Research priorities in Primary Ciliary Dyskinesia - patients´ perspective

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Introduction

Patients can provide important perspectives for health research that may differ from those of health care professionals. This is particularly important for a rare disease with many knowledge gaps like primary ciliary dyskinesia (PCD). We aimed to identify research priorities in PCD from the patients' perspective.

Methods

We present the first part of a two-part mixed methods study. We conducted, transcribed, coded and thematically analysed in-depth semi-structured interviews with adults and teens with PCD or parents of children with PCD. Invited participants included purposefully selected patients from the Swiss PCD registry, volunteers through PCD support UK, and the BEAT-PCD network.

Results

We interviewed 22 participants including 9 adults, 9 parents, and 4 adolescents from 7 countries mainly Switzerland, and UK.

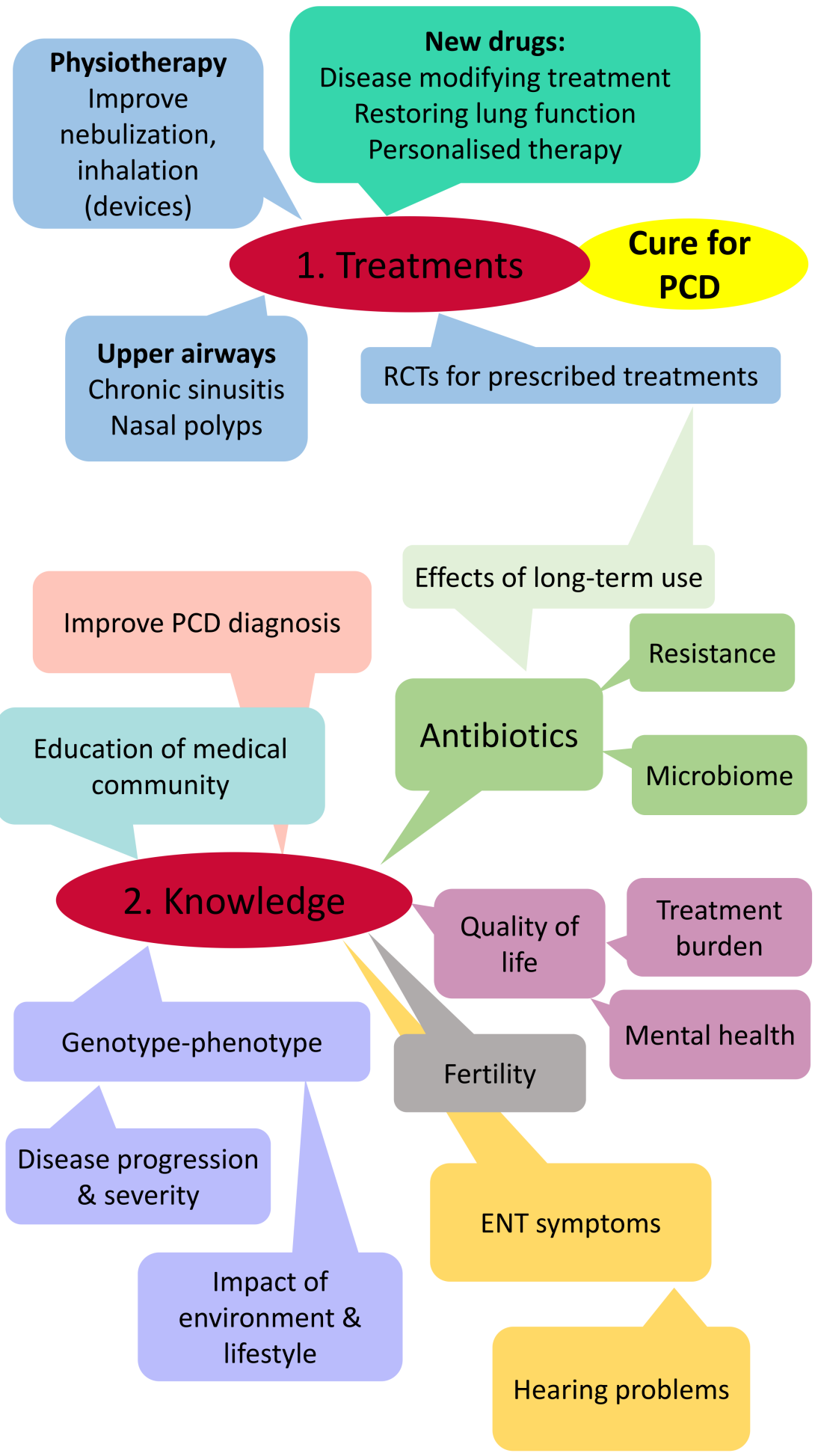
Country of interviewed participants (N=22)	
Switzerland	9
UK	7
Other European countries	3
Non-European countries	3

The main theme we identified as priority for PCD research was related to 1. **treatments**: evaluation and development of PCD-specific treatments but also evaluation of currently prescribed treatments in clinical trials (Figure 1). A second important theme was related to increasing 2. **knowledge** about the disease. Research on areas besides the lungs was highlighted including research on ENT symptoms (particularly hearing problems), fertility, mental health, and treatment burden. Other themes covered were improving PCD diagnosis, understanding of disease progression, and disease severity, and the impacts of environmental or lifestyle changes on symptoms.

"What I want more than anything… What I want is **a PCD version of the cystic fibrosis medication that works**! THAT'S what I want! But it's very difficult!"

"Maybe one day… maybe at the end of my lifetime, we will be **doing personalised genetic treatments**. I wonder… it would be nice to think we will."

Figure 1: Main research priorities suggested by participants



Conclusion and next steps

These results will be used to develop an online survey that will be translated in multiple languages and circulated worldwide in collaboration with the BEAT-PCD network, the European Lung Foundation, and PCD patient advocacy groups.

Final study results will help PCD researchers to establish research priorities, addressing patients' wishes and needs.

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Funding: SNF PZ00P3_185923, Foundation Johanna Dürmüller-Bol