BURDEN OF ALBINISM - CREATION OF A QUESTIONNAIRE

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BACKGROUND

Albinism comprises a group of autosomal recessive diseases that are characterized by poor vision and a variable hypopigmentation phenotype. The prevalence of all known forms of albinism appears to be 1 : 17 000 newborns (with a range of 1:10 000–20 000).

A comprehensive literature review showed that no tool can assess the burden experienced by individuals who present with albinism, although such a tool is needed and would be beneficial for clinicians and patients alike. Additionally, this tool would also allow the impact of albinism care to be evaluated.

Within the framework of the RADICO project, the FIMARAD group began an initiative to create a specific questionnaire designed to evaluate the burden created by albinism.

METHOD

The questionnaire was devised using standardized methodology for developing and validating questionnaires on the quality of life of subjects (Seidenberg et al., 1994), according to the following chronological structure: conceptual phase, development phase, and then validation phase.

A multidisciplinary working group was assembled, including experts on questionnaire design and development, dermatologists specializing in care for patients with albinism, and representatives of the Genespoir association.

Validated questionnaires : *Short-Form-12 (SF12), the Body Image States Scale (BISS) and Daily Life Quality Index (DLQI)* were also administered to ensure external validity.

RESULTS

Based on an initial verbatim report, the workgroup compiled a list of items that were transcribed and reformulated into questions. During this phase, 65 items were defined, reorganized and regrouped according to the content. Then, they were reduced to 24 questions (so-called « conceptual » questionnaire). A total of 87 patients attending the clinic were invited to participate, of whom 63 agreed to participate.

Of these 63 individuals, 58% were females and 42% were males. The mean age was 43.98±19.4 years [38.7-49.25]. No significant differences by gender (p=0.13) were observed. Of the participants, 46.2% of men and 52.8% women were employed. Regarding the diagnostic delay, 23.8% declared having experienced a misdiagnosis; this diagnosis had a clinical origin for 46% of participants and a genetic origin for 33% of them. Of these , 58% were women. During the validation phase, principal component analysis (PCA) was conducted on the 24 items, which allowed the questionnaire to be reduced to 20 questions [Q].

The standardized regression coefficients were all greater than 0.5 for their corresponding factors. Based on their normalized regression coefficients, each group of questions was linked to one of the following four dimensions, with each dimension consisting of at least three questions : « Live with » (8 Q), « Daily life » (3 Q), « Resignation » (3 Q), and « Fear of the future » (6 Q). All dimensions correlated well with the overall BoA score. Cronbach's α was 0.92 for the entire BoA scale, confirming excellent internal coherence. Intradimensional coherences all demonstrated excellent reliability (α> 0.65).

To verify the external validity, the correlation coefficients of the questionnaire were also calculated with the following validated questionnaires : SF12, BISS, and DLQI. A strong correlation was found; thus, external validity was confirmed.

The BoA score for each situation confirmed the sensitivity of the questionnaire; indeed, those who experienced a misdiagnosis had a Burden score that was significantly higher than those who had not. Similarly, those who had prominent albinism and those who had a significant alteration of their visual acuity also had significantly higher Burden scores than those who did not fit into these categories.

	PCS	MCS	DLQI	BISS	GLOBAL SCORE	SCORE FACTOR 1	SCORE FACTOR 2	SCORE FACTOR 3	SCORE FACTOR 4
PCS (SF12)	1.00000	0.06986	-0.56161	0.17791	-0.52808	-0.27096	-0.65457	-0.34930	-0.14496
MCS (SF12)	0.06986	1.00000	-0.48964	0.55391	-0.54514	-0.53865	-0.38715	-0.57915	0.05643
DAILY LIFE QUALITY INDEX	-0.56161	-0.48964	1.00000	-0.48176	0.67974	0.52040	0.65349	0.52183	0.12680
BODY IMAGE STATES SCALE	0.17791	0.55391	-0.48176	1.00000	-0.61403	-0.64792	-0.38433	-0.57044	-0.06093

DISCUSSION

This questionnaire represents the first specific assessment tool for evaluating the burden of albinism. It is easy to use and relatively quick to complete, which will allow the burden to be evaluated over time with a reproducible questionnaire. This questionnaire can also be used before and after treatments to evaluate their effect.

CONCLUSION

To ensure that this questionnaire can be used by as many people as possible, cultural and linguistic validation in US English was conducted with the original French version.

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