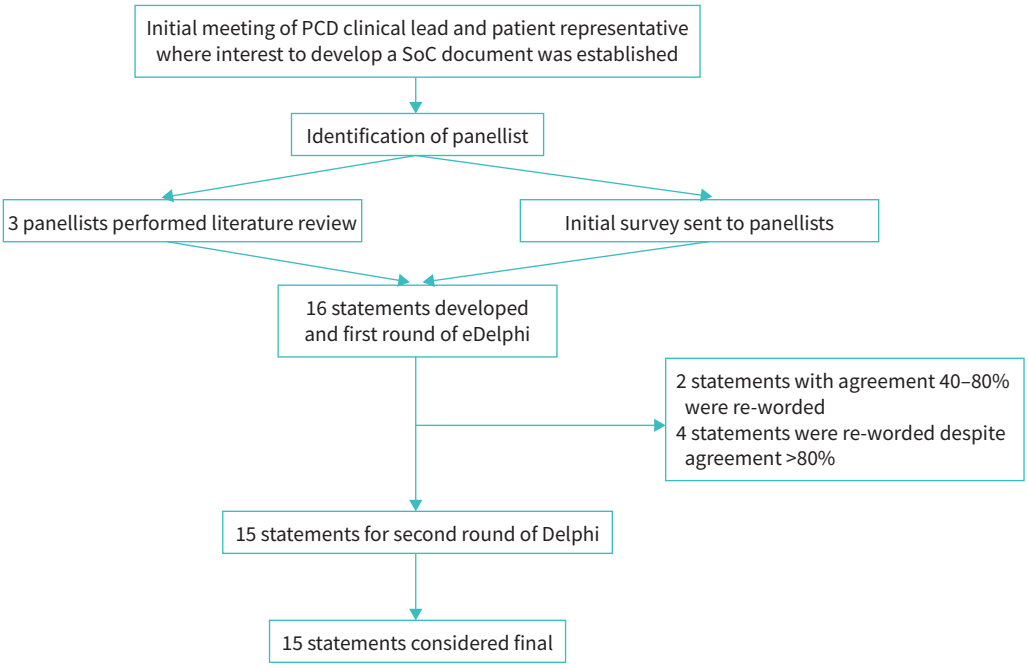


FIGURE 1 Flowchart of the eDelphi process. PCD: primary ciliary dyskinesia; SoC: standards of care.

**TABLE 1** Statement for standards of care agreed by the panel (in brackets the grade of evidence is reported)

| | Statement | % agreement and round |
|---|---|---|
| **1.** | All individuals with PCD (confirmed or highly likely [7]) should have early access to a national PCD management service with specified MDT capacity [D]. | 100% agreement - first round |
| **2.** | All individuals with PCD (confirmed or highly likely) should be reviewed at least annually by the specialist PCD management service MDT [D].<br>a. At annual review, people with PCD should be offered a review with an ENT consultant who has a specialist interest and knowledge in PCD management [D].<br>b. All children with PCD should be offered an audiology review at least annually. Older children and adults with PCD should be offered an audiology review at diagnosis (if late) and as clinically indicated [D]. | 100% agreement - first round;<br><br>93% agreement - first round |
| **3.** | Routine review should be completed at a local respiratory unit or PCD management service centre every 3 months for children and as clinically indicated for adults. Support and coordination of care is available from the PCD management service as required. | 100% agreement - first round |
| **4.** | Respiratory samples should be sent at each clinic review for culture. Samples should be processed as extended culture (as per CF standards) at annual review and additionally if clinically indicated [D]. | 100% agreement - second round |
| **5.** | CXR surveillance in children should be considered annually and as clinically indicated. CXR should be performed as clinically indicated in adults [D]. | 89% agreement - second round |
| **6.** | A chest CT should be performed as clinically indicated for children and adults. It should be particularly considered at transition to adult services if no previous CT available [C]. | 100% agreement - second round |
| **7.** | Unless a patient's PCD is known to be caused by a gene that does not cause laterality defects, all patients should have an echocardiogram and abdominal ultrasound to identify situs abnormality or cardiac defects. Patients in the adult service might require these tests if there is no previous documentation [D]. | 90% agreement - second round |
| **8.** | All patients able to perform spirometry should do so at routine and annual review [C]. | 93% agreement - first round |
| **9.** | As clinically indicated, full lung function measurements, hypoxic altitude simulation testing, lung clearance index, oximetry and sleep efficiency assessment should be available [D]. | 87% agreement - first round |
| **10.** | A full set of routine bloods, including full blood count, U&E, LFTs, but also immunoglobulins, functional antibodies, total IgE, RAST to aeroallergens and *Aspergillus* markers should be performed at diagnosis and repeated as clinically indicated [D]. | 93% agreement - first round |
| **11.** | Genetic testing for PCD should be performed during diagnostic work-up or as soon as possible for a patient with known PCD [D]. | 100% agreement - first round |
| **12.** | An individualised airway clearance therapy routine should be given to every individual with PCD and assessed as part of the annual review process [D]. | 100% agreement - first round |
| **13.** | Prompt recognition and treatment for pulmonary exacerbation is essential [D]. | 100% agreement - first round |
| **14.** | A structured transition programme should be implemented to allow adolescents to have the opportunity to be involved in their transition [D]. | 93% agreement - first round |
| **15.** | Fertility should be discussed during transition to adult care, and adults should have local access to genetic counselling and a fertility service [D]. | 93% agreement - first round |

PCD: primary ciliary dyskinesia; MDT: multidisciplinary team; ENT: ear nose and throat; CF: cystic fibrosis; CXR: chest radiography; CT: computed tomography; U&E: urea and electrolytes; LFTs: liver function tests; IgE: immunoglobulin E; RAST: radioallergosorbent test.

earlier diagnosis and improvement in clinical care [13, 14] with patients willing to travel distances to ensure they have access to specialised care [9]. Centres with expertise in the management of PCD and the development of dedicated care pathways in France have led to the improvement in lung function and reduction in hospital admissions for children with PCD [14]. Furthermore, many individuals with PCD highlighted that the delay in reaching a diagnosis has led them to have mistrust in the healthcare system [15]. Specialist referral centres for the management of the condition might help overcome this mistrust.

Despite many patients and families being willing to travel to receive specialist care [9], the panel suggested that the management services are responsible for completing annual reviews and coordinating care with local providers, where patients might receive routine care as appropriate. While the COVID pandemic accelerated a shift towards telehealth in several chronic conditions, including CF [16], and in the initial survey among panellists the role of remote review was assessed, the final consensus preferred not to comment on face-to-face *versus* remote consultation (Statements 2 and 3).

At annual review, patients should have appropriate assessment of their recent clinical history with a focus on number of exacerbations, lung function, respiratory sample culture, blood tests, observations and physical examination, assessment of nutritional status as well as physiotherapy review (Statements 2, 4, 8,

10 and 12). Encompassed in these measurements are the core outcome set for clinical trials recommended by the BEAT-PCD (Better Experimental Approaches to Treat PCD) consensus statement and by the recent consensus on blood testing in PCD [17, 18]. Monitoring of similar clinical parameters has also been included in the North American PCD Foundation recommendations on monitoring and treatment of PCD, though with some differences in the recommended frequency of measurement [19]. In adults with bronchiectasis, clinical parameters collected routinely in clinical practice, such as age, body mass index (BMI), forced expiratory volume in 1 s ($FEV_1$), history of hospital admission and exacerbations, Medical Research Council (MRC) dyspnoea score and microbiological data as well as pre-existing imaging data can predict future risks of exacerbations and mortality as well as quality of life [20, 21]. It is conceivable that similar data can aid the stratification of risk in individuals with PCD, where recognition of exacerbation is often particularly difficult due to the characteristic daily symptoms.

Guidelines on bronchiectasis recommend yearly lung function for adults and 3–6 monthly for children [22, 23], compared with more frequent monitoring of $FEV_1$ in CF recommended at every clinical review [24]. The role of lung function and spirometry specifically in PCD is unclear. There is a significant intra-subject variability of spirometry values, possibly related to the volume and burden of secretions [25]. This notwithstanding, the panel suggested routine monitoring of spirometry to start in childhood as soon as age appropriate (Statement 8). There is evidence of reduced lung function from early childhood in people with PCD, associated with progressive decline in a proportion of patients [6, 26, 27]. Furthermore, a recent large genotype –phenotype correlation study confirmed previous findings of smaller scale cohort studies [26, 28–30], showing that individuals with specific genotypes (*i.e.* CCDC39 and CCDC40) appear to have lower lung function [31]. As such, routine monitoring of lung function will provide valuable information to assess if $FEV_1$ could be an objective measure for disease progression and response to treatment in real life, both in the short term and in the long term and could further understanding of the factors impacting on lung function in PCD.

In view of the lack of any curative treatment, current options focus on preventing and managing disease complications. As such the mainstay for treatment consists in aiding the impaired mucus clearance, symptoms management and treatment of pulmonary exacerbations. While there is no evidence to support any specific airway clearance technique [32], good concordance with chest physiotherapy is a key factor contributing to lung function in the long term [33], despite patients not perceiving a short-term impact [34]. The panel emphasised the importance of regular, at least yearly, assessment and reviews with a specialist physiotherapist to form a personalised and tailored treatment plan for airway clearance, which could include mucolytics such as hypertonic saline as required (Statement 12).

Recently, a survey of people with PCD in the UK suggested that access to treatment for acute exacerbations, defined as per recent BEAT-PCD consensus statement [35], can be difficult for patients and their caregivers [36]. Urgent access to clinical review *via* the local team and/or the specialist service is therefore advisable in the event of acute illness. Treatment for pulmonary exacerbation should be supported by an individualised management plan that is established at annual review, and that can be enacted by general practitioners or local teams. Further assessments are required during and/or after treatment especially if there was no significant clinical response.

In CF and non-CF bronchiectasis, the importance of microbiological monitoring of respiratory secretions is well established. Pathogens such as *Pseudomonas aeruginosa* are associated with an increased burden of exacerbations and progression of lung disease [28, 37, 38]. Monitoring of respiratory samples is also relevant for individuals with PCD, as detailed in the BEAT-PCD consensus on infection prevention and control strategies in PCD [39]. The panel recommended to use sputum samples for all those who can expectorate both at annual review and at times of clinical instability, and induced sputum should be considered for those unable to expectorate (Statement 4). Access to bronchoscopy should be available for all those patients not responding to appropriate antibiotic treatment and should be considered in newly diagnosed younger children to assess infection and colonisation status. This is of relevance for identification of *P. aeruginosa* as recommended also for children and adolescents with bronchiectasis [22].

Although the most common pathogens isolated in respiratory samples of individuals with PCD are *Streptococcus pneumoniae*, *Haemophilus influenzae* and *Moraxella catarrhalis*, sputum should be processed using CF standards, by using various selective media to enhance the recovery of *P. aeruginosa* and other Gram-negative bacteria such as *Burkholderia cepacia* complex (BCC) [40]. Chronic colonisation with *P. aeruginosa* in PCD is associated with more extensive computed tomography (CT) radiological changes and lower baseline lung function, although causality has not been confirmed [40–43]. The recent isolations of BCC in individuals with PCD [44] confirms the importance of microbiological analysis as

done for processing CF sputum. While the American recommendations for monitoring of PCD suggest respiratory samples to be sent for non-tuberculous mycobacterium cultures only when individuals are not responding to treatment [19, 43], the panel recommended yearly assessment and as clinically indicated, especially in view of data supporting the use of preventative azithromycin in PCD [45], in line with the BEAT-PCD statement on infection control [39].

The panel suggested that imaging could be performed annually for surveillance in children *via* chest radiography and as required in adults, whereas a chest CT should be conducted if clinically indicated or upon entry into the service for adults. In view of the significant proportion of individuals with laterality defects and congenital heart disease, the panel proposed that individuals with PCD should have at least one abdominal ultrasound and echocardiogram in their records, unless a patient's PCD was known to be caused by a gene that does not cause laterality defects (Statements 5, 6 and 7).

Individuals with PCD can develop bronchiectasis, but also atelectasis, and have features of mucus impaction and mucus plugging, with middle and lower lobe predominance [46]. Often the parenchymal changes can occur before lung function is affected. Furthermore, chest radiographs are not as sensitive as CT imaging. Despite the use of significantly lower levels of radiation in modern CT scans, risk of exposure with cumulative dosing from CT scanning needs to be considered. Using CT scanning, a correlation was found between age and severity of lung parenchymal abnormalities, suggesting progression of structural changes over time [41, 47]. Further research is required to assess the evolution and progression of structural lung changes in people with PCD over time and to further develop and validate severity scores and grading scores specifically for PCD, to evaluate their potential as outcome measure in PCD [48].

Chronic rhinosinusitis and middle ear disease are a major cause of morbidity and affect the quality of life (QoL) of people with PCD [1, 2, 49]. Management, especially that of chronic otitis media with effusion, differs in individuals with PCD from those without PCD [50]. As such, the panel recommended that as part of the annual review process, each person with PCD should be offered review by a specialist ENT consultant with an interest in PCD. Audiology assessment should be performed at least annually in children as this could avoid speech delay, and as required in adults with PCD (Statement 2). Sino-nasal irrigation can alleviate the morbidity associated with chronic rhinosinusitis, as a relapse of symptoms frequently occurs after surgery. Specific evidence is, however, lacking.

Sperm flagellum and motile cilia in the reproductive tract can be affected in people with PCD. Hence, people with PCD might suffer with infertility and subfertility, and women might be at higher risk of ectopic pregnancies [51, 52]. A recent survey among individuals with PCD in Europe highlighted that fertility is a topic of importance and interest for people with PCD, but only half of the respondents received care from a fertility specialist with a minority being referred by their PCD team [53]. To overcome this and in the interest of people with PCD, the panel suggested that adults with PCD should have local access to genetic counselling and fertility services. Furthermore, in the same survey, patients were often not satisfied with the information they received on fertility and expressed the need for a more comprehensive discussion [54]. While research is ongoing in developing links between specific genetic variants, ultrastructural defects and fertility in individuals with PCD, the panel suggested that fertility should always be discussed during transition to adult care (Statement 15).

To achieve this, the panel also recommended that a structured transition programme should be implemented with regular meetings between teams for continuity of care and support of patients. Adolescents and young adults are at a key and critical point in their life and those living with a chronic health condition experience the general issues of entering adulthood intertwined with the need to take and then maintain responsibility of their own disease and care. Planning of transfer to adult care should be done in partnership with the adolescent individual, who should have the opportunity to meet the adult team ahead of transfer of care. The standardised "ready, steady, go" transition programme is recommended [55] (Statement 14).

People with PCD might struggle not only due to physical symptoms of the condition, but also because of the implications of such symptoms and their diagnosis on their QoL. Previous studies [9, 15] highlighted that some experience frustration with the lack of knowledge of PCD and face a personal dilemma on whether to disclose and how to explain their condition due to the risk of being stigmatised. Some patients conceal their symptoms of PCD to avoid embarrassment [56]. It appears that while QoL worsens with age, symptoms impact it even in childhood and teens with PCD and the diagnosis also negatively impacts their parent [49, 55, 56]. This together with the impact of having a lifelong condition can affect QoL and mental health in individuals with PCD and a recent survey of patients in the UK highlighted difficulties in accessing psychology services as a main issue of the service [36, 57]. Despite not formalising it in a statement, the

panel suggested that patients with PCD should have an assessment of QoL, with consideration of an objective tool as part of this assessment and consider referral to psychology services as required.

Participation in research, both prospective and retrospective, as well as in national and international registries is recommended to allow for further benchmarking and improvement of quality of services. This latter point leads to a further role of management services in promoting knowledge of PCD to patients and families but also to other healthcare professionals, to raise awareness of the condition, to reduce time to diagnosis and to emphasise the potential severity of the condition. The registries will also help in defining long-term longitudinal outcomes for people with PCD.

### *Limitations and strengths*

This document aims to give a comprehensive overview, discussing the main aspects of monitoring and care for individuals with PCD. However, the literature search that supported development of statements and panel discussion has been completed as a narrative review and is not fully structured as in a systematic review.

To ensure adequate representation and consideration of topics that might be relevant in specific ages (*i.e.* referral to fertility care, genetic counselling for adults and regular audiology assessment to avoid delay in speech), the panel consisted of an equal number of adult and paediatric respiratory clinicians all with a significant expertise in the care of people with PCD. Patients' representatives from PCD Support UK and from each centre also contributed to the development and review of the statements to guarantee a patient-centred approach. However, all panel members work in England within the established highly specialised services for PCD, thus limiting the generalisability of the document.

As this consensus statement aimed at creating a framework for the day-to-day management of individuals with PCD, it focused mostly on the medical aspects of management. However, as concordance with treatment and optimisation of care is strictly linked with patient education and engagement, the lack of a statement addressing this aspect is a limitation of our work, and should be considered in a future revision of the statements.

### *Conclusion*

It is only in recent years that the provision of care for patients with PCD has been organised into specialist paediatric and adult management services in England. Overall, this consensus statement builds on previous documents on standards for diagnosis and care for PCD [58], which were developed before the PCD management services were established in England. With the further understanding of the complexity in the diagnosis, correlations of genotype and phenotype and the potential of clinical trials becoming available, the panel and patients' representative in England felt the need to ensure harmonisation of care across the country. Our aim with this statement is to set out standards for the provision of care for individuals with PCD. This will encourage consistency in practice and enable a benchmark for future provision.

Provenance: Submitted article, peer reviewed.

Conflict of interest: G. Spoletini is an associate editor of this journal. S.B. Carr has received NIHR grants (HTA, RfPB and Programme Development grants as co-investigator). K. Dexter is the unpaid chair of PCD Support UK. L. Dixon is the unpaid vice-chair of PCD Support UK. C. Hogg received consultancy fee from ReCode Therapeutics. A. Jones has received speakers' honoraria from Vertex Pharmaceuticals. M. Loebinger received consultancy fees from Armata, 30T, AstraZeneca, Parion, Insmed, Chiesi, Zambon, Electromed, Recode, Boehringer Ingelheim, Ethris, Mannkind and AN2 Therapeutics. J.S. Lucas is an unpaid medical advisor for PCD Support UK, on the Scientific Advisory Board for PCD Research, is Work Package Lead for BEAT-PCD ERS CRC and a Trustee for Allergy, Asthma and Immunology Research (AAIR) Charity; has received grants from AAIR Charity, LifeArc, Medical Research Council, National Institute for Health Research and Great Ormond Street Hospital Children's Charity; and has received funding for associated research to her institution from LifeArc Translational Rare Respiratory Disease Centre for which she is Co-Leader. D. Patel is an unpaid medical advisor for PCD Support UK and is part of the BEAT-PCD patient engagement work package group. S. Range is an unpaid medical advisor for PCD Support UK. The other authors have no conflicts of interest to disclose in relation to the present manuscript.

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