Experiences of parents whose young child has been diagnosed with primary ciliary dyskinesia

Corine Driessens1, Siohan Carr1, Edel Clough1, Fiona Copeland1, Sharon Dell1, Lucy Dixon1, Amanda Harris1, Rebecca Knibb1, Margaret Leigh1, Manjith Narayanan1, Beatrice Redfern1, Evie Robson1, Michael Sawras1, Lynne Schofield1, Kelli Sullivan1, Myra Tipping1, Nhu Tran1, Woolf Walker1, Jane S Lucas1, and Laura Behan2

1Affiliation not available
2University of Southampton

February 24, 2021

Abstract

Primary ciliary dyskinesia is an incurable, rare, inherited, chronic condition. Treatment includes regular clearing of airway mucus, aggressive treatment of infections and management of hearing loss. Caregiver burden has not been explored, hence we interviewed 18 English speaking mothers and 6 English speaking fathers of children under 6 years who were diagnosed with PCD around the world. The parents described how the child’s diagnosis, treatment regimen, and health status impacted their life. They discussed the impact of the COVID-19 pandemic and they talked about the different ways they cope with challenges that arise. The need for integrated social care is discussed.

Experiences of parents whose young child has been diagnosed with primary ciliary dyskinesia by

Corine Driessens1,2,3
Siobhan Carr5
Edel Clough8
Fiona Copeland8
Sharon Dell6
Lucy Dixon8
Amanda Harris1,3
Rebecca Knibb4
Margaret Leigh9
Manjith Narayanan11
Beatrice Redfern8
Evie Robson10
Michael Sawras7
Lynne Schofield10
Kelli Sullivan¹²
Myra Tipping⁸
Nhu Tran⁸
Woolf Walker³
Jane S. Lucas¹,³ ORCID ID: 0000-0001-8701-9975
Laura Behan¹

1. School of Clinical and Experimental Sciences, University of Southampton Faculty of Medicine, Southampton UK
2. NIHR Applied Research Collaboration Wessex, University of Southampton, Southampton, UK
3. Primary Ciliary Dyskinesia Centre, University Hospital Southampton NHS Foundation Trust, Southampton UK.
4. Department of Psychology, School of Life and Health Sciences, Aston University
5. Primary Ciliary Dyskinesia Centre, Royal Brompton & Harefield Hospital NHS Foundation Trust, London UK
6. Department of Paediatrics, University of British Columbia, CA
7. Hospital for Sick Children (SICKKIDS), University of Toronto, CA
8. PCD Family Support Group, UK
9. Department of Paediatrics, University of North Carolina at Chapel Hill, USA
10. Primary Ciliary Dyskinesia Centre, Leeds Children’s Hospital at Leeds Teaching Hospitals NHS Foundation Trust, UK
11. Primary Ciliary Dyskinesia Centre, University Hospitals of Leicester NHS Foundation Trust, UK
12. Department of Medicine, University of North Carolina, Marsico Lung Institute, Chapel Hill, North Carolina.

Competing interest
On behalf of all authors, the corresponding author states that there are no competing interests to declare.

Abstract
Primary ciliary dyskinesia is an incurable, rare, inherited, chronic condition. Treatment includes regular clearing of airway mucus, aggressive treatment of infections and management of hearing loss. Caregiver burden has not been explored, hence we interviewed 18 English speaking mothers and 6 English speaking fathers of children under 6 years who were diagnosed with PCD around the world. The parents described how the child’s diagnosis, treatment regimen, and health status impacted their life. They discussed the impact of the COVID-19 pandemic and they talked about the different ways they cope with challenges that arise. The need for integrated social care is discussed.

Keywords: primary ciliary dyskinesia, caregiver burden, chronic illness, preschool, COVID-19, Quality of Life

Introduction
Primary ciliary dyskinesia is a rare (1:10,000) genetic lung disease caused by impaired clearance of mucus and debris from the airways. Individuals with PCD often present with unexplained neonatal symptoms often, requiring respiratory support (Mullowney et al., 2014). They continue to have persistent lung and nasal symptoms in infancy, with frequent infections. Progressive chest symptoms persist throughout life and include daily wet cough and recurrent chest infections almost invariably leading to irreversible bronchiectasis by adulthood (Lucas et al., 2019). Cilia do not clear fluid from the middle ear causing frequent ear infections, hearing impairment, delayed speech and learning. Dysfunction of the embryonic nodal cilia causes situs inversus (mirror image positioning of organs in the body) in 50% of cases, sometimes associated with congenital heart disease (Best et al., 2019).
Despite symptoms from birth diagnosis is often delayed, primarily because of lack of awareness amongst clinicians (Behan et al., 2016). Major efforts to improve knowledge of paediatricians has reduced the average age of diagnosis from 6 years in 2009 to 2.6 years in 2015 (Kuehni et al., 2010; Rubbo et al., 2020). There is no cure, and management aims to control symptoms, improve hearing, treat infections and delay lung damage (Lucas et al., 2017a). Most importantly, PCD patients need to adhere to daily time-consuming treatment airway clearance regimens aimed at facilitating mucus clearance (Schofield et al., 2018).

From birth, the parents of children living with PCD are faced with the challenges of caring for a child with a (un)diagnosed chronic condition. Parents not only assist the child in activities of daily living, but also accommodate the nutritional, physical, social, emotional, medical and financial needs associated with the chronic health condition. This has been shown to differentially impact parental wellbeing. Pelentsov et al. (2016) for instance describes that parents of a child with a rare chronic condition felt socially isolated as others did not understand what the parent was going through. Many mothers reported that caring for a child with a chronic condition required adjustment in their employment. Germeni et al. (2018) found that parents of children with chronic conditions try to reconstruct some form of normality once diagnosis has been made. The stability of this normality depends heavily on the fluctuations in health status of the child with the chronic health condition and ability to control these fluctuations (Dudeney et al., 2017).

To date, the impact of child’s PCD on parental wellbeing has not been well documented. A previous study has indicated that a small group of Italian mothers of children (age 6 to 16 years) living with PCD experienced more distress than mothers of children who did not have a chronic health problem (Carotenuto et al., 2013). Another study found that Turkish parents to children (age 6 to 16 years) diagnosed with cystic fibrosis experienced significantly more caregiver burden than a small group of parents to children diagnosed with PCD (Keniş-Coşkun et al., 2019). As so little knowledge exists, we conducted this qualitative study to explore the impact of caring for a young child diagnosed with PCD on English speaking parents from around the world.

Methods

The study is reported according to the Consolidated criteria for Reporting Qualitative health research (COREQ) (Tong et al. 2007). The study received ethical approval from the Southampton and South West Hants Research Ethics Committee (06/Q1702/10; University of Southampton ERGO#53155), University of North Carolina Internal Review Board (IRB#09-1099), and Hospital for Sick Toronto Children Research Ethics Board (REB#1000024263).

Recruitment

English speaking parents of young children (<6yrs old) who had received a diagnosis of PCD according to international guidelines (Lucas et al., 2017b), were recruited by PCD specialist’s team from three PCD centres in England (Southampton, London, Leeds) and two PCD reference centres in North America (Chapel Hill, USA; Toronto, Canada) between November 2019 and November 2020. In addition, recruitment advertisements were circulated among the members of the PCD Support Group UK. Clinical teams recruited twenty-one parents, and three members of the PCD Support Group agreed to participate. This method of convenience sampling was used as PCD is a rare disorder. If both parents agreed to participate in the study, they were interviewed separately, except in one case where the parents were interviewed together.

Participants

Eighteen mothers and six fathers of 20 children participated. Detailed information on the child is provided in table 1. Most parents resided in the UK (n=16), 3 in Canada, 2 in USA, 1 in Georgia, 1 in Ireland, and 1 parent in the Netherlands. Four parents completed secondary education, 3 completed further education, 6 completed professional training, and 11 completed higher education. Fourteen of the parents were part-time or fulltime employed, 2 of the parents were self-employed. Five of the parents self-identified as homemakers and 3 as unemployed.

**TABLE 1 INSERT HERE**
Data collection

Following consent, semi-structured interviews were conducted with participants over phone or via videoconferencing. Interviews followed an interview guide exploring experiences around birth, child development and progression symptoms, healthcare needs child, impact child’s PCD on parent and thoughts about the future. The interview guide was developed by CD after a literature review and refined by a panel PCD clinicians and patient representatives (Appendix 1). CD, a psychologist experienced in mixed research methods, conducted most of the interviews. LB, experienced in developing Quality of Life scales for individuals living with PCD (Lucas et al., 2015; Behan et al., 2017; Behan et al., 2019) and trained as a qualitative interviewer, conducted 3 interviews and validated thematic coding. Both interviewers were not known to any of the participants. Interviews with participants ceased when no new information emerged during the conversations. Data saturation was assessed periodically by CD and LB. All interviews were digitally recorded and transcribed verbatim. Interview length ranged from 30 minutes to 125 minutes with most interviews lasting between 45 and 60 minutes.

Data analysis

The transcripts were analysed using nVivo 12.0 (QSR International, 1999). An inductive approach using reflexive thematic analysis was adopted (Braun and Clarke, 2019). The first 10 interviews were coded independently by CD and LB. Themes were identified inductively and similar subthemes were grouped together under an overarching thematic framework (Supplementary material). Initial coding was cross-compared and a preliminary thematic framework was agreed upon. The remaining interviews were analysed by CD. The thematic framework was further refined by findings of the subsequent interviews and through discussion with the research team (see table 2).

“TABLE 2 INSERT HERE”

Results

This paper concentrates on the impact of PCD on the parents. Five themes created an understanding of the parent’s experiences: Impact of child’s PCD diagnosis on parent; Impact of child’s treatment regimen on parent; Impact of child’s health status on parent; parent’s coping strategies; parental concerns for the future. Under these overarching themes we identified several subthemes.

Impact of child’s PCD diagnosis on parent

For those children with situs inversus, the abnormal organ arrangement was often picked up at the 20-week pregnancy scan. Parents described that once this was identified they were exposed to multiple prenatal tests and had to have regular appointments. Parents spoke of this as being a very difficult time:

“in antenatal stage at my 20-week scan, where you know I was going there really excited to find out what, am I going to have a boy or a girl. Half-way through the scan the lady who was doing the scan kind of went quiet and then said that she needs to get her colleague. . . . and I just straight away knew there was something wrong. The other lady came in and she did say that she can’t see his stomach. They got me an appointment the following day to see a consultant and then we discovered that his stomach is placed on the other side. So, through the antenatal stage I had lots of scans, I went to see the doctor, cardiologist to check his heart. Then I had a c section . . . . it was just horrendous. . . . they took him out, I had him with me just a few seconds and they take him to intensive care unit because they had to check him . . . . I couldn’t see him for like 5 hours, I didn’t know what was going on.” (mother 15)

Parents credited the identification of abnormal situs to an earlier diagnosis for their child. Some parents remembered PCD being mentioned as a possibility during their pregnancy or shortly after.

“So, that then triggered lots of things put in place for as soon as he was born. So we had like heart tests and things like that and obviously one of the things that they tested for, was PCD. . . . So it was just really quick. Like we were kind of aware that it was something that he might have even before, I think, I don’t know if PCD was mentioned before he was born, but pretty soon afterwards.” (mother 4)
All parents noticed the child’s symptoms at birth, yet only 11 of the 20 children were diagnosed at this time. During the pre-diagnostic period, parents had no knowledge of how to support and care for their child; when reflecting on this, parents recollected their helplessness and emotional distress.

“No body ever really knew what it was, so I think that was the hardest for us, those four years of just what could it be? Could it be something you know life threatening? Who knows what it could be. . . . . before he was diagnosed, I was always worried and sometimes I would put it in the back of my mind for a few weeks and not think much of it until he got like a really bad cold . . . . we were always worried because we knew something was wrong.” (mother 10)

A considerable number of parents reported frustration and feeling dismissed when raising the issue of their child’s symptoms to medical experts or to their extended family and friends; many were told their child would probably grow out of their symptoms:

“And every doctor we went to was like ‘he will grow out of it’ or ‘it is probably reflux.’ He was on reflux medication, didn’t help. They always had some sort of excuse, they used to just shrug it off.” (mother 10)

These experiences differed from parents who already had experience of PCD. In the following narrative a mother who already had an older child with PCD describes the emotional impact of the pre-diagnostic period of her older child compared of her youngest child.

“when our first born was diagnosed, I mean oh my gosh, it was horrific. I was petrified, I was scared, I didn't know what was going on, we didn't have a clue what we were facing, didn't have a clue what they were testing her for, so it was very much a case of we were scared. Whereas with our current child we knew exactly what they were testing him for, we were 99% sure that he had it (PCD), so actually it was more sadness that our thoughts, our worse fears kind of being confirmed I guess.” (mother 3)

Once the child was diagnosed, most parents felt mixed emotions. They had feelings of relief now having a name for their child’s condition mixed with feelings of sadness because the child had a life-long condition that always needed to be managed.

“we were relieved and sad at the same time. Because PCD, it’s something he will have for the rest of his life, and something he will have to deal with for the rest of his life. You know, we were sad that he did have a condition that would affect him for the rest of his life. . . . . we were happy because now he is being treated, so now we can actually get things with his diagnosis so we are relieved that we have one.” (mother 10)

Many parents did not process their child’s PCD diagnosis immediately, but just focused on dealing with daily challenges. These parents described getting overwhelmed when faced with a major challenge.

“I think the hardest thing we ever found to deal with was the hearing aids. . . . . Which was just because we’d coped so far, like he was 18 months when he had hearing aids in and I think that, at that point we were like, actually it’s ok to not feel ok about this. Whereas up until that point we were like ok, yeah, we can do this, we can do this!” (mother 4)

Impact child’s treatment regimen on parent

Parents caring for their first child with PCD described feelings of worry and stress, having responsibility for PCD management. Treatment regimens were described as overwhelming particularly in the early stages of diagnosis but became easier over time.

“when we started treatment yes, it was like a big whole day, I was either preparing or giving a treatment, so there was nothing I could do, no cleaning, no dinner, nothing. And it was like how, can I do this like for ever and this was really hard. But later, I was little faster and now it is like everything is getting better.” (mother 8)

Parents of children diagnosed with PCD changed their daily schedule to accommodate the child’s daily PCD treatment regimen. Most parents provided the child with 1 to 3 daily airway clearance (physiotherapy treatment) sessions totalling 20 to 360 minutes each day. For most parents it took 1 to 2 years to settle
into a lifestyle that accommodated the medical needs of their child. While some parents shared the daily treatment burden, for most couples it was the mother who took responsibility. Parents scheduled or planned each day to accommodate the daily treatment or they developed a new daily routine. One mother described:

“living with PCD it’s a lifestyle really, it’s something that we fit it all into the daily routine.” (mother 1)

Parents described having to wake up earlier in the morning and return from work or family excursions earlier in the evening to support their child with the PCD treatment.

“... it has to be timed very well and done in a specific order... we can’t just do things spontaneously, you know like a family with children with, who don’t have the condition, you know if they want to change it up at the last minute, they can do that, whereas we couldn’t, if we went out for a certain period time for example, I couldn’t just say, yeah let’s just stay it’s fine we’ll just stay later, because it might be that I don’t have a certain medication on me” (mother 3)

Parents generally prioritised treatment over any other activity. At this young age the child needed parental supervision to adhere to the daily treatment. The child didn’t always sit still for treatment and adherence could be challenging for parents. Some parents used a role model (older sibling, parent) to convince their children to do daily treatment. Some parents explained to their child that the treatment will keep the germs out and will make them feel better. For most parents repeated reminders, chasing, struggles, and restraining were commonplace. Other parents did not make a big fuss but handled it matter of fact as this father explains.

“he does have this aerobika thing that he does after the nebuliser, so it’s like, it’s this whole process, he does it twice a day but he’s used to it, we don’t make a big fuss about it, .......and then antibiotic, but he doesn’t know what it is, what it does. He just knows he has to take it.” (father 4)

Parents also cuddled with their child or used other methods to calm the child so the daily treatment would not upset them. Various parents bribed their child to do daily treatment, but most parents used electronic distraction to encourage adherence to daily treatment.

“I mean we always put him in front of the telly and do and try and to make it like a nice thing, rather than something boring, so you know it’s part, his bedtime routine it works brilliantly because it’s like 10 minutes of chill out calm before stories and bed, so that works really well, but in the morning he just wants to play now, he doesn’t want to sit down or do it, so he does do it, but it’s bribed with you can have this at the end of it (Laughter) anything to get him to do it.” (mother 4)

A number of parents felt unsure whether they were doing the daily treatment correctly and several found it hard and emotional to enforce treatment when the child was upset.

“it is about emotionally as well, it wasn’t really comforting ... she is looking at you and you are actually giving her physio and you want her to have the head mask as well and you know she doesn’t know what is happening.....to be honest that was actually really really difficult. ......... she was getting very very edgy, like she was crying and really upset and we were supposed to keep her there for like 5-10 minutes, I mean, depending on how we feel, like if she is coughing or not.” (mother 2)

The parents described their frustration at the continuity of the condition and treatments. They explained how important it is for their child to do the treatment every day in order to stay well. They felt time pressured in the morning to complete the PCD treatment before they did any other activities. The also described not being able to follow their own interests or schedule alone time.

“it’s harder for me in this aspect, to not to be able to go whenever you want ... the thing for me is that I don’t have enough time to do everything what I want to do...... I am more at home, at home and taking care of my kids, that’s what changed I guess. I’m not going out as much as I used to” (mother 8)

As PCD is a rare disease, parents often spoke of their difficulty when dealing with medical professionals who don’t have an awareness of PCD. Children with PCD are often prescribed antibiotics to treat repeated ear
and lung infections. Parents discussed challenges they experienced in getting antibiotic prescription from a GP or filled by the pharmacy.

“to get a new prescription, and if that’s something different I have to explain again, and then sometimes they need to call the specialist. . . . sometimes the doses is different than they used to give, it’s higher . . . sometimes the treatments are for four weeks instead of one and they’re like ‘we need to ask questions’, I understand that, but sometimes I get so tired of explaining myself again.” (mother 13)

A few parents worried about the side effects of antibiotics on their child’s health and they feared that antibiotics would stop working in the long run as their child is taking them quite often. Parents also faced issues where their child became upset at having to take antibiotics due to the ‘nasty taste’.

“one of the things which is quite difficult is to get to take his antibiotics. . . . as he’s got older that’s become more tricky and we’ve had to hide it in food. Because sometimes you’ve got to give him this stuff 3 times a day and it’s not particularly flavoured nicely, so he’s very aware that this is medicine that he’s having and he doesn’t want to have it so, um obviously that’s something that is, you can overcome it but sometimes it’s quite tricky.” (father 2)

In some healthcare settings children diagnosed with PCD attended a one-stop multidisciplinary clinic once every 3 months for review by a PCD respiratory consultant, respiratory physiotherapist, PCD nurse specialist, and sometimes a psychologist, dietician, and/or Ear-Nose-Throat consultant. In countries with fragmented PCD care, the parent was responsible for coordinating all appointments and communication with medical specialists needed for their child’s wellbeing.

Most parents valued an update on the medical status of their child and felt involved in the medical decision making regarding their child. Parents described their medical team as family and felt they could contact their medical team whenever they had questions or issues.

*They have been so awesome and I feel so close to all of them. They are almost like family now (laughs) . . . .I can’t say anything negative about them. I feel included in every decision.* (Mother 13)

In some instances, parents did not feel supported by the PCD healthcare team. Some experienced language barriers, unclear information, a lack of practical support, felt not listened to, or the parent felt powerless to refuse required PCD healthcare. The appointments were time consuming, sometimes requiring the parent and child to be at the hospital for several hours. Working parents often needed to claim a holiday or took unpaid leave to accompany their child to these appointments. For a few parents it was quite emotional to attend clinic. As this mother explained:

“I find it quite difficult because it is a reminder I think of how serious his condition is and I struggle with going . . . . it would just bring up a lot of emotions and I’d get really upset and I think I get really fearful even though they are a really . . . .they’re really supportive . . . . having a whole team of people was frightened me at first and . . . .um . . . .it . . . because it made everything so . . . .I mean it was very serious to me anyway . . . .but it just . . . made it even more serious . . . .and more scary but . . . .I knew that they were there for . . . .for a good reason, I did not want my son to have all this . . . .need, all these people” (mother 7)

It could also be quite stressful due to the emotional/behavioural response of the child. As mentioned by this mother, parents needed to prepare their child for the visit and often rewarded them for compliant behaviour:

“sometimes when I say ‘hospital’ he thinks he’s being hospitalised, and that’s something in his mindset. If he says something like that, it makes me a bit sad. I have to explain to him which hospital it is, it’s really important to prepare him for that. Yeah, to prevent that he . . . he thinks he has to be hospitalised . . . . I have to reward him afterwards. And we try to make it as positive as possible” (mother 13)

The parents tried to stay calm when they saw their children struggle with the procedures and did not like to restrain their child as this father explained:

“The hard bits I suppose are when (child’s name) has to undergo medical bits and pieces like nasal brushings
and things that he doesn't want to do because either it hurts or it's just an invasion of his privacy” (father 2)

The worst time for parents was when their child was hospitalized. They described it as a draining time, full of emotions and anxiety where they felt out of control. Not being able to be together as a family and care for any additional children had an emotional impact on the parent.

“It’s horrible, it’s the worst time... I used to think it was hard when (child) ended up in hospital and at the time that was like the end of the world. You know, we had one child and that child was in hospital and you worried about that child and it was just awful. But actually, now having 2 with it and one without – having 3 children, what actually now has materialised is hideous, as a mother having to pick between your children, having to choose between them.” (Mother 3)

Impact child’s health status on parent

In general, parents did not anticipate their child being born with health problems when they decided to start a family. Now that they were living with a child with a long-term health condition, they sometimes wished the child did not have PCD, were wondering why this happened to them or wished that there was a cure. Emotionally, the worries about PCD never left some parents. For some parents PCD worries were in the back of their mind surfacing as soon as the child was feeling unwell, the child needed to visit a doctor or therapist, or they received PCD related correspondence. Parents wanted their child to live as normal a life as possible as explained by the following mother:

“I want my children to experience as normal a life as they possibly can and so for me, that includes travelling. So you know, yes we take those risks and we just go for it! And we do it because, you know I don’t want to have the regret of not having done it.” (mother 3)

For other parents the PCD worries were with them every day. Always being on high alert can be overwhelming. These parents felt worn out and overrun by PCD.

“I do feel I’m quite stressed, I’m quite run down. I just feel everything’s sort of on top of me at the moment .... it’s just constant, sort of appointments and phone calls and ordering her medication and chasing it up and having to call this person and that person and so on and so saying they don’t know, then try this, it just goes round in circles sometimes, so I do get a bit frustrated and angry” (mother 14)

This distress had led to sleep deprivation, stress, and feelings of unhappiness, sadness, depression, and anxiety. Five of 10 parents reporting mental health symptoms received a clinical diagnosis and/or treatment for depression, anxiety. One parent reported that her physical health was affected, she now received treatment for stress-induced high blood pressure.

On the practical side the parents did anything they believed would keep their child well such as being vigilant not to expose the child to infections, behaving more cautiously around other people, assessing the child’s health condition during the day as well as night, assessing the risk level of each activity, ensuring the child attended medical appointments, making sure the child had tissues to wipe their running nose, and prolonged breastfeeding. They were on constant alert to make sure the child did not develop a cold or became unwell. A few parents even checked their child with a stethoscope or oximeter on a daily basis, various parents regularly measured the child’s temperature, other parents kept a diary/record, or closely monitored the child’s symptoms. The parents mentioned a variety of emotions when child was unwell including that they were worried, scared, anxious, upset, or sad.

As the following mother described some children with PCD can become unwell very quickly:

“With her it comes on so sudden, so you don’t really notice a change until it’s too late and you need to get her into the hospital on to oxygen. Because she could be fine, the last time we had it, she went to bed, she’d been at nursery all day, she had her tea, she went to bed, she was fine – I noticed she was a bit wheezy, so I gave her blue inhaler, I waited didn’t have any impact on her, gave it to her again and she was just getting
worse and worse and worse. It was just so sudden like within a couple of hours we were having to call an ambulance out.” (mother 11)

Hospitalizations were described as being horrific and overwhelming; it made the parents feel out of control. The following mother disclosed:

“I hate it, it makes me cry every time. Because it just takes me back to when she was little when the first time we went into hospital and we were in there for 3 weeks. And it felt like we were never going to get out.” (mother 11)

Most couples had to adjust work commitments once they started caring for a child with PCD. One of the partners usually stopped employment, started working from home, became self-employed, minimalized work hours, and/or took a less demanding vocational role. The other parent was no longer able to make overtime and needed to take holidays or unpaid leave to accompany the child to medical appointments. Losing this additional income financially affected most families living with a young child diagnosed with PCD. The couple felt that one of them needed to have the flexibility to attend to the child’s daily and emergency health needs. The following mother explained why she started working part-time:

“I work part time, I work 2 days a week, because after my daughter got diagnosed with PCD it was just too much I just couldn’t cope working any more than 2 days, um I just was really run down and tired and taking too much time off. I wasn’t sleeping well, I had to go down doing 2 days. Even so I do 2 days, I’m still struggling, I’m just thinking I might have to give up work, but financially we can’t cope to be honest if I did sort of give up work.” (mother 14)

Another mother explained that she had to stop working out of fear her children would pick up illnesses in other people’s care and how this had a major impact on her and her own life expectations.

“I stopped working so I could be here so they wouldn’t be at day care and wouldn’t get exposed to so many germs. ........ you know I gave up my career so that I could better take care of the kids. I don’t work anymore. So this is the worst thing for me, if we are talking about me as a mother. I always thought that I would have a career and a family too, close together and right now I can only I don’t have any career right now and I studied a lot and when I meet my old friends, you know from university or from school, they are like, oh my god, I don’t believe you don’t work! This is what everybody expected or I was expecting from myself.” (mother 8)

The child’s genetic diagnosis prompted the parents to reflect on family goals and some parents discussed their decision not to have any more children.

“we always thought we have 2 children, but after having a child with PCD we decided not to have another child because we did not want to risk that we would pass the ...the faulty genes on again and have another child with PCD. ...um I think very much ...not so much from a selfish point of view as in we don’t think we’ll be able to cope with that...uhm...but we felt that it was not fair to risk having another child and passing on the condition again” (mother 5)

Socially many parents felt their child’s PCD care did not leave any room for spontaneity or time for themselves. They did not get a break, were unable to go out much. The PCD care interfered with their ability to see friends/colleagues/family and PCD limitations had changed family activities. They no longer had time for hobbies or even time to spend on their appearance. Although the PCD diagnosis of the child had placed a strain on many marriages, most couples had been able to work through this and came out the stronger for it.

“I think, for me and my husband, we’re a pretty good team actually. He, generally, in our general kind of marriage, we parent very similarly, but obviously it adds a bigger impact on us, on our marriage for sure because we’re making decisions that perhaps other parents don’t actually have to make. So it definitely puts a bigger impact on our marriage, but generally we’re pretty good at dealing with that.” (mother 3)
In families with other children without PCD, parents sometimes believed these children lacked parental attention.

“I know it has had a big impact on my 2 other children, because of this condition that my child has PCD, you know. Because they can see I give her more time and I don’t have enough time for them and even if I do, I’m just too tired that my attention is not with them, especially when it comes to homework and just other stuff, you know cos I’m just physically run down and tired doing all the chest physio and stuff like that. I get tired myself, you know what I mean.” (mother 14)

Parents received support from friends and family, however reported feeling that nobody understood exactly what they are going through. The following mother explained:

“. . . maybe they try to understand our position, but do they really get it? No, absolutely not! I don’t think, that’s a slight on anybody else, that’s just unless you’re living it, I guess you can’t really understand it. No more that I could understand somebody else’s position with perhaps a different condition.” (mother 3)

Parent’s coping strategies

There was wide variation in the way parents coped with the challenges of caring for a child with PCD. We have classified the different styles of coping into support seeking, problem-focused, emotional-focused, and cognitive-adaptive coping strategies. Some parents discussed how support from partner, family, healthcare providers, online PCD support groups had enabled them to cope when child was hospitalized or unwell. Some parents sought medical information to explain why their child was unwell while other parents needed to talk to somebody who was willing to just listen.

Several parents dealt with the challenges of raising a child with PCD pragmatically, by taking things day-by-day, and focusing on the problems at hand. They became knowledgeable about PCD and sometimes learnt new skills to take control, as highlighted by the following mother:

“T’m actually qualified now, so I trained to do home IVs, so I can actually administer once they have a PICC line fitted and the consultant decides they’re ok to be in my care at home. I actually bring them home and I do the IV myself because again it’s given us the ability to back together as a family and I think that’s the biggest care – sometimes not being able to be together as a family.” (mother 3)

Some parents tried to forget their worries and supressed negative emotions that surfaced due to the challenges of raising a child with PCD, for example by crying, exercising, keeping busy, thinking positive thoughts, having fun and laughing a lot. Some found themselves taking prescribed mental health medication, whereas other parents positively re-assessed the situation so it no longer induced negative emotional responses through techniques such as praying, not thinking about PCD, wishing for the miracle of a cure, or knowing that today is a bad day but hoping that tomorrow might be a good day. The most popular form of adaptive cognitive coping was the parent comparing their situation with somebody who was worse off. As this father described:

“I currently have in my team at work somebody who’s having a baby who’s got a tumour on her brain and it’s about viability of the baby, so when you think about what could be happening and that fact that my son has wonderful life and is very happy and he actually talks fine and he’s you know got a zest for life, he’s enthusiastic and he’s running around actually compared to perhaps what other people are going through, this really isn’t as big a deal as it could have been.” (father 2)

Parental concerns for the future

Parents who were thinking about the future mentioned their anticipation of explaining PCD to their child when they are old enough to understand.

“I feel confident in terms that we’re doing the right thing at the right stage of his life at the moment. I feel nervous that we’ll get it wrong later on, especially as we have to start explaining things in more detail to him and I worry what his reaction with be.” (father 2)
The expected social impact of PCD symptoms and the child’s potential emotional response are well described in these two narratives:

“Currently my son is very confident, but kids can be very cruel and I think if he’s going to continue speaking very, very unclearly, they might pick up on him. . . . I don’t know, but you know simple things like, he can’t say finish, he’s saying pinish, and you know for 4½ its still quite cute, but when he’s 6, 8 or 10, it’s not going to be cute any more, it will become an issue for him” (mother 15)

“I think he will be more affected by it when other children get annoyed by it, so like he coughs a lot, you know and I can just imagine him sitting in class coughing and someone’s going “ooh, that’s not very nice”. So, it’s that, that we need to be like teaching him how to do it nicely, and make sure he blows his nose, and all of those things, just to support the social aspect of it. I guess it’s that side of it that needs to be carefully managed and supported as well because otherwise he will be physically fine but mentally not, so . . . we need him to be able to cope with that side of it.” (mother 4)

Another concern that was frequently mentioned was progression of disease severity. Some parents worried that lung damage might lead to lung transplant, progression of hearing loss, and even PCD cutting their child’s life short. Currently, parents were in control of PCD treatment and adherence, but some worried whether their child would continue to manage their treatments adequately when older.

“my child will become more independent with treatments and stuff and we’re helping her to achieve that, but that’s quite tough to achieve that, because you want them to have the best possible treatment and in my eyes, the best possible treatment is me being in control of that treatment. . . . So that concerns me. Have I brought her up in the best possible way I can for her to really comprehend how serious this condition is and that PCD if it’s well maintained. . . . she can live. . . as normal a life as she allows herself to live, by the treatment she continues to do?” (mother 3)

Many parents thought about how PCD might limit the child’s social activities, development, and career goals.

“it’s going to have a big impact in her daily life, daily routine, because she will have to be home to do her therapy and stuff like that. She won’t be able to do all the normal things her friends do.” (mother 14)

The most frequently raised concern for the future was fertility. As discussed by one mother, parents worried how fertility treatment would impact their child and whether the grandchildren would inherit this genetic disorder.

“. . . what I’m really concerned about also, is whether he’ll be able to have children one day. Because, I know the kind of science is moving in the right direction and there might be some further development, but from what I understand for males with PCD to actually conceive the child naturally it can be difficult. So he might need some extra help and I know that for some males that might be an issue, they won’t be an alpha male. Being able to fully, not just perform, but what I’m saying is being able to do all those things that they are kind of natural to male, and it might affect his confidence and self-esteem and mental health in the future.” (mother 15)

COVID-19 pandemic and caring for a young child with PCD

Three months into this project the COVID-19 pandemic started. As we wanted to focus on everyday experiences of parents caring for a young child with PCD, our data collection stopped from March to July 2020, the first wave in the COVID-19 pandemic in the UK. COVID-19 was rarely reported among children diagnosed with PCD (Pederson et al., submitted) and as the world tried to return to some form of normality, the data collection for this study resumed from August to December 2020. The interviews did not focus on the effect of COVID-19 on caring for a child with PCD, but parents spontaneously brought up the impact COVID-19.

Parents discussed how anxiety-inducing it is caring for a young child with PCD due to COVID-19. One mother described how she worried about her child going to school and that he wasn’t shielded anymore:
"I think also, with COVID-19, that’s a completely different story. COVID-19 itself is really anxiety provoking and for someone who, you know their son has PCD, where we don’t fully know how he’s going to react because I understand this is pretty much case by case, the anxiety is even higher. He has to go to school because there isn’t a shielding group 3 anymore. God knows what he is going to bring back from that school….You know any kind of social contact,…outside of our house, so that’s even more frightening for us" (mother 15)

Some parents deferred start of school for an additional year. All parents mentioned that they had deferred any holiday activities. Several parents mentioned that the COVID-19 pandemic had impacted on their child’s social interaction outside of the immediate household, especially ability to attend sport activities. COVID-19 caused an additional level of social isolation for the family caring for a vulnerable child. This mother mentioned that shielding during the beginning of the COVID-19 pandemic impacted on social development of her child:

"when we had to keep him inside for like 3 months and I was at home with him, shielding that affected him and when we first came back to our lives and seeing other people he was really quite shy he didn’t want to see anyone again….that way of keeping him inside for so long it was hard for him to get back into society and stuff like with people again.” (mother 17)

Most frequently mentioned was the enhanced vigilance around infections since the start of the pandemic, for example cleaning hands regularly, deterring the child from putting items into their mouths, washing clothes of family members who had attended outdoor activities, cleaning groceries coming into the house. General PCD care was sometimes delayed, routine clinic appointments postponed or conducted online, which especially impacted children with hearing problems and speech delay. The mother of one child reported:

“he had his hearing tested twice before lockdown before the March lockdown and then all the appointments got postponed. So, what they said is that he’s got a glue ear and he needs to be monitored closely but has not had any tests ever since” (mother 15)

Discussion

To the best of our knowledge, this is the first study exploring parental experiences of caring for pre-school children with PCD. It provides valuable insights into parental adjustment and adaptation to the diagnosis and management of their child’s PCD. Diagnosis of this rare and persistent condition was a disruptive life event. Adjustment to a new normality was a gradual process, facilitated by getting a diagnosis and an explanation for the child’s symptoms. Some families lived in countries where PCD health care was coordinated through a multidisciplinary team, whilst other families received fragmented care and were personally responsible for coordinating appointments with different specialties. As the COVID-19 pandemic started part way through the study, the information collected also covers PCD care during this unpredictable stressful life event.

As there is no cure for PCD, the health status of the child is dependent on PCD management. Parents found the additional effort to keep their young child healthy and happy was not only physically but also emotionally taxing. Although parents recognized the benefits of a PCD treatment regimen, similar to the reports of parents with other chronic conditions (Hawkins et al., 2020), the commitment provided challenges to the fulfilment of their social, vocational, and parenting roles. Parents were not able to attend social activities to a level previously enjoyed. The parent had to be more cautious in their interaction with others and PCD caring instructions prevented participation in social activities.

Consistent with the experiences of parents caring for children with other lifetime chronic conditions (Hatzmann et al., 2014) the parent’s vocational role and functioning was often affected. As reported by other studies (DeRigne & Porterfield, 2017) it was usually the mother who experienced changes in work status. In this study the parents often reported a need for more flexible, less demanding vocational roles. Families were often financially affected by combined reduction in working hours, as well as efforts to save for future healthcare procedures (e.g. fertility treatments).

Most parents felt a constant worry for their child’s wellbeing. About 50 percent of the parents reported recurrent feelings of anxiety and depression and 20 percent of the parents disclosed formal mental health
diagnosis or treatment. Although it seems that a lot of parents responded emotionally to the caregiving responsibility, the assumed prevalence of mental health problems from this informal source of reporting is similar to the existing lifetime mental health prevalence in the general population (Schaefer et al., 2017). More formal psychiatric epidemiological testing among caregivers of individuals living with PCD would be appropriate to determine a more accurate prevalence of mental health problems among this population.

Limitations

Although we recruited an international, diverse sample of parents caring for young children diagnosed with PCD, this group of parents might not be representative of the population of parents caring for young PCD children as 11 of the 24 participants were recruited from Southampton Children’s Hospital UK, more mothers than fathers participated, and most participants had a white ethnic background while the prevalence of PCD is higher in other ethnic groups (O’Callaghan et al, 2010). In addition, due to the partially retrospective aspect of the project, we cannot exclude the possibility of recall bias when the parents commented on historic events.

Practical implications

The findings of this study reveal that lack of awareness of PCD by clinicians sometimes limits disease management as it impedes GP care, emergency care, and pharmaceutical care. Due to the rarity of PCD the symptom pattern is not always recognized early in life, thereby delaying diagnosis. Over the past decade, diagnostic awareness campaigns have been able to lower the age of diagnosis of this rare hereditary chronic condition in some countries. It would be prudent to extend these awareness campaigns and improve the knowledge of health care providers about PCD and other rare diseases.

The impact of PCD care on the parent can be overwhelming at times. It induces time constraints and requires not only physical but also emotional efforts. The mental health and resilience of caregivers is important to the quality of care provided. Parents of children living with PCD are often unable to get a regular break from their caring responsibilities. Caregiver burden might be eased and more personalized care provided if PCD health care was integrated with social care, coordinating health care services with support for tasks of daily living and engagement with local community (Stein, 2001). Currently PCD services are geared towards providing appropriate services to cover the child’s PCD medical needs, but some parents reported a need for respite care; support with practical implementation of PCD management at home; caregiver appreciation; financial advice and support; awareness of community resources. A more holistic PCD care is responsive to meet not only the health care needs but also cover the social care needs of the family living with PCD.

Conclusion

This is the first international study examining the experiences of parents whose young child has been diagnosed with PCD. The findings from this study can be used to advise clinical practice in the holistic management of the child’s care. To end with these words from one of the parents:

_on the positive side it’s changed my view of life, I’m much more kind of, I appreciate him, I appreciate every, every second of his life and am much more and I’m much more in wonder I think of him. So I think there are real positives. And I couldn’t love him any more, because you just love them so much because you want them to be well and you know you want them to, yeah I mean it’s a real kind of protective instinct that takes over, but it’s got a real positive side, I think if you want it to it can have a real positive side to it (mother 18)_

Acknowledgements

We thank the patients and their families for participating in the study. Our thanks are also extended to PCD centres who recruited parents of young children diagnosed with PCD and the PCD Support Group UK who advertised this study on their social media. We like to thank Lynn Reeves and Samantha Packham for their support to the project management and administration of this research.

Declarations
Ethical approval

Ethical approval for the work presented in this manuscript was obtained from Southampton and South West Hants Research Ethics Committee (REC - 06/Q1702/109; with University of Southampton (ER-GO#53155). S Approval was also obtained in United States of America from the University of North Carolina Internal Review Board (IRB#09-1099), and in Canada from Hospital for Sick Children Research Ethics Board (REB#1000024263).

The work has therefore been performed in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki and its later amendments.

Consent for publication

Written consent from all individuals participating in this study has been obtained.

Competing interest

On behalf of all authors, the corresponding author states that there are no competing interests to declare.

Funding Statement

This study/project is funded by the National Institute for Health Research (NIHR) Research for Patient Benefit programme (NIHR 200470). The views expressed are those of the authors and not necessarily those of the NIHR or the Department of Health and Social Care. The UK National PCD Service is commissioned and funded by NHS England. PCD research in Southampton is supported by NIHR Southampton Respiratory Biomedical Research Unit and NIHR Wellcome Trust Clinical Research Facility.

Author Contributions

JSL and LB conceived and designed the study, secured funding and co-lead the governance of the study. CD developed the interview guide with LB and JSL, and led data collection, analysis, and drafting of this manuscript. LB assisted in data collection and analysis. RK provided qualitative research expertise. WW AH JSL SC SD MS ML KS LS ER MN provided guidance on clinical/demographic data collection forms, recruited the participants and provided clinical data. FC, LD, and EC supported participant recruited and guided the study with their lived experiences. BF, MT, and NT provided guidance on clinical/demographic data collection forms and guided the study with their lived experiences. All authors contributed in writing this paper and approved the final version.

REFERENCES


Eva SL Pedersen, Myrofora Goutaki, Amanda L Harris, Lucy Dixon, Michele Manion, Bernhard Rindlisbacher, COVID-PCD patient advisory group, Jane S Lucas, Claudia E Kuehni SARS-CoV-2 infec-


### Table 1: Child characteristics

<table>
<thead>
<tr>
<th>N=20 (%)</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Gender</td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>14 (70%)</td>
</tr>
<tr>
<td>Female</td>
<td>6 (30%)</td>
</tr>
<tr>
<td>Ethnicity</td>
<td></td>
</tr>
<tr>
<td>White</td>
<td>15 (75%)</td>
</tr>
<tr>
<td>Asian</td>
<td>3 (15%)</td>
</tr>
<tr>
<td>Other</td>
<td>2 (10%)</td>
</tr>
<tr>
<td>Ages</td>
<td></td>
</tr>
<tr>
<td>1 year</td>
<td>2 (10%)</td>
</tr>
<tr>
<td>2 years</td>
<td>4 (20%)</td>
</tr>
<tr>
<td>3 years</td>
<td>3 (15%)</td>
</tr>
<tr>
<td>4 years</td>
<td>5 (25%)</td>
</tr>
<tr>
<td>5 years</td>
<td>6 (30%)</td>
</tr>
<tr>
<td>Birth order of child</td>
<td></td>
</tr>
<tr>
<td>First</td>
<td>8 (40%)</td>
</tr>
<tr>
<td>Second</td>
<td>9 (45%)</td>
</tr>
<tr>
<td>Third</td>
<td>2 (10%)</td>
</tr>
<tr>
<td>Fifth</td>
<td>1 (5%)</td>
</tr>
<tr>
<td>Siblings with PCD</td>
<td></td>
</tr>
<tr>
<td>None</td>
<td>7 (35%)</td>
</tr>
<tr>
<td>Younger</td>
<td>9 (45%)</td>
</tr>
<tr>
<td>Older</td>
<td>5 (25%)</td>
</tr>
<tr>
<td>Missing</td>
<td>7 (35%)</td>
</tr>
<tr>
<td>Situs abnormalities</td>
<td></td>
</tr>
<tr>
<td>10 (50%)</td>
<td></td>
</tr>
<tr>
<td>Prophylactic antibiotics</td>
<td>7 (35%)</td>
</tr>
<tr>
<td>Respiratory infection(s) in past year</td>
<td>19 (95%)</td>
</tr>
<tr>
<td>Hospital Admissions in past year</td>
<td>6 (30%)</td>
</tr>
</tbody>
</table>

### Table 2: Thematic framework

<table>
<thead>
<tr>
<th>Major Themes:</th>
<th>Parent’s reaction to:</th>
<th>Parent’s reaction to:</th>
<th>Parent’s reaction to:</th>
<th>Parental:</th>
<th>Parental:</th>
</tr>
</thead>
<tbody>
<tr>
<td>PCD diagnosis</td>
<td>Treatment regimen child</td>
<td>Health status child</td>
<td>Coping</td>
<td>Concerns for future</td>
<td></td>
</tr>
<tr>
<td>child</td>
<td>Daily treatment: Parent-child interaction</td>
<td>Emotional</td>
<td>Support seeking</td>
<td>Explaining PCD</td>
<td></td>
</tr>
<tr>
<td>Prenatal experiences</td>
<td>Emotional impact on parents</td>
<td>Social/emotional impact PCD</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Feelings before Medical procedures and medication</td>
<td>Health</td>
<td>Problem-focused</td>
<td>Social/emotional impact PCD</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Appendix 1

**Interview guide for development of HRQoL questionnaire: QOL-PCD^{PR}**

The interviews will be conducted in a semi-structured way. The questions below are meant to serve as a guide; the researcher will adapt each interview according to the relevance of the different section to the interviewee. The sections in red will only be asked if there is time.

**Introduction**

Thank you very much for agreeing to participate in this study.

As mentioned in the Participant Information Sheet, this interview is to help us to understand how your child’s PCD effects them, and you. This information will be used for the development of a questionnaire which can be used in clinics or research to monitor how PCD is effecting your quality of life. The interview is being recorded but the recording will be kept confidential, we won’t feed anything back to your medical team or anyone else, and you won’t be identified in any reports or presentations. Before we go further please can you let me know if you have any questions about the research?

Once any questions are answered, You have previously signed a consent form for this study, please can you confirm that you are still happy to participate, and that you are happy to be recorded?

The interview will take roughly an hour so I would like to confirm with you that we do have sufficient time for this. Obviously if you would like to stop at any time please let me know and we can stop, or arrange a convenient time to resume the interview, whichever you prefer.

Do you have any (further) questions for me before we start?

**General**

Tell me about what is it like for you having a child with PCD?

What is it like for your child having PCD?

**Diagnosis**

- When did you first recognise your child was experiencing symptoms caused by PCD?
- Can you tell me about how your child was diagnosed?
- What words best describe what you felt like before your child was diagnosed?
- What words best described what you felt like when you heard the diagnosis?
• What was your response when you learned your child’s diagnosis?

Additional questions
Did having a diagnosis to explain your child’s symptoms change things?
Has your life changed since having a child with PCD?

Symptoms
• Can you tell me about what symptoms your child experiences on a day to day basis?
• How do you monitor your child’s PCD signs and symptoms?
• How do these symptoms affect your child on a daily basis? (Prompt: cover physical, emotional and social functioning)
• How does your child experiencing these symptoms affect you on a daily basis?

Treatment burden
• What type of things are you doing for your child’s PCD care?
• How do you incorporate your child’s treatment regimen in your daily life?
• How does the treatment regimen affect your child?
• When planning things like holidays or family activities are there things that have to be considered because of your child’s PCD?

Additional questions
Did you make any changes to family life to accommodate your child’s treatment?

Care received
• Can you talk about your child medical care?
• How do you feel your child treatment is going?
• Do you feel like you are being involved in the decisions that are being made about your son’s/daughter’s care and can communicate your concerns to the clinical staff?
• What words best describe what you feel like when your child is not feeling well or even has to be hospitalized? What has helped you deal with this?
• Have you ever had to calm your child for medical care or treatment? How did that make you feel?

Additional questions
• How do you make sure your child does his/her treatment as prescribed?
• Do you feel the clinical staff listens to what you have to say and understands what you are saying?
• Do you feel you can communicate new signs and symptoms of PCD effectively to the clinical staff?
• Do you feel you know as much as you want to know about PCD and the treatment for your child? Do you feel the clinical staff provides you with the information you are looking for?

Social Functioning and Role
• Do you believe you have changed now that you are caring for a child with a chronic illness?
• How has having a child with PCD impacted on your relationships with others? (Prompt: address family, friends, spouse, children and colleagues)
• Has having a child with PCD lead to new relationships in your life?
• How has having a child with PCD impacted on your personal, family or career goals? (Prompt: does your child’s chronic illness create any financial concerns in the family (e.g. travel costs to hospital; care arrangements siblings; cost special food or things; trouble maintaining employment?)
• Do you feel like extended family members and the community understand what PCD is and what you as a family are going through?
• Do you feel like your child’s PCD has an impact on your child in terms of his or her development? (Prompt: Address physical, emotional and social functioning. Under social functioning address relationships with family members i.e. siblings, and those outside of the home)

Additional questions

Do you discuss your child’s medical condition openly with them? Do you feel that you can explain it to them in a way that they can understand it? What is it like for you when you have to talk to your child about PCD, their treatment, going to the hospital, the side-effects they are experiencing, etc?

Emotional functioning

What words best describe what you feel like now in the current situation?

How does PCD affect your child emotionally at the moment? How has it impacted him or her in the past?

Concerns about the future

Do you have any concerns about your child’s future?

Additional sections

Coping

Some people caring for a child with a long-term condition have said there are times when they wondered how they would manage with the situation, have you ever felt this? If yes how did you deal with this? What things were helpful at that time?

Transitions

Every parent has to get used to leaving their child in someone else’s care.

Have you left your child in other people’s care (address partner, family member/friend, school/daycare)

What do you feel when you leave them in other people’s care (if worried ask what they are worried about)