

Health-related quality of life in primary ciliary dyskinesia (PCD): Translation, cross-cultural adaptation and content validation of the Italian QOL-PCD questionnaire

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Abstract

Background

Primary ciliary dyskinesia (PCD) is a rare genetic disorder characterized by impaired mucociliary clearance, chronic respiratory symptoms, and reduced health-related quality of life (HRQoL). Although the disease-specific Quality of Life Questionnaire for PCD (QOL-PCD) is available in several languages, no Italian version has been available to date. This study aimed to translate, culturally adapt, and assess the content validity of the Italian QOL-PCD.

Methods

The original English QOL-PCD questionnaires for children, adolescents, adults, and parents/caregivers were translated into Italian using a standardized forward–backward translation process. Cross-cultural adaptation included expert review and harmonization with the original developer. Cognitive interviews were conducted with 20 participants using a think-aloud process to assess comprehensibility, relevance, and acceptability. Content validity was evaluated by three PCD experts using item- and scale-level Content Validity Indices (I-CVI and S-CVI).

Results

Four Italian versions of the QOL-PCD were successfully translated. Cognitive testing showed that instructions, items, recall period, and response options were clear and relevant, requiring only minor linguistic modifications. No new issues emerged, indicating adequate saturation. Content validity was high, with most items achieving perfect expert agreement (I-CVI = 1.00). Scale-level content validity was excellent, with S-CVI values of 1.00 (child), 0.95 (adolescent), 0.99 (adult), and 0.86 (parent/caregiver).

Conclusions

The Italian QOL-PCD questionnaires were successfully translated and culturally adapted. The instruments were well understood by patients and caregivers and showed good to excellent content validity across all versions. These findings support future psychometric validation and the use of the Italian QOL-PCD in clinical practice, research, and international clinical trials.

1. INTRODUCTION

Primary ciliary dyskinesia (PCD) is a hereditary genetic disease that affects the structure and movement of motile cilia, thereby compromising mucociliary clearance of the airways.¹ This can result in neonatal respiratory distress, recurrent sinus and bronchopulmonary infections, otitis media, bronchiectasis,

organ laterality defects and subfertility.¹ Recent genetic discoveries have led to an increase in the estimated incidence from 1 in 15,000–1 in 130,000 births to approximately 1 in 7,600, with higher frequencies observed in populations with founder variants.¹ Nevertheless, the condition remains underdiagnosed, with a significant number of patients receiving an incorrect diagnosis of idiopathic bronchiectasis.¹

The diagnostic pathway for PCD is complex and requires a multidisciplinary approach, as no single diagnostic test is sufficient. Current diagnostic algorithms integrate nasal nitric oxide measurement, high-speed video microscopy analysis, transmission electron microscopy, and genetic testing.¹ Despite improvements in supportive care, no disease-modifying therapies capable of restoring ciliary function are currently available. However, since bronchiectasis in PCD is caused by chronic neutrophilic inflammation, new drugs have been recently studied showing improvements in recurrence of respiratory infections and lung function by Dipeptidyl peptidase-1 (also known as cathepsin C) inhibition.^{2,3} Consequently, clinical management focuses on slowing disease progression, treating respiratory exacerbations, preventing complications, and ultimately improving health-related quality of life (HRQoL)⁴.

People with primary ciliary dyskinesia (PW-PCD) experience respiratory symptoms with chronic productive cough, production of purulent sputum and recurrent infections requiring multiple daily treatments which impose a considerable burden on patients and their families. The symptom and treatment burden negatively impact their physical, emotional, and social HRQoL.⁴ This lifelong disease burden and persistent symptomatology can negatively affect treatment adherence and contribute to disease progression, including decline in lung function.⁴ In addition, previous studies have shown that individuals with PCD report more symptoms of anxiety and depression than the general population, which negatively impact their HRQoL.^{5,6} In this context, systematic assessment of HRQoL represents a crucial component of comprehensive patient care.

Alongside conventional physiological measures such as spirometry, lung clearance index and the six-minute walking test, use of disease-specific instruments capture the specific challenges in daily functioning experienced by PW-PCD and are more sensitive to change. Regulatory agencies, including the U.S. Food and Drug Administration and the European Medicines Agency, require the use of patient-reported outcome measures in medical product development to ensure that the patients' perceptions of symptom improvement and impacts on HRQoL are captured.^{7,8}

To address the need for disease-specific HRQoL assessment in PCD, the QOL-PCD questionnaire was developed, with versions available for children, adolescents, and adults.^{9,10} The original English version was developed based on qualitative individual and group interviews with specialists, pediatric and adult patients with PCD, and parents of pediatric PCD patients. Draft measures were subsequently refined through cognitive interviews. The English QOL-PCD instruments have been validated, representing a crucial step toward their use in both research and clinical practice. Each age version includes between 34 and 40 items depending on whether it is the child, adolescent, adult or parent proxy version. Over

time, the questionnaires have been translated and culturally adapted into multiple languages and implemented across various countries, including Cyprus, Israel, Brazil and Poland.^{11,14}

Despite the availability of validated QOL-PCD questionnaires in several languages, no disease-specific HRQoL instrument has been available for Italian-speaking PW-PCD, limiting both routine assessment of patient-reported outcomes and participation in international collaborative studies.

The aim of this study was to describe the process by which the original English QOL-PCD instruments were translated into Italian and cross-culturally adapted according to a standardized, well-established procedure.

2. METHODS

The four versions of the questionnaire were translated from English into Italian. A forward then backward translation of the instruments was performed using independent translators who speak English and are experts in PCD at Bambino Gesù Children's Hospital in Rome which is part of the European Reference Network LUNG, with review by one of the original authors of the QOL-PCD. The process followed the guidelines for translation of PROs published by the FDA and EMA.¹⁵ Cognitive testing interviews were conducted after the translation process using a “think aloud” approach.¹⁶ A flowchart of the development and cross-cultural adaptation of the Italian language versions of QOL-PCD is summarized in Fig. 1. Written consent and assent were obtained. The study was reviewed and approved by the Ethics Committee of Bambino Gesù Children's Hospital (Institutional Review Board, IRB). The study was conducted in accordance with the ethical principles of the Declaration of Helsinki and its subsequent amendments.

2.1 Translation and adaptation. The initial step was the translation of the English versions of the QOL-PCD child, adolescent, parent-proxy, and adult into Italian. The forward translations were completed by two independent translators who are native Italian speakers, also speak fluent English, and are experts in PCD (psychologist, pediatric pulmonologist). This first step was followed by a teleconference with one of the authors (Dr. Alexandra L. Quittner) to discuss and resolve discrepancies and perform a cultural adaptation. This yielded the “*consensus forward*” Italian version. Those versions were then *back-translated* into English by 2 new, independent Italian speakers who are also fluent in English. Next, a second teleconference was conducted with the author to ensure that the items conveyed the original meaning (“harmonization”), utilized simple medical terminology, and were culturally equivalent to the original English.

2.2 Cognitive Testing

After completing the questionnaire, participants took part in an individual cognitive debriefing interview. During this “think aloud” interview, they were asked how they interpreted each question, whether they could rephrase it in their own words, what came to mind when reading it, and if the rating scales were clear and easy to use.¹⁶ This process was repeated for every item. Respondents were also asked for

their opinions on the questions, including whether any terms were unclear or if any important aspects of PCD’s impact on their HRQoL were missing (Table 1). All feedback was recorded and used by the expert team to refine the questionnaire further. The feedback was recorded and used for further revisions by the experts. Cognitive testing interviews were conducted with 5 parents of children with PCD, 5 children with PCD, 5 adolescents with PCD, and 5 adults with PCD (Table 1) (*N* = 20). The cognitive testing questions were administered after every four items. The final versions of the QOL-PCD were administered to the patients and parents of minor children to assess the feasibility and practicality of the tool. After completion of the measure, specific probes were utilized to assess the clarity of the items, ease of completing the rating scales, and to ensure that the content was comprehensive (Table 2). Any controversies related to ambiguous wording were resolved, and a consensus translation was finalized. Minor cultural differences were identified and discussed, and all remaining discrepancies were resolved through consensus.

Table 1
QOL-PCD Cognitive Interview

QOL-PCD Cognitive Interview	
Instruction	- <i>What do you think about these instructions?</i> - <i>Is there anything that should be changed to make them clearer?</i> - <i>Did you clearly understand what you are being asked to do?</i> - <i>Is it clear that you are asked to consider the last 7 days?</i>
Part 1 and Part 2	- <i>What were you asked in these questions?</i>
EVERY 4 QUESTIONS	- <i>Can you explain it in your own words?</i>
(up to the symptom questions and for the symptoms)	- <i>Do you think any word is too difficult?</i> - <i>Which word(s) would you use instead?</i>

2.3 Content Validity

Based on the original English version, four prototype Italian QOL-PCD questionnaires were developed, one for each target population: children, adolescents, adults, and parents of children with PCD. For evaluation of content validity, all of the experts reviewed the items for the Italian prototype version and compared them to the original.¹⁷ All items were evaluated by three experts in PCD, who assessed their relevance in terms of semantic, idiomatic, cultural, and conceptual equivalence.

The content validity index (CVI) was calculated at both the item level (I-CVI) and scale level (S-CVI). Three experts with extensive clinical experience in PCD independently rated the relevance of each item on a 4-point Likert scale (1 = *not relevant*, 2 = *somewhat relevant*, 3 = *quite relevant*, 4 = *highly relevant*).

The I-CVI for each item was computed as the proportion of experts assigning a rating of 3 or 4. The S-CVI was calculated as the average of the I-CVI values across all items. Content validity was considered acceptable if the S-CVI values exceeded 0.80.¹⁸

3. RESULTS

Cognitive Testing. The sample included 20 participants divided into four groups: parents of children aged 6–12 years ($N = 5$; 80% female, 20% male), children aged 6–12 years ($N = 5$; 80% female, 20% male), adolescents aged 13–17 years ($N = 5$; 100% male), and adults aged ≥ 18 years ($N = 5$; 100% female). Characteristics of the sample are reported in Table 2. Results indicated that the questions were relevant and clear, the response options were easy to use, and the measure was all-inclusive, capturing patients’ symptoms and challenges. Only a few specific words were difficult to understand (e.g., “chest hurt”; used a simpler word for “chest” for young children); minor edits were made to produce the final Italian versions. Overall, the specific items of the translated versions were easily understood by patients and parents of minor children. These terms were reviewed by the expert committee and revised to clarify their meaning. Notably, after the fifth interview in each group, no new comments emerged and no further revisions were deemed necessary (i.e., saturation was met).

The final, cross-culturally adapted Italian versions of the QOL-PCD questionnaire retained the core characteristics of the original English instruments. Minor modifications to spelling and punctuation were made to ensure standardization of format and layout in line with the original questionnaires.

Table 2
Sociodemographic characteristics of the sample ($N = 20$)

Group	N	Gender	Mean age (years)
Parents of children aged 6–12 years	5	4 Females, 1 Male	41.6
Children aged 6–12 years	5	4 Females, 1 Male	11.0
Adolescents aged 13–17 years	5	5 Males	15.6
Adults (≥ 18 years)	5	5 Females	21.8

Content Validity. The I-CVI values were high across all items, with the majority of items achieved perfect agreement (I-CVI = 1.00). A small number of items showed lower agreement (I-CVI = 0.67), indicating minor discrepancies among experts. The S-CVI values demonstrated excellent overall content validity for all versions of the questionnaire, with values of 1.00 for the child version, 0.95 for the adolescent version, 0.99 for the adult version, and 0.86 for the parent version (Table 3).

Table 3
Content validity index (CVI) for the PCD-QOL questionnaire: Children, adolescents, adults, and parents/caregivers

Children		Item Expert 1	Expert 2	Expert 3	Questionnaire Experts in agreement	Item I- CVI
	1	Yes	Yes	Yes	3	1
	2	Yes	Yes	Yes	3	1
	3	Yes	Yes	Yes	3	1
	4	Yes	Yes	Yes	3	1
	5	Yes	Yes	Yes	3	1
	6	Yes	Yes	Yes	3	1
	7	Yes	Yes	Yes	3	1
	8	Yes	Yes	Yes	3	1
	9	Yes	Yes	Yes	3	1
	10	Yes	Yes	Yes	3	1
	11	Yes	Yes	Yes	3	1
	12	Yes	Yes	Yes	3	1
	13	Yes	Yes	Yes	3	1
	14	Yes	Yes	Yes	3	1
	15	Yes	Yes	Yes	3	1
	16	Yes	Yes	Yes	3	1
	17	Yes	Yes	Yes	3	1
	18	Yes	Yes	Yes	3	1
	19	Yes	Yes	Yes	3	1
	20	Yes	Yes	Yes	3	1
	21	Yes	Yes	Yes	3	1
	22	Yes	Yes	Yes	3	1
	23	Yes	Yes	Yes	3	1
	24	Yes	Yes	Yes	3	1

Abbreviation: PCD-QOL, primary ciliary dyskinesia-quality of life.

Note: *Adults*: *18 = I feel comfortable blowing my nose in front of others, *28 = You felt congested in the chest; *Parents/Caregivers*: *23 = My child's chest was congested.

Children		Item Expert 1	Expert 2	Expert 3	Questionnaire Experts in agreement	Item I- CVI
	1	Yes	Yes	Yes	3	1
	25	Yes	Yes	Yes	3	1
	26	Yes	Yes	Yes	3	1
	27	Yes	Yes	Yes	3	1
	28	Yes	Yes	Yes	3	1
	29	Yes	Yes	Yes	3	1
	30	Yes	Yes	Yes	3	1
	31	Yes	Yes	Yes	3	1
	32	Yes	Yes	Yes	3	1
	33	Yes	Yes	Yes	3	1
	34	Yes	Yes	Yes	3	1
Adolescents	1	Yes	Yes	Yes	3	1
	2	Yes	Yes	Yes	3	1
	3	Yes	Yes	Yes	3	1
	4	Yes	Yes	Yes	3	1
	5	Yes	Yes	Yes	3	1
	6	Yes	Yes	Yes	3	1
	7	Yes	Yes	Yes	3	1
	8	Yes	Yes	Yes	3	1
	9	Yes	Yes	Yes	3	1
	10	Yes	Yes	Yes	3	1
	11	Yes	Yes	Yes	3	1
	12	Yes	Yes	Yes	3	1
	13	Yes	Yes	Yes	3	1
	14	Yes	Yes	Yes	3	1

Abbreviation: PCD-QOL, primary ciliary dyskinesia-quality of life.

Note: *Adults*: *18 = I feel comfortable blowing my nose in front of others, *28 = You felt congested in the chest; *Parents/Caregivers*: *23 = My child's chest was congested.

Children		Item Expert 1	Expert 2	Expert 3	Questionnaire Experts in agreement	Item I- CVI
	1	Yes	Yes	Yes	3	1
	15	Yes	Yes	Yes	3	1
	16	Yes	Yes	Yes	3	1
	17	Yes	Yes	Yes	3	1
	18	Yes	Yes	Yes	3	1
	19	Yes	Yes	Yes	3	1
	20	Yes	Yes	Yes	3	1
	21	Yes	Yes	Yes	3	1
	22	Yes	Yes	Yes	3	1
	23	Yes	Yes	Yes	3	1
	24	Yes	Yes	Yes	3	1
	25	Yes	Yes	Yes	3	1
	26	Yes	Yes	Yes	3	1
	27	Yes	Yes	Yes	3	1
	28	Yes	Yes	Yes	3	1
	29	Yes	Yes	Yes	3	1
	30	Yes	Yes	Yes	3	1
	31	Yes	Yes	Yes	3	1
	32	Yes	Yes	Yes	3	1
	33	Yes	Yes	Yes	3	1
	34	Yes	Yes	Yes	3	1
	35	Yes	Yes	Yes	3	1
	36	Yes	Yes	Yes	3	1
	37	Yes	Yes	Yes	3	1
	38	Yes	Yes	Yes	3	1

Abbreviation: PCD-QOL, primary ciliary dyskinesia-quality of life.

Note: *Adults*: *18 = I feel comfortable blowing my nose in front of others, *28 = You felt congested in the chest; *Parents/Caregivers*: *23 = My child's chest was congested.

Children		Item Expert 1	Expert 2	Expert 3	Questionnaire Experts in agreement	Item I- CVI
	1	Yes	Yes	Yes	3	1
Adults	1	Yes	Yes	Yes	3	1
	2	Yes	Yes	Yes	3	1
	3	Yes	Yes	Yes	3	1
	4	Yes	Yes	Yes	3	1
	5	Yes	Yes	Yes	3	1
	6	Yes	Yes	Yes	3	1
	7	Yes	Yes	Yes	3	1
	8	Yes	Yes	Yes	3	1
	9	Yes	Yes	Yes	3	1
	10	Yes	Yes	Yes	3	1
	11	Yes	Yes	Yes	3	1
	12	Yes	Yes	Yes	3	1
	13	Yes	Yes	Yes	3	1
	14	Yes	Yes	Yes	3	1
	15	Yes	Yes	Yes	3	1
	16	Yes	Yes	Yes	3	1
	17	Yes	Yes	Yes	3	1
	18*	Yes	Yes	No	2	0.67*
	19	Yes	Yes	Yes	3	1
	20	Yes	Yes	Yes	3	1
	21	Yes	Yes	Yes	3	1
	22	Yes	Yes	Yes	3	1
	23	Yes	Yes	Yes	3	1
	24	Yes	Yes	Yes	3	1

Abbreviation: PCD-QOL, primary ciliary dyskinesia-quality of life.

Note: *Adults*: *18 = I feel comfortable blowing my nose in front of others, *28 = You felt congested in the chest; *Parents/Caregivers*: *23 = My child's chest was congested.

Children		Item Expert 1	Expert 2	Expert 3	Questionnaire Experts in agreement	Item I- CVI
	1	Yes	Yes	Yes	3	1
	25	Yes	Yes	Yes	3	1
	26	Yes	Yes	Yes	3	1
	27	Yes	Yes	Yes	3	1
	28*	Yes	Yes	No	2	0.67*
	29	Yes	Yes	Yes	3	1
	30	Yes	Yes	Yes	3	1
	31	Yes	Yes	Yes	3	1
	32	Yes	Yes	Yes	3	1
	33	Yes	Yes	Yes	3	1
	34	Yes	Yes	Yes	3	1
	35	Yes	Yes	Yes	3	1
	36	Yes	Yes	Yes	3	1
	37	Yes	Yes	Yes	3	1
	38	Yes	Yes	Yes	3	1
	39	Yes	Yes	Yes	3	1
	40	Yes	Yes	Yes	3	1
Parents/Caregivers	1	Yes	Yes	Yes	3	1
	2	Yes	Yes	Yes	3	1
	3	Yes	Yes	Yes	3	1
	4	Yes	Yes	Yes	3	1
	5	Yes	Yes	Yes	3	1
	6	Yes	Yes	Yes	3	1
	7	Yes	Yes	Yes	3	1
	8	Yes	Yes	Yes	3	1

Abbreviation: PCD-QOL, primary ciliary dyskinesia-quality of life.

Note: *Adults*: *18 = I feel comfortable blowing my nose in front of others, *28 = You felt congested in the chest; *Parents/Caregivers*: *23 = My child's chest was congested.

Children		Item Expert 1	Expert 2	Expert 3	Questionnaire Experts in agreement	Item I- CVI
	1	Yes	Yes	Yes	3	1
	9	Yes	Yes	Yes	3	1
	10	Yes	Yes	Yes	3	1
	11	Yes	Yes	Yes	3	1
	12	Yes	Yes	Yes	3	1
	13	Yes	Yes	Yes	3	1
	14	Yes	Yes	Yes	3	1
	15	Yes	Yes	Yes	3	1
	16	Yes	Yes	Yes	3	1
	17	Yes	Yes	Yes	3	1
	18	Yes	Yes	Yes	3	1
	19	Yes	Yes	Yes	3	1
	20	Yes	Yes	Yes	3	1
	21	Yes	Yes	Yes	3	1
	22	Yes	Yes	Yes	3	1
	23	Yes	Yes	No	2	0.67*
	24	Yes	Yes	Yes	3	1
	25	Yes	Yes	Yes	3	1
	26	Yes	Yes	Yes	3	1
	27	Yes	Yes	Yes	3	1
	28	Yes	Yes	Yes	3	1
	29	Yes	Yes	Yes	3	1
	30	Yes	Yes	Yes	3	1
	31	Yes	Yes	Yes	3	1
	32	Yes	Yes	Yes	3	1

Abbreviation: PCD-QOL, primary ciliary dyskinesia-quality of life.

Note: *Adults*: *18 = I feel comfortable blowing my nose in front of others, *28 = You felt congested in the chest; *Parents/Caregivers*: *23 = My child's chest was congested.

Children		Item Expert 1	Expert 2	Expert 3	Questionnaire Experts in agreement	Item I- CVI
	1	Yes	Yes	Yes	3	1
	33	Yes	Yes	Yes	3	1
	34	Yes	Yes	Yes	3	1
	35	Yes	Yes	Yes	3	1
Abbreviation: PCD-QOL, primary ciliary dyskinesia-quality of life. Note: <i>Adults</i> : *18 = I feel comfortable blowing my nose in front of others, *28 = You felt congested in the chest; <i>Parents/Caregivers</i> : *23 = My child's chest was congested.						

Table 3. Content validity index (CVI) for the PCD-QOL questionnaire: Children, adolescents, adults, and parents/caregivers

4. DISCUSSION

This study described the translation, cross-cultural adaptation, cognitive testing, and content validation of the original English QOL-PCD questionnaires into Italian. Four Italian versions were developed, targeting children, adolescents, adults, and parents/caregivers of children with PCD. These measures are the first Italian, disease-specific HRQoL instruments available for PW-PCD. This work also represents the first formal cross-cultural adaptation of a PCD-specific patient-reported outcomes (PROs) measure for the Italian-speaking population. Overall, our findings indicated that the Italian QOL-PCD preserved the conceptual content of the original English instrument, was well understood by respondents across different age groups, and showed satisfactory to excellent content validity.¹⁹ All of these processes were conducted in collaboration with one of the authors of the original English QOL-PCD measures.

Assessment of HRQoL is increasingly recognized as an essential component of comprehensive care for PW-PCD. Although physiological and clinical indicators such as lung function, pulmonary exacerbations, microbiological status, and imaging results document important information about disease severity, they do not fully capture the patient's subjective experience of living with a chronic respiratory disease.²⁰ Disease-specific PROs provide the perspective of the person or caregiver who is living with the disease. Importantly, they evaluate the impact of the disease on the "whole person" including their daily symptoms, treatment burden, emotional well-being, social functioning, and physical functioning.

Cognitive testing demonstrated that the items and rating scales were well-understood. Participants reported that the instructions, recall period, response options, and questionnaire items were clear and relevant to their experience. Only minor linguistic modifications were required following participant feedback. Importantly, no new concerns emerged after the fifth cognitive interview within each respondent group, suggesting that adequate saturation had been achieved for the purposes of linguistic and cultural adaptation. These findings support the feasibility and validity of administering the Italian QOL-PCD across different age groups and respondent categories.

Content validity analyses also confirmed the adequacy of the translated instruments. Most items achieved complete agreement among experts, and all four questionnaire versions demonstrated scale-level content validity indices above the recommended threshold with a very small number of items showing lower item-level agreement (I-CVI = 0.67). Interestingly, the items generating lower agreement highlighted some of the most challenging aspects of cross-cultural adaptation. One item referred to feeling comfortable blowing one's nose in front of other people, a situation whose interpretation may vary according to age, social norms, and cultural attitudes toward visible symptoms. Another item concerned the sensation of a "congested chest," a subjective symptom that may encompass different meanings in Italian, including mucus retention, chest heaviness, or difficulty clearing secretions. These findings illustrate the complexity of translating PROs, where conceptual equivalence is often more important than literal linguistic correspondence. Importantly, cognitive debriefing confirmed that the final wording was understandable and acceptable to patients and caregivers, supporting retention of these items in the Italian version.

Our findings are closely aligned with previous translation and adaptation studies of the QOL-PCD in other languages including Hebrew, Portuguese for Brazil and Polish.¹¹⁻¹⁴ In particular, the Hebrew translation study used a similar methodology, including forward translation, back-translation, expert review, and pretesting in 20 participants, and reported comparable content validity values across the four questionnaire versions. The similarity between the Hebrew and Italian findings supports the robustness of the translation and cultural adaptation framework used for the QOL-PCD and suggests that this approach can be successfully applied across different linguistic and cultural settings.¹²

The potential applicability of the Italian QOL-PCD in routine clinical care is also supported by recent evidence from the Polish validation study, which showed that QOL-PCD questionnaires can be administered during follow-up visits and generally require only a short completion time.¹⁴ This is important because, in real-world clinical settings, the time available during consultations is often limited, and discussion of how patients experience their disease may be insufficient. A brief, disease-specific questionnaire may help structure this discussion and provide clinicians with information that might otherwise remain unexplored. Moreover, the Polish study suggested that perceived quality of life may differ across countries and cultural contexts, reinforcing the need for linguistically and culturally adapted versions rather than relying on the original English instrument alone.¹⁴

The availability of all four Italian versions is another important strength of this work. PCD affects patients throughout life, but the impact of the disease varies according to developmental stage and disease progression. Children, adolescents, adults, and parents/caregivers may experience and report disease burden differently, particularly in relation to treatment burden, school or work limitations, emotional well-being, social participation, and family routines.²¹ Having age-specific Italian instruments will allow future studies to explore how the impact of PCD changes across the lifespan, will facilitate longitudinal assessment from childhood to adulthood and the testing of new treatments in clinical trials. The development of patient registries is another important step in advancing new treatment approaches

and including PROs is critically important. The EMBARC Patient Registry collects data on HRQoL using the QOL-B and a new pediatric bronchiectasis registry has recently been launched.^{22,23}

Currently, most of the prescribed treatments for PCD represent symptomatic treatment, largely borrowed from the cystic fibrosis literature, with little evidence of their efficacy. However, new treatments have recently been approved to treat bronchiectasis with potential applications to PCD. Many new medications are being tested in Phase 3 clinical trials in PCD and having valid translations of these PROs will facilitate inclusion of Italian patients and parent caregivers.

This study has several strengths. First, all four age-specific versions of the QOL-PCD were translated and culturally adapted, allowing assessment across pediatric, adolescent, adult, and parent/caregiver populations. Second, the process involved both PCD experts and members of the target population, ensuring that the final version was clinically relevant and understandable to patients and families. Third, content validity was assessed quantitatively using both item-level and scale-level CVI, providing objective evidence of expert agreement regarding item relevance. Finally, the translation process was conducted in close connection with one of the original authors, supporting consistency with the source instrument.

Some limitations should be acknowledged. The sample size used for cognitive testing was small, with five participants in each respondent group. However, this approach is consistent with previous QOL-PCD translation studies and with the aims of cognitive debriefing, particularly in the context of a rare disease¹². In addition, no additional comments emerged after the fifth interview in each group, suggesting that further interviews were unlikely to identify new issues. The most important limitation of the present study is the absence of a comprehensive psychometric validation. Although content validity and comprehensibility were demonstrated, further studies are required to assess internal consistency, test-retest reliability, construct validity, responsiveness, and measurement invariance. Most importantly, the Italian QOL-PCD measures need to demonstrate responsivity to change related to a new medication or intervention, which would also facilitate identification of the minimal important difference score (MID) for each scale.

Future, multicentre studies will be needed to confirm the psychometric properties of the Italian QOL-PCD and to evaluate its performance across different clinical settings and age groups. In particular, future research should investigate associations between QOL-PCD scores and clinical indicators such as lung function, respiratory exacerbations, microbiological status, hearing symptoms, treatment burden, and disease severity. This is highly relevant in the context of rare diseases, where PROs are increasingly recognized as essential for understanding the real impact of the disease and its treatment from the patient's perspective.

In conclusion, the Italian versions of the QOL-PCD questionnaires were successfully translated, culturally adapted, and validated. The final instruments were easily understood by patients and parents/caregivers and demonstrated satisfactory to excellent content validity across child, adolescent, adult, and parent/caregiver versions. These findings support the Italian QOL-PCD as a promising disease-specific

tool for assessing HRQoL in Italian-speaking people with PCD. Given the rarity of PCD, the availability of a harmonized Italian QOL-PCD provides a foundation for future psychometric validation studies and may facilitate the integration of Italian-speaking patients into international research initiatives, registries, and clinical trials. In clinical practice, the Italian QOL-PCD will help clinicians identify patients who experience greater difficulties with daily treatments and support more individualized strategies to improve disease management.

Abbreviations

Primary ciliary dyskinesia (PCD)

Health-related quality of life (HRQoL)

People with primary ciliary dyskinesia (PW-PCD)

Content validity index (CVI)

Item level (I-CVI)

Scale level (S-CVI)

Declarations

AI-Assisted Language Editing

During the preparation of this work, we used ChatGPT (version GPT-3.5, OpenAI) to optimize the readability and wording of the manuscript. After using this tool, the authors reviewed and edited the content as required and take full responsibility for the content of the publication.

Declaration of interests

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Consent for publication

Consent for publication was obtained from the participants included in the study.

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Author Contribution

Author Contributions (CRediT):SG and NU: Conceptualization, Metodology, Writing – original draft, Writing – review & editing.AB: Writing – original draft, Writing – review & editing.SP and DM: Methodology, Investigation, Data curation.RC and ALQ: Supervision, Project administration.All authors: Writing – review & editing.

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Figures

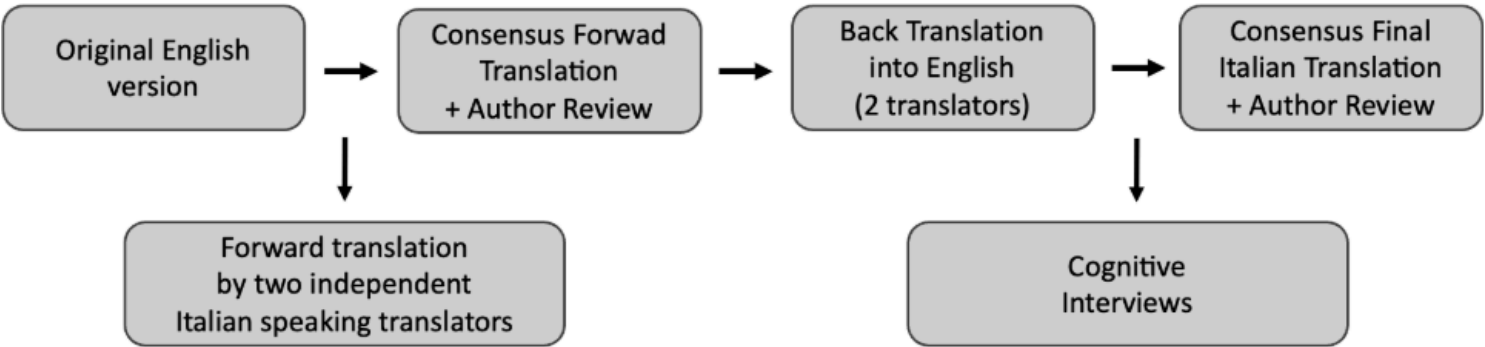


Figure 1

The development of the Italian-language version of the QOL-PCD

Supplementary Files

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