

PCQoL: A Quality of Life Assessment Measure for Pachyonychia Congenita

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Background: Pachyonychia congenita (PC) is a rare but often debilitating, dominantly inherited disorder. New treatments require more accurate instruments for evaluating changes in the quality of life in these patients.

Objectives: This study was undertaken to develop and validate a quality of life questionnaire for PC patients (PCQoL).

Methods: Relevant factors influencing quality of life in PC patients were identified and incorporated into the well-recognized, general questionnaire, the Dermatology Life Quality Index (DLQI), to establish a disease-specific measure, the PCQoL. Classical test theory (CTT) and Rasch analysis (RA) were used to analyze and validate the PCQoL.

Results: CTT analysis established test–retest reliability and internal consistency for the PCQoL. Concurrent and construct validity for the DLQI and the PCQoL were also validated. Chi-square–based infit and outfit statistics indicated that the Rasch model fits the observed responses very well. RA reconfirmed reliability, internal consistency, reasonable homogeneity, construct validity, and the presence of three RA-based domains.

Conclusion: The PCQoL questionnaire is a measure validated by both CTT and RA. It appears to be a valuable tool in measuring quality of life modifications in PC individuals with keratoderma.

Contexte: La pachyonychie congénitale (PC) est une affection rare mais souvent débilitante, transmise selon le mode dominant. L'arrivée de nouveaux traitements exige l'élaboration d'instruments plus précis d'évaluation des changements qui se sont produits dans la qualité de vie des personnes touchées par la maladie.

Objectifs: L'étude visait à élaborer et à valider un questionnaire sur la qualité de vie des patients atteints de PC (PCQoL).

Méthode: Des facteurs pertinents, ayant une incidence sur la qualité de vie des personnes atteintes de PC ont été cernés, puis intégrés dans le questionnaire général, reconnu en dermatologie, le Dermatology Life Quality Index (DLQI), afin que soit élaborée une mesure propre à la maladie, le PCQoL. La théorie classique des tests (TCT) et l'analyse de Rasch (AR) ont servi à l'analyse et à la validation du PCQoL.

Résultats: L'analyse selon la TCT a permis d'établir la fiabilité de test-retest et la cohérence interne du PCQoL. Ont également été confirmées la validité conceptuelle et la validité concourante du DLQI et du PCQoL. D'après les statistiques *Infit* et *Outfit* reposant sur le khi carré, le modèle de Rasch convient très bien aux réactions observées. De son côté, l'AR a reconfirmé la fiabilité, la cohérence interne, l'homogénéité raisonnable, la validité conceptuelle et la présence de trois domaines fondés sur l'AR.

Conclusions: Le questionnaire PCQoL est un instrument de mesure validé à la fois par la TCT et l'AR. Il semble un bon instrument de mesure des modifications de la qualité de vie chez les personnes atteintes de PC, accompagnée de kératodermie.

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PACHYONYCHIA CONGENITA (PC) is a rare, autosomal dominantly inherited disorder of keratin generally associated with thickened nails, a particularly painful palmo-plantar keratoderma, and a variety of cutaneous cysts. The disorder is associated with mutations in one of five keratin

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genes, *KRT6A*, *KRT6B*, *KRT6C*, *KRT16*, or *KRT17*, with the resultant phenotypes now classified as PC-6a, PC-6b, PC-6c, PC-16, and PC-1.¹ The most distressing symptom and physical limitation in PC is the painful and debilitating plantar keratoderma. This pain limits mobility and is worse with continued standing or walking. It endures for hours at the end of the day, and many patients resort to walking on hands and knees when at home. The more labor intensive the work, the greater are the calluses and resultant pain. Some patients are wheelchair restricted, whereas others use walking aids. To date, no specific questionnaire addresses the characteristic features affecting quality of life (QoL) in PC patients. A number of treatments for PC are nearing the clinical trial phase. Treatments reaching this point include simvastatin, which was shown to have an inhibitory effect on the *KRT6A* promoter, small interfering ribonucleic acid (siRNA), and rapamycin.²⁻⁴ New assessment measures are being developed to document the efficacy of treatment. These include the use of a treadmill to assess improvement in mobility.

Establishing a clinically and statistically valid measure for PCQoL will be valuable in assessing disease-relevant change during planned future clinical interventions. This index will be used to detect when significant change has occurred and has been based on the Dermatology Life Quality Index (DLQI).⁵

Methods

Question Design and Participants

In 2004, the nonprofit organization Pachyonychia Congenita Project established a registry collecting clinical and genetic data. Initially, the questionnaire used in the registry was developed by a joint patient/physician panel and, in addition to demographics, provided detailed information regarding whether and to what extent patients were affected by described clinical features of PC. In 2006, following the initial experience with 100 patients, the questionnaire was shortened. The PC registry now has 467 genetically confirmed PC patients. In addition, 15 patient support meetings have been held, with 510 PC patients participating. We used the questionnaires and patient interviews to structure questions that were relevant to QoL in patients with PC.

Based on the DLQI, there are 12 questions in the PCQoL (Appendix). The PCQoL is meant as a disease-specific tool to assess the response of treatment in PC patients. The emphasis is on the physical domain as this is clearly the most significant determinant of QoL in these patients. As in the DLQI, there are four possible answers.

Responses for each question were assigned scores as follows: “not at all” = 0, “a little” = 1, “a lot” = 2, “very much” = 3. A few questions had a fifth choice of “not relevant,” which was assigned a score of 0.

Patient Selection

The PCQoL questionnaire was administered to PC patients, who were recruited through the Pachyonychia Congenita Project. All participant data included in the analysis were from patients with a confirmed PC keratin mutation. Inclusion criteria were disease duration of at least 6 months, active PC causing self-reported functional impairment, no significant psychiatric comorbidity, no comorbid process with similar effects on QoL, age > 18 years, fluency in English, and ability to provide informed consent. Seventy-six patients participated with the following genetic subtypes of PC: 32 patients with PC-6a, 5 with PC-6b, 2 with PC-6c, 28 with PC-16, and 9 with PC-17.

Ethics Approval

Ethics approval for this study was obtained from University of Saskatchewan Research Ethics Board. The use of the PC registry data was approved by the Western Institutional Review Board (study #20040468).

Statistical Analysis

Traditional analysis based on classical test theory (CTT) and Rasch analysis (RA) were performed to evaluate test–retest reliability, internal consistency reliability, dimensionality, floor and ceiling effects, concurrent validity, and construct validity. Using RA, an item-person map, bubble chart, and category probability curve were generated. CTT analysis was performed using SPSS version 19 (IBM SPSS Statistics, Armonk, NY). RA was performed using *Ministep* software version 3.73 (Winsteps, Lincare & Wright, Chicago, IL).

A summary of the criteria used is provided in Table 1.

Results

Classical Test Theory

Test–Retest Reliability

The test–retest reliability of the control group (intraclass correlation coefficient [ICC] = 0.81, 95% CI = 0.68–0.89, $p < .001$) was slightly less than that of the PC group (ICC = 0.90, 95% CI = 0.84–0.94, $p < .001$). The ICC values exceed

Table 1. Classical Test Theory Validation Criteria

| Statistical Test | PCQoL | DLQI | Interpretation |
|--|---|--|--|
| Test-retest reliability criterion: ICC ≥ 0.70 for a sample size of at least 50 ²⁸ | ICC = 0.90 | ICC = 0.81 | Readministration of the questionnaire at another time point will lead to a similar score if the medical condition is stable |
| Internal consistency criterion: 0.70 < Cronbach α < 0.90 ²⁹ | $\alpha = 0.88$ | $\alpha = 0.75$ | Measures the extent to which all items in the PCQoL relate to one another; all questions are worthy of retention |
| Exploratory factor analysis (EFA) criterion: the Bartlett test should be significant so that the correlation matrix is not an identity matrix ⁶ | 3 domains | 3 domains | To identify the underlying dimensions or factors; to determine if our questionnaire is multidimensional |
| Kaiser-Meyer-Olkin measure of sampling adequacy > 0.5 ⁷ | | | |
| Multidimensionality in EFA criteria: Kaiser rule (K1): retain all components with eigenvalue ≥ 1.0 | | | |
| Scree test: retain all components before the drop ceases toward less steep decline | | | |
| Proportion of variance: factors with explained variance exceeding 5% ⁹ | | | |
| Ceiling and floor effect criterion: if 15% or more of respondents achieved the highest or lowest scores ³⁰ | No effect found | | Indicator for higher QoL (ceiling effect) and lower QoL (floor effect) than the average patient; if no floor or ceiling effect is found, this means that the respondent's condition (QoL) has not improved or has deteriorated beyond what the questionnaire can measure |
| Concurrent validity (PCQoL vs DLQI) | Statistically significant strong correlation between PCQoL and corresponding DLQI scores in each domain ^{10,31,32} | Domain Spearman Correlation between DLQI and PCQoL | This validity compares the new instrument concurrently with an established measure for QoL; since the DLQI is widely used and validated, the strong correlations support PCQoL validity |
| Construct validity (PC vs controls) criteria: Convergent validity: strong item-domain correlation (≥ 0.40) ¹¹ | Highest correlation of questions with domains to which they were assigned; weak correlation of questions with all other domains | 1 0.989 2 0.975 3 0.953 | The extent to which PCQoL scores relate to each other and their domains; construct validity was met as questions correlated highest to the domain they belonged to and weaker in all other domains |
| Divergent validity: weak correlation between item and all other domains ^{10,33} | | | |

DLQI = Dermatology Life Quality Index; ICC = intraclass correlation coefficient; PC = pachyonychia congenita; PCQoL = quality of life assessment measure for pachyonychia congenita.

Table 2. Cronbach α Coefficient for Control and PC Groups

| Group | First Response | α if Item Deleted | Second Response | α if Item Deleted | All Responses | α if Item Deleted |
|---------|---------------------|--------------------------|--------------------|--------------------------|--------------------|--------------------------|
| Control | 0.758 ($n = 101$) | 0.782 (Q3) | 0.784 ($n = 61$) | 0.821 (Q3) | 0.752 ($n = 61$) | 0.788 (Q3) |
| PC | 0.873 ($n = 75$) | 0.876 (Q11) | 0.860 ($n = 61$) | 0.866 (Q11) | 0.877 ($n = 61$) | 0.881 (Q11) |

PC = pachyonychia congenita; Q = question.

the minimum threshold of ICC ≥ 0.70 (see Table 1), confirming the repeatability/reliability of the PCQoL questionnaire. Thus, if a patient's medical status is stable, when the questionnaire is administered at two different time points, the patient's scores should be similar.

Internal Consistency Reliability

The Cronbach α coefficient scores were marginally higher for the PC group than for the control group but within the acceptable range for internal consistency (Table 2). This test determines the internal consistency of responses between questions of the PCQoL for both groups. The column " α if Item Deleted" shows the highest achievable value of α that can be obtained when a specific question is dropped and is indicated in brackets in Table 2. Exclusion of any of the 12 items in either group did not increase α significantly, suggesting that all questions are worthy of retention.

Dimensionality

The validity of exploratory factor analysis (EFA) was verified first by the Bartlett test, which was found to be highly significant ($p < .001$).⁶ The Kaiser-Meyer-Olkin measure of sampling adequacy of 0.82 and 0.78 for PCQoL and DLQI, respectively, exceeds 0.5 in both cases, indicating that factor analysis is appropriate.^{7,8} Based on the criteria, an eigenvalue above 1.0, a scree plot with a sharp drop in eigenvalues, and items with a factor loading of 0.4 or more (see Table 1), three factors were revealed in both the DLQI and the PCQoL. This suggests that both questionnaires are multidimensional.

Floor and Ceiling Effects

These effects are present when respondents believe that their condition (QoL) has improved or deteriorated beyond that which the questionnaire can measure. There were no floor or ceiling effects for factors 1 and 2 in the PCQoL (see Table 1). Factor 3 was found to have no ceiling effect; however, some floor effects were noted, suggesting overall good QoL.

Validity

The following types of validity were analyzed for the PCQoL scale.

Concurrent validity. A statistically significant strong Spearman correlation of ≤ 0.95 was found between PCQoL first response scores and corresponding DLQI scores in each factor, confirming concurrent validity for the PCQoL. Given that similar scores were reported and the DLQI is a valid dermatologic questionnaire, this validates the PCQoL.

Construct validity All questions in the PCQoL showed the highest correlation (≥ 0.40) with factors to which they were assigned than with other factors.⁹⁻¹¹ This supports construct validity for the three factors determined by EFA.

Item Response Theory: Rasch Model

Model Data Fit

Values of the person and item infit and outfit statistics were found to be within the acceptable range (mean square residual [MNSQ] 0.5 to 1.5; standardized mean square residual [Z-standardized (Zstd)] -2.0 to 2.0) (Table 3).^{12,13} The chi-square probability ($p < .001$) indicates that our questionnaire was measuring a dominant rather than a unidimensional scale. This suggests that patients and questions were both representative of the disease process.

Internal Consistency

The item and person reliability indices were 0.97 and 0.87, respectively, verifying PCQoL as a reliable instrument (see Table 3). The person reliability index is an indicator for overall precision. High values suggests that the instrument is well targeted for PC patients. The high item reliability index indicates that the responses to items are consistent.

Construct Validity

Construct validity was determined by the item separation index of 6.24 and a person separation index of 2.61 (see

Table 3. Overall Rasch Model Fit Summary Statistics

| | Item | | | | Person | | | |
|--------------------|-------|------|--------|------|--------|------|--------|------|
| | Infit | | Outfit | | Infit | | Outfit | |
| | MNSQ | Zstd | MNSQ | Zstd | MNSQ | Zstd | MNSQ | Zstd |
| Mean | 1.05 | 0.0 | 0.99 | -0.3 | 0.97 | -0.1 | 0.99 | -0.1 |
| Standard deviation | 0.37 | 2.0 | 0.31 | 1.4 | 0.60 | 1.3 | 0.85 | 1.3 |
| Maximum values | 1.93 | 3.2 | 1.69 | 2.0 | 3.99 | 4.8 | 6.24 | 4.9 |
| Minimum values | 0.57 | -3.3 | 0.63 | -2.3 | 0.29 | -2.2 | 0.27 | -2.2 |
| Separation index | | 6.24 | | | | | 2.61 | |
| Reliability index | | 0.97 | | | | | 0.87 | |

MNSQ = mean square of measure; Zstd = Z-standardized.

Table 3). The item and person separation indices suggest that items and persons are well separated or distinct. Our separation index values indicate that the PCQoL has good construct validity.

Item Data Fit

Fit statistics, difficulty measures, and the standard error for each item were determined. Infit and outfit MNSQ indices for all items except question 11 (infit: MNSQ 1.93, Zstd 3.20; outfit: MNSQ 1.69, Zstd 1.60) are in the acceptable range.¹³ It indicates that responses based on guesses and careless mistakes have no significant effect on our results.

Item-Person Fit

Overall, the item-person (Figure 1) map shows fairly good spread of persons and items along the scale, indicating a good overall match between item difficulties and person abilities. The bubble chart (Figure 2) illustrates the fitness of person ability and item difficulty measures simultaneously, along with the magnitude of measurement error. Question 1, located leftmost in the chart, has the most regular answer pattern and is marginally overfit to the model. Greatly and poorly affecting QoL items were found to be questions 8 and 11.

Response Categories Structure

The criteria for differences between response categories hold well for all categories except between categories 2 and 3, which are 0.95 logit apart (Figure 3).¹⁴ Although the Lincare criterion was not fully satisfied, there is no need to reorganize the four rating scales because the average measures and threshold were found to increase monotonically with

categories, the probability curve shows an ordered distinct pattern with good transition between all categories, and infit and outfit MNSQ for each category were within the acceptable range.

RA-Based Principal Component Analysis of Residuals

Principal component analysis of residuals (PCAR) identified three domains for the PCQoL. Domain 1 contains questions 1, 2, and 5. Domain 2 contains questions 6, 7, and 8. Domain 3 contains questions 3, 4, 9, 10, 11, and 12. Thus, our questionnaire is multidimensional.

Validity

All questions showed the highest correlations (≥ 0.40) with domains to which they were assigned than with other domains.¹¹ Correlations between an item and the other dimensions were low, confirming convergent and divergent validity.^{9,10}

Validation of Domains Based on RA

To confirm the unidimensionality of the Rasch model domains, variance, eigenvalues, and convergent and divergent validity were analyzed. The Rasch component explained 74.5%, 68.1%, and 60.3% of the variance for each domain with first contrast eigenvalues of 1.7 or less.^{15,16} Thus, all three Rasch domains satisfy the criteria for unidimensionality.

Discussion

Disease-specific QoL measures are important in focusing attention to those aspects of life that are particularly pertinent to a specific disease. Treatment success is



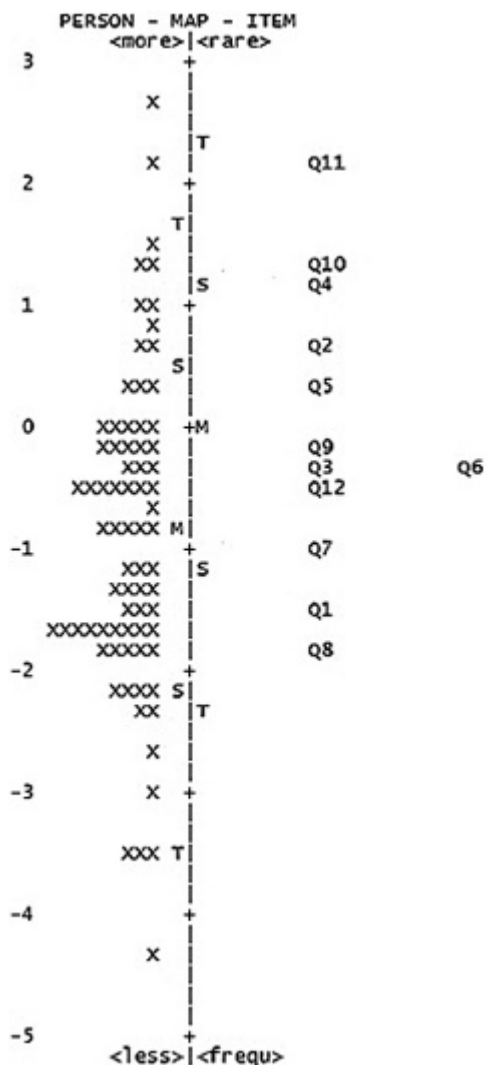


Figure 1. Item-person map of 12 items (PCQoL questions). The distribution of Rascha analysis-calibrated participant scores is on the left side of the vertical line, and items are located on the right. M = the person and item mean; S = 1 SD away from the mean; T = 2 SD away from the mean; X = a patient.

assessed not only by measuring changes in a particular disease state but also how the QoL of the sufferer has improved. Several clinical trials for PC are being considered, and a disease-specific QoL measure has been developed and validated in this study.

Our CTT analysis shows that test-retest reliability for both control and PC groups demonstrated very good reproducibility. Thus, if a patient’s clinical condition remains the same and the questionnaire is readministered, a very similar score will be obtained. The Cronbach α test confirmed or validated the internal consistency for the PCQoL. It also suggested that all 12 questions are worthy of retention. Our construct validity finding of three factors

is in agreement with our RA findings as well as with other reports.¹⁷ Floor and ceiling effects demonstrated an overall good spread for QoL assessment, indicating that the participant’s condition (QoL) did not improve or deteriorate beyond the measurement capacity of the PCQoL. Concurrent validity was verified by comparing PCQoL scores to the DLQI, measured essentially at the same time. Given that the DLQI is widely used and validated, these associations support PCQoL validity.¹⁸ Construct validity was confirmed because questions correlated highest with the factor to which they belonged.

To overcome several limitations of CTT, modern measurement methods, commonly referred to as “item response theory” (IRT), are employed.^{19–21} One of these models, the Rasch measurement model, estimates the “goodness of fit” between item difficulty and person ability.²² Rasch models have been used in the validation of a series of questionnaires and are essential in evaluating measurement tools in specific disease cohorts.^{23,24}

Overall, all items of the PCQoL scale fit to the Rasch model very well. The standard deviation for person estimates is slightly larger than the corresponding item estimates, indicating a slightly wider spread within person measures. Internal consistency demonstrated the PCQoL to be a reliable instrument (see Table 3). Our item reliability value of 6.24 suggests excellent reproducibility when these patients are given a similar QoL questionnaire. A person separation index of 2.61 indicates that patients have reasonable spread along the scale (ie, are well represented). Our separation index values indicate that PCQoL has good construct validity. Person reliability was lower than item reliability, which may be due to the inherent variability in individualized behaviors specifically in a large sample with few items.²⁵

Item hierarchy of PCQoL in Figure 1 indicates that question 11 (“Over the past week, how much has your skin caused any sexual difficulties?”) least affects QoL, whereas question 8 (“Over the past week, has the pain in your feet made it difficult for you to do any sport?”) greatly affects QoL (see Figure 1). Overall, we found that personal relationships were the least affected by PC, whereas leisure activities were affected the most. To ensure appropriate ordering of our scoring system, patterns of multiple response categories were analyzed by category probability graph and Andrich threshold estimates for the transition from one rating scale to another.²⁶ Our analysis showed an ordered set of response thresholds. There is no evidence of inappropriate use of the scale categories, no underuse categories, and no inversions. The scoring system was found to be reasonably spaced for all items, indicating that

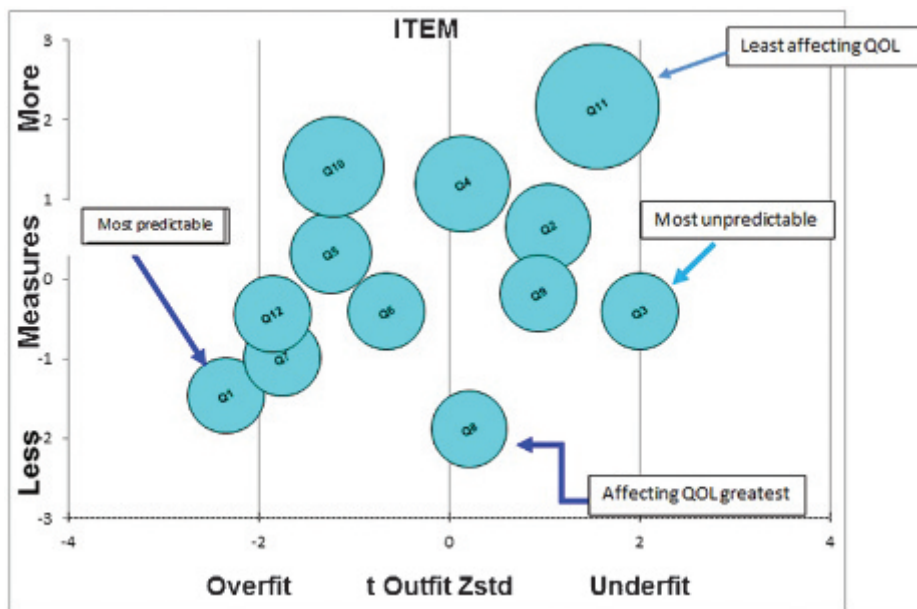


Figure 2. Bubble chart showing outfit mean square standard statistics versus item measure. The t Outfit Zstd is a standardized indicator of how well item performances fit the Rasch model's requirements. QOL = quality of life.

the response design, that is, four options for each question, was appropriate (see Figure 3). Thus, collapsing of response categories is not required.

Factor analysis demonstrated a three-factor structure that broadly corresponds to the three domains determined by PCAR. The difference in item groupings is likely due to different methodologies. Conventional factor analysis identified factors within a correlational matrix, whereas PCAR is based on the existence of the multidimensionality in the residuals.

Our overall analysis supports the PCQoL as a useful measure for assessing QoL in PC patients, particularly those with significant keratoderma. It may not be the most useful tool to assess QoL in PC patients who have *KRT17*

mutations, where large disfiguring cysts and scarring are prominent. The PCQoL has better validity and responsiveness for QoL changes than the DLQI for PC patients. This study has shown that a brief, direct, interviewer-administered PCQoL questionnaire can provide useful data on the QoL of patients with PC. Recently, Fu and colleagues examined PC genotypes and subsequent physical limitations on a Likert scale.²⁷ They found that clinical severity depends on the genotype.²⁷ However, this study is the first to formally create and critique a questionnaire for patients with PC. We hope that our questionnaire will be used as an objective marker for the treatment of PC patients in the clinical setting.

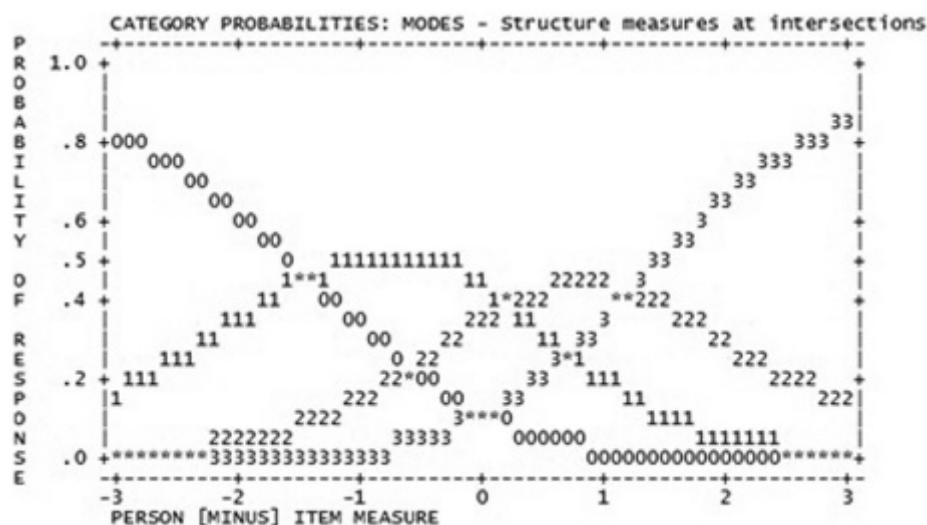


Figure 3. Probability curves of the PCQoL four-category rating scale.



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Appendix. PCQoL Questions and the DLQI Equivalents

| <i>PCQoL Question</i> | <i>DLQI Equivalent</i> |
|---|--|
| Q1. Over the past week, how much pain have you had in your feet? | Over the last week, how itchy, sore, painful or stinging has your skin been? |
| Q2. Over the past week, how much alcohol or medication did you have to use for your PC pain? | None |
| Q3. Over the past week, how embarrassed or self-conscious have you been because of your PC? | Over the last week, how embarrassed or self-conscious have you been because of your skin? |
| Q4. Over the past week, how much have your nails interfered with your daily work? | None |
| Q5. Over the past week, how much has the pain in your feet disturbed your sleep? | None |
| Q6. Over the past week, how much has your PC interfered with you going shopping or looking after your home or garden? | Over the last week, how much has your skin interfered with you going shopping or looking after your home or garden? |
| Q7. Over the past week, how much has the pain in your feet affected any social or leisure activity? | Over the last week, how much has your skin affected any social or leisure activities? |
| Q8. Over the past week, has the pain in your feet made it difficult for you to do any sport? | Over the last week, how much has your skin made it difficult for you to do any sport? |
| Q9. Over the past week, has your PC prevented you from working or studying? | Over the last week, has your skin prevented you from working or studying? |
| If “No,” over the past week, how much trouble has your PC been a problem at work or studying? | If “No,” over the last week, how much has your skin been a problem at work or studying? |
| Q10. Over the past week, how much has your skin created problems with your close friends or relatives? | Over the last week, how much has your skin created problems with your partner or any of your close friends or relatives? |
| Q11. Over the past week, how much has your PC caused any sexual difficulties? | Over the last week, how much has your skin caused any sexual difficulties? |
| Q12. Over the past week, how much of a problem has it been to take care of your PC? | Over the last week, how much of a problem has the treatment for your skin been, for example, by making your home messy or by taking up time? |

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DLQI = Dermatology Life Quality Index; PC = pachyonychia congenita; PCQoL = quality of life assessment measure for pachyonychia congenita.

