Turk Thorac J 2017; 18: 19-22 DOI: 10.5152/TurkThoracJ.2017.16049

ORIGINAL ARTICLE

Quality of Life Questionnaire for Turkish Patients with Primary Ciliary Dyskinesia

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Abstract

OBJECTIVES: Primary ciliary dyskinesia (PCD) is a major cause of progressive lung disease, and physiological measures do not reflect the impact of the disease on patients' daily symptoms or physical and social functions. We need valid and reliable health-related quality-of-life (HRQOL) measures in PCD to assess the symptoms and daily functions from the patient's perspective. Our aim was to develop a Turkish translation of PCD-specific HRQOL questionnairre to be used as outcomes in clinical trials.

MATERIAL AND METHODS: This study was conducted at the Division of Pediatric Pulmonology, Hacettepe University Faculty of Medicine and the Division of Pediatric Pulmonology, Marmara University Faculty of Medicine. Forward and back translations were performed by three different translators. We recruited participants with PCD from different age groups of both sexes, with an aim to represent a wide spectrum of disease severity and performed the prototype of the translation in these participants.

RESULTS: Five participants from each age group [children (6-12 years), teenagers (13-17 years), adults (18+ years) and parents of children aged from 6 to 12 years] responded to the HRQOL questionnaire. Content analysis of the questions included the following domains depending on age: Respiratory Symptoms, Physical Functioning, Emotional Functioning, Treatment Burden, Ears and Hearing, Sinus Symptoms, Social Functioning, Role Functioning, Vitality, Health Perceptions, School Functioning, Eating and Weight. After the participants have completed the questionnaire, a cognitive debriefing interview was conducted with them, and the results of the interviews were used to form a final version of PCD-specific HRQOL, ready for formal validation.

CONCLUSION: A Turkish translation of PCD-specific HRQOL questionnaire was developed to meet the standards set by international guidelines. This questionnaire is expected to be useful as end points in clinical trials for monitoring health outcomes and for improving clinical decisions.

KEYWORDS: Primary ciliary dyskinesia, quality of life questionnaire, forward translation

Received: *30.11.2016* ***Accepted:*** *29.12.2016*

INTRODUCTION

Primary ciliary dyskinesia (PCD) is a clinical disorder characterized by chronic lower and upper respiratory tract infections associated with impaired ciliary motility. It has clinical and genetic heterogeneity, and it is mostly inherited as an autosomal recessive disease. Its incidence has been reported to be 1 in 4,000-40,000 live births in various studies conducted to determine its frequency in different societies [1,2]. In our country, where the rate of consanguineous marriage is high, the incidence of PCD is also estimated to be high; however, its exact incidence is unknown. An abnormal ciliary structure and function are found in PCD. Recurrent pulmonary infections due to mucociliary clearance dysfunction in the respiratory system; lower and upper respiratory tract diseases such as bronchiectasis, sinusitis, rhinitis, and decreased hearing; infertility due to sperm immotility in men; and decreased fertility and ectopic pregnancy in women are observed. Situs inversus occurs in 30-50% of cases. On the other hand, hydrocephaly, ectopic pregnancies, and heterotaxia are less frequently seen [1-4].

While following PCD patients, parameters specific to the disease are used. Spirometry is an insensitive method for evaluating advanced lung damage. Although high-resolution computed tomography of the lungs is a useful technique in the monitoring of patients, it is impossible to perform tomography for all patients at certain intervals in practice [5-8]. Therefore, a quality of life scale is needed to evaluate the effects of PCD on patients [9].

At present, it is known that the quality of life scales, which are gradually gaining importance, are used as essential tools for assessing patients in clinical research studies as well as in the daily follow-up of chronic disease patients [9-11].

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In this study, the aim was to apply a pretest to PCD patients in various age groups and to the parents of PCD patients between the ages of 6 and 12 years to give the final form of the prototype scale of the quality of life scale for PCD patients (PCD-QOL scale, version 4.3) obtained after translating the English version into Turkish. It is suggested that the use of the Turkish version of these scales prepared for PCD will be beneficial in the follow-up of these patients and in clinical research to be performed on this disease with which an increasing number of people are diagnosed.

MATERIAL AND METHODS

This is a questionnaire study conducted by the Department of Pediatric Chest Diseases in the Faculty of Medicine at Hacettepe University and the Department of Pediatric Chest Diseases in the Faculty of Medicine at Marmara University. Ethical approval was received from the Non-interventional Clinical Research Ethics Board at Hacettepe University. Written informed consent was obtained from patients and their parents while completing the quality of life scale.

The study included children older than 6 years, adolescents and adults who were diagnosed with PCD, and the parents of PCD patients in the age group of 6-12 years. Those who did not complete the questionnaire were excluded. The diagnosis of PCD was established considering the patients' medical histories, clinical and radiological findings, and results of nasal nitric oxide measurements, electron microscopy, video microscopy, and genetic analysis.

In the study, with the permission of Dr. Jane Lucas from the group developing the scale and in accordance with the protocols prepared on this issue, the PCD-QOL scale (April 15, 2014, version 4.3) was first translated into Turkish by two researchers separately and the obtained form was re-translated into English by another researcher. The "prototype scale" was obtained by discussing every step with the developers of the scale on the phone [12].

In this study, the prototype scale was given the final form after being applied as a pretest to patients in different age groups and to the parents of children in the age group of 6-12 years. The plan was to apply it to larger groups including PCD patients and their parents for a validity-reliability study (Figure 1).

Statistical Analysis

For analyzing data, IBM SPSS (IBM Statistical Package for Social Sciences; Armonk, NY, USA) Windows 22.0 was used and descriptive statistical methods (mean, standard deviation, minimum, and maximum) were employed. Results were evaluated at the 95% confidence interval and significance at the level of $p<0.05$.

RESULTS

The patients were divided into groups including the parents of PCD patients, adult PCD patients, adolescent (age group of 13 and 18 years) PCD patients, and pediatric (age group if 6 and 12 years) PCD patients. The Turkish prototype of the scale was applied to five patients from each group in a quiet place in the outpatient clinic. The application of the prototype scale to individuals from each group (5 parents of PCD patients, 5 PCD patients in the age group of 6 and 12 years, 5 PCD patients of the age group of 13 and 18 years, and 5 adult PCD patients) and the cognitive review of responses with a one-to-one interview were performed in the Department of Pediatric Chest Diseases in the Faculty of Medicine at Hacettepe University. Patients of both sexes with different severities of the disease were preferred.

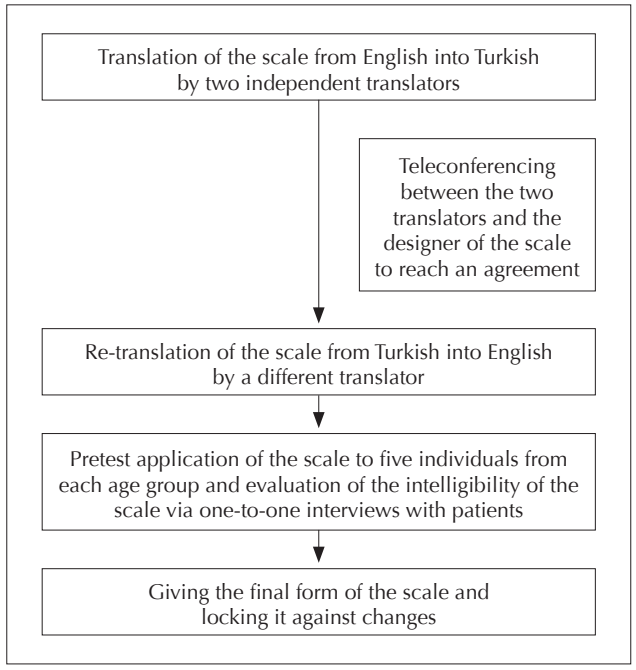

Figure 1. The algorithm of the procedures during the period in which the quality of life scale was given the final form

The following path was followed while applying the prototype scale (Figure 1). The prototype scale was completed by the patients by considering the instructions in the form. After completing the scale, a one-to-one interview was performed and the patients were asked to state what they understood from each question in their own words and what the question recalled at first reading. The patients were asked about the basis of their response to each question. They were asked how they found the questions, whether there were unclear words, and whether there were points that were important for PCD but overlooked. For an item or instruction with alternative words or explanations in the scale, the patients were questioned about which one would have been better. Moreover, the patients were asked about what they felt more comfortable with in their daily language. These views were recorded. In this way, the final form of the QOL-PCD scale that would be used in the actual validity study was obtained.

The quality of life scale included sub-groups involving different situations according to the different ages. These subgroups are shown in Table 1. The scale consisted of 37 items for the patients in the age group of 6-12 years, 41 items for the parents of the patients in the age group of 6-12 years, 43 items for the patients in the age group of 13-18 years, and 49 items for the patients in the age group of 18 years and above.

DISCUSSION

The aim of this study was to translate the English version of the PCD-QOL scale (version 4.3) into Turkish to follow the clinical course of the disease and to evaluate the effects of new treatments on patients. There are a few parameters that have been developed for this purpose in the follow-up of PCD patients.

Table 1. The sub-groups of the quality of life scale in different age groups

Measurement	Children in the age group of 6-12 years	Adolescents in the age group of 13-18 years	Parents of children in the age group of 6-12 years	Adults in the age group of 18 years and above
Evaluation of physical functions	X	X	X	X
Evaluation of emotions	X	X	X	X
Evaluation of treatment	X	X	X	X
Evaluation of ears and hearing	X	X	X	X
Evaluation of respiratory symptoms	X	X	X	X
Evaluation of sinus symptoms	X	X	X	X
Evaluation of social functions	X	X		X
Evaluation of social role		X		X
Evaluation of vitality		X	X	X
Evaluation of health perception			X	X
Evaluation of school functions			X	
Evaluation of nutritional status			X	

The quality of life scale has questions in the sub-groups including the evaluation of physical functions, emotions, treatment, hearing, respiratory symptoms, sinus symptoms, social functions, social role, vitality, health perception, school functions, and nutritional status, which were developed considering the different age groups.

In the quality of life scale for PCD, the aim was to evaluate the effect of respiratory symptoms on the daily life functions of patients by performing a cognitive evaluation with open-ended questions [12]. These questions are similar to those in other scales that are used for patients with cystic fibrosis and bronchiectasis, including respiratory symptoms such as chronic cough, shortness of breath, sputum will be better instead of phlegm, and headache, but the newly prepared quality of life scale also includes questions on symptoms specific to PCD patients such as nasal discharge, nasal obstruction, chronic otitis, and hearing problems [13-16]. The reason for adding these items is to question the symptoms developing in association with decreased mucus clearance from the lungs, nose, sinuses, and middle ear due to ciliary dysfunction [1-3,17].

Cystic fibrosis is a disease presenting with gastrointestinal symptoms and lower respiratory tract symptoms, and ear and hearing problems are rarely seen in this disease. Different from cystic fibrosis, symptoms related to the gastrointestinal system are rare in PCD patients. Despite the absence of pancreatic insufficiency findings in patients, a loss of appetite is observed during the periods of pulmonary exacerbation. These findings are mostly reported by parents. These kinds of questions were asked to the parents of children in the age group of 6-12 years [9].

There are differences in the quality of life scale according to ages. For younger ages, because symptoms for health perception and vitality evaluation are not dominant, there are no questions on these issues. However, it was observed that PCD affected vitality in adolescent patients. Another advantage of this scale is that it allows a comparison of the children's perception of symptoms with their parents' perception.

In a study by Dell et al. [9], it was specified that the development of the quality of life scale for PCD would be effective in the clinical course of the disease [9]. Lucas et al. [11] from England assessed the applicability of the quality of life scale in adult PCD patients, and it was decided that this test could be used after cognitive evaluations [18].

In conclusion, it is suggested that the quality of life scale for PCD patients can be commonly used in clinical studies and in the clinical follow-up of patients.

Ethics Committee Approval: Ethics committee approval was received for this study from the ethics committee of Hacettepe University School of Medicine.

Informed Consent: Written informed consent was obtained from patients who participated in this study.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept - H.U.Ö.; Design - N.E., H.U.Ö., B.K.; Supervision - H.U.Ö., B.K.; Resources -N.E., H.U.Ö.; Materials - N.E., H.U.Ö.; Data Collection and/or Processing - N.E., H.U.Ö.; Analysis and/or Interpretation - N.E., B.K., H.U.Ö.; Literature Search - N.E., H.U.Ö.; Writing Manuscript - N.E.; Critical Review - H.U.Ö., B.K.; Other - N.E., H.U.Ö., B.K.

Acknowledgements: We would like to thank to Prof. Jane Lucas for the contribution of Quality of life Questionnaire preparation; also we would like to thank to Laure Behan for the administration of interviews between the translators, and for the contribution of the develeopment of the last survey form.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study has received no financial support.

NOTE: This questionnaire can be obtained from Professor Dr Uğur Özçelik and Dr Nagehan Emiralioğlu in Hacettepe University Faculty of Medicine Department of Pediatric Pulmonology and Professor Dr Bülent Karadağ in Marmara University Faculty of Medicine Department of Pediatric Pulmonology. This questionnaire can only be used and copy with the agreement and permission of the consortium of developers Prof Dr Jane Lucas, Sharon Dell, Margaret Leigh, Alexandra Quittner. After the validation of English version of this questionnaire Turkish validation will be performed.

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