Proposal of a Portuguese Tool for Quality Assessment of Genetic Counselling: a New Tool for Healthcare Professionals

Proposta de Uma Escala Portuguesa para a Avaliação da Qualidade do Aconselhamento Genético: Uma Nova Ferramenta para os Profissionais da Saúde

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ABSTRACT
Introduction: The lack of tools for quality assessment of genetic counselling is recognized in national and international studies. The correlation of quality of healthcare practice with greater satisfaction of patients and affected families is also well established. The present study describes the development and validation of the first Portuguese scale for quality assessment of genetic counselling practice. It encompasses a proposal of a new tool for the evaluation of the process by professionals.
Material and Methods: The definition of an initial pool of items and their organization was based on a literature review and identification of the main genetic counselling dimensions as well as the theoretical dimensions of the Reciprocal Engagement Model. After a pre-test validation the scale was submitted to psychometric validation using a sample of 30 participants who evaluated 81 genetic counselling sessions.
Results: Based on statistical and empirical criteria the best items were selected. The final 50 items- version comprises five dimensions: education, counselees' characteristics and decision-making, therapeutic relationship, effects of the process on the counselees and services organization.
Discussion: Results showed consistent psychometric properties of the scale supported on theoretical and practice concepts of genetic counseling. The reduced number of participants involved in psychometric validation is a limitation of the study, which reflected the reduced number of professionals in genetic healthcare services.
Conclusion: The scale proposed at this study is a novel and multidimensional instrument that aimed to contribute to the improvement of genetic counselling practice in Portugal.
Keywords: Genetic Counseling; Genetic Services; Outcome Assessment (Health Care); Portugal; Psychometrics

RESUMO
Introdução: A falta de ferramentas para avaliar a qualidade do aconselhamento genético é uma limitação reconhecida em estudos nacionais e internacionais. É conhecida também a relação da qualidade da prática nos cuidados de saúde com uma maior satisfação dos doentes e das famílias afetadas. O seguinte estudo apresenta a construção e validação da primeira escala portuguesa para avaliação da qualidade da prática do aconselhamento genético. Engloba uma proposta de ferramenta para a avaliação do processo pelos próprios profissionais.
Material e Métodos: Iniciou-se este estudo pela revisão da literatura e identificação das principais dimensões do aconselhamento genético. De seguida, procedeu-se à elaboração dos itens e à sua organização mediante as dimensões teóricas do Modelo do Envolvimento Recíproco. Após um pré-teste alcançou-se a versão que foi proposta para validação a uma amostra de 30 participantes, que avaliaram 81 sessões de aconselhamento genético.
Resultados: Através de critérios estatísticos e empíricos selecionaram-se os melhores itens, ficando a escala constituída por 50 itens. Esta versão da escala compreende cinco dimensões: educação, características do consultando e tomada de decisão, relação terapêutica, efeitos do processo no consultando e organização do serviço.
Discussão: Os resultados mostraram que se trata de uma escala válida, com características psicométricas consistentes e fundamentada em simultâneo do ponto de vista teórico-prático do aconselhamento genético. O número reduzido de participantes envolvidos na validação da escala constitui uma limitação, que reflete o número reduzido de profissionais a exercer nesta área dos cuidados de saúde.
Conclusão: A escala proposta neste estudo é um instrumento pioneiro, multidimensional que pretende contribuir para a qualidade da prática do aconselhamento genético em Portugal.
Palavras-chave: Aconselhamento Genético; Avaliação de Resultados (Cuidados de Saúde); Portugal; Psicometria; Serviços de Genética

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INTRODUCTION

Genetic counselling has gradually emerged as a new interdisciplinary healthcare field, particularly within the departments of genetics.[1] Internationally and particularly in Europe, an increasing recognition of the imperative need for providing genetic counselling at the time of genetic testing has occurred, aimed at a safe practice and informed decision-making.[2,3]

In Portugal, genetic counselling has been delivered by Medical Genetics specialists since its development as a specialty, which was mainly linked to the implementation of the Predictive Testing Protocol (*Protocolo de Testes Preditivos*) in major late-onset neurological diseases, with a particularly high prevalence in Portugal.[4,5] The need to provide genetic counselling to risk individuals was therefore defined in the 90s and information support, decision making and psycho-social support are the key elements of the protocol. Predictive (pre-symptomatic) genetic testing allowing for the identification of mutations linked with these incurable diseases is probably a unique setting to the professionals within the departments of genetics in Portugal for learning and skill acquisition. This is still an unrecognised occupation in Portugal even though an academic pathway related to a professional master's degree has been already developed for non-physician healthcare professionals aimed at training in specific skills regarding genetic counselling.[5]

Increasing national research in counselling and psycho-social genetics has been found over the past few years as a starting point for the improvement of clinical practice in healthcare services.[6-11] Current qualities and constraints in the area of genetics both in primary care and in specialty departments have received growing attention by physicians, researchers and healthcare regulation authorities.[5,8,12,13] Human resource shortage and the need for training in specific skills are the major limitations and therefore genetic counselling has been mainly ensured by genetic physicians, neurologists and obstetricians, constrained by long waiting lists in some departments of genetics.[13] Limitations regarding the definition of indicators of effectiveness and expertise as well as the lack of good practice national recommendations in genetic counselling have been described by genetics professionals in a recent Portuguese study[12] and this reality is not limited to Portugal.[14]

Few instruments are generally available for quality assessment in genetic counselling[15-19] and those that are mostly used are mainly focused on the possible effects of counselling[20] rather than on the process components aimed at ensuring patient outcomes.[16,21] Apart from the fact that few scales have been developed within the area of genetic counselling, it is worth mentioning that they are all focused on very specific areas and none has ever been adapted to the Portuguese context.[19,22,23] In fact, scales aimed at the separate assessment of satisfaction, awareness, decision making, coping strategies and family communication, among others, have traditionally been used.[15,23] Most of these instruments are mainly related to psychological or medical instruments that were adapted to genetic counsel-

ling and therefore none is comprehensive enough for the full assessment of the relevant characteristics regarding quality, counselling process and outcomes.[15,23] The lack of conceptually reasoned and empirically validated assessment scales prevents from an adequate service audit, as well as the appropriate study of its limitations and potentialities and the comparison between different models of provision.[24,25] For this reason, the development of instruments aimed at the assessment of clinical practice and measurement of expertise parameters is on its own a contribution to improvement.[14]

The need for the development of an assessment scale of genetic counselling that could fill in those gaps has emerged in Portugal from the present situation. This study is aimed at the description of the construct validity of the first Portuguese scale for quality assessment of genetic counselling, in addition to the description of its psychometric properties including principal component analysis, internal consistency and dimension correlation.

MATERIAL AND METHODS

Methodological approach: search for theoretical and practical model of the scale

A theoretical model is useful as a guide to the configuration of the basic structure of the scale and this was the initial stage of the study, focused on the search for an appropriate theoretical framework. In addition, a theoretical and practical model allows for the definition of tenets and goals of counselling which, in turn, may guide the identification of which dimensions of genetic counselling should be studied.[20]

Genetic counselling was historically based on medical, educational and mental health models.[26,27] Counselling has been considered as a kind of genetic social work aimed at the presentation of a psycho-social approach to genetic conditions, according to McCarthy Veach *et al.* (2003) (originally), Sheldon Reed and Joan Marks.[27] This approach was focused on the recognition that patients exist within a family and social system and patient assessment in a vacuum makes no sense. Kessler (1997) has subsequently described teaching and counselling models as the two professional approaches to genetic counselling.[28,29] The teaching model, imported from medicine, was mainly aimed at providing information to patients and facilitate their own decision making. In addition, the counselling model, derived from the area of mental health, was based on a patient-centred approach.[29]

The Reciprocal-Engagement Model[28] was developed in an effort to obtain the first definition of genetic counselling, with contributions from all the previous models and was selected as theoretical framework for the scale that was designed in this study (Fig. 1).

This model was developed from a consensus conference with a group of professionals with longstanding expertise in the area of genetic counselling and the following basic tenets were considered: information is crucial,

relationship is integral to counselling, patient autonomy must be supported, patients are resilient and their emotions matter.[28] Different goals, strategies and specific behaviours are associated with each tenet of the model, aimed at reaching positive outcomes (Appendix 1: https://www.actamedicaportuguesa.com/revista/index.php/amp/article/view/9997/5454).

As far as we know, this scale describes the first experience in the use of this model as a theoretical and practical baseline reference for the development of an instrument for practice assessment.

Validity of the scale: pre-test procedures and content validity

The first item formulation was based on bibliographic research aimed at the identification of major dimensions and the way some subjects were described in literature. In addition, Portuguese studies regarding the perception of professionals and patients on the relevant criteria for a quality practice have been analysed.[11,12,21,25] Cultural validity of the quality indicators of genetic counselling provided to the Portuguese population (counsellors and patients) was based on those previous qualitative studies and the first version of the scale emerged from the current context of healthcare in Portugal.

Subsequently, the scale format has been defined, as well as the different response options. The first version was subsequently submitted to exploratory pre-test involving experts in the area. Cognitive interviewing was used in pre-test validity of the initial scale[30] and a five-expert panel has been involved (two geneticists, two genetic counsellors and one psychologist). A cognitive interview consists of a process of administration of questions aimed at obtaining additional information on the meaning of the items within a scale
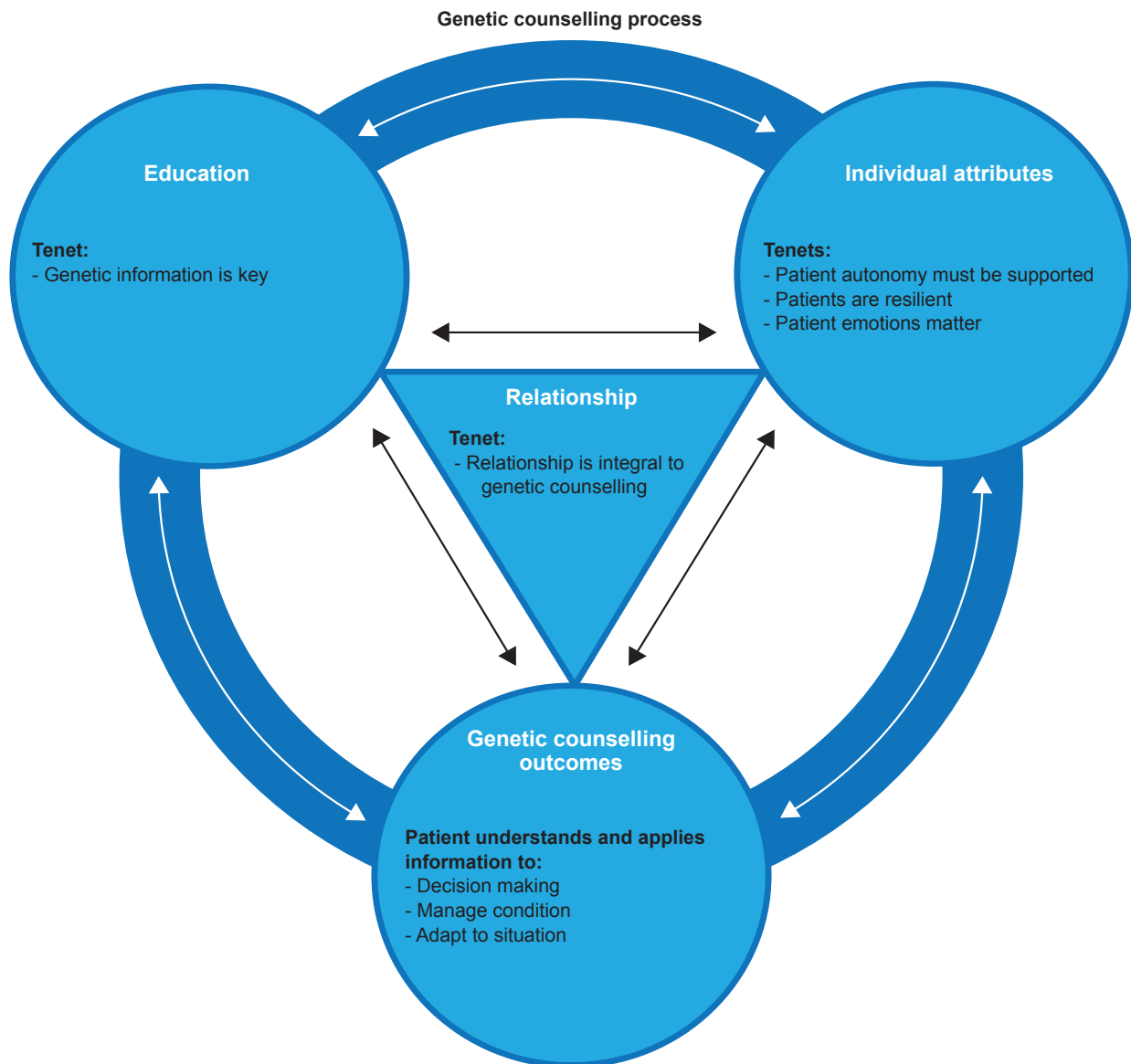

Figure 1 – Reciprocal-Engagement Model28- dimension and tenet layout
Each element interacts with every other element. None stand alone or work in isolation.

and understanding the thought process of the interviewed.[31]

An item assessment has been mostly suggested, according to criteria such as formulation of goals, simplicity, relevance, range, clarity, response alternatives, ambiguities and constraints. In whole, the scale was analysed regarding instruction adequacy, detection of misconceived items, sensitivity of questions and items, quality of response options, range of contents and relevance for practice assessment.[32] Content was subsequently analysed according to the Portuguese context and evidences of the clinical practice.

Psychometric validity: data collection procedure

A list of all the departments providing genetic counselling consultations in Portugal was developed. Subsequently, departments were contacted by email and were visited upon agreement on the participation in the study and copies of the scale and an information sheet were provided, whilst its goals and all the ethical issues regarding data confidentiality were discussed.

Only the professionals actually involved in genetic counselling consultation were eligible for the study: geneticists, oncogeneticists and genetic counsellors. Due to a small target population and the need for a credible statistical analysis, each professional was asked to complete the scale three times, focused on three sessions. A sealed envelope has been provided together with each set of scales and each participant was asked to seal it and return it to us.

Data analysis

Data analysis was carried out by using the SPSS version 24 software and principal component, internal consistency and item correlation have been analysed and aimed at construct validity of the scale, considering a 0.05 level of significance.

RESULTS
First version of the scale

Five item sets regarding quality assessment of practice, 75 items in total, were included in the scale, representing relevant aspects and dimensions of genetic counselling. Items were organised based on the Reciprocal-Engagement Model and its dimensions. The first set of items concerned the tenet of relevance of information in genetic counselling practice, within the 'Education' dimension. The second set was related to the approach of emotional and motivational issues, to patient's individual characteristics and the support to decision making, regarding the 'Individual Attributes' dimension of the model. The third set of items was related to relational and communication issues in genetic counselling, to the tenet that patient concerns and doubts are all the more effectively approached, the stronger, more empathetic and respectful towards patient's autonomy the counsellor-patient relationship will be, in order to respond to all medical, educational and psycho-social issues, within the 'Relationship' dimension. The fourth set of items was focused on patient outcomes of counselling, related to the 'Genetic Outcomes' dimension. Finally, the fifth set of items was related to 'Department's Activity' which was considered as a crucial aspect for practice assessment, even though not included in the model.

In addition, age and gender of the participants have been collected. A five-point Likert-type response scale has been used, in which 1 corresponded to a very low level of the evaluated aspect and 5 to a very high level, with the option for a 'non-applicable' response (considering the diversity of typologies of genetic counselling consultation).

The analysis of pre-test cognitive interviews allowed for the definition of different recommendations for the revision of the scale. Items were assessed by the experts according to their compliance with the target population or cultural context, a clear item formulation, the consistency of certain words or sentences, the possible redundancies, among other aspects. Some changes were made in the formulation of words and expressions as a result of this methodological procedure, as well as in the presentation format of some items. Some items were removed and other added, while a 74-item total remained.

Psychometric validity of the scale
Participants

Seven departments of genetics and other 10 hospital institutions with the provision of genetic counselling currently exist in Portugal. Five from these agreed with the participation in the study, as well as four other institutions with the provision of genetic counselling, which were used to enhance the small number of professionals within the departments of genetics. Therefore, 30 participants from eight hospital institutions have participated in the study, with ages ranging 26 to 60 (M = 39.38; SD = 10.38, 20 female: 66.7%).

Great interest and good feedback regarding the study and the scale were shown by most departments, with a certain curiosity towards the theoretical model on which the study was based. This positive acceptance was not only found during the visits to the departments of genetics as well as in national and international meetings in which the study was presented.

Principal component and the internal consistency analysis

At this stage, a separate analysis of each dimension has been considered. It is worth mentioning that the three first dimensions relate to criteria that should be addressed by genetic counselling practice, including information relevance, the approach to emotional issues and individual characteristics as well as relational and communication aspects. The fourth and fifth dimensions are different aspects, involving the approach to the outcomes of genetic counselling and the activity indicators of the departments, respectively. Therefore, the scale is not intended to provide a global measurement of these aspects that are actually different.

Principal component analysis was based on sample adequacy and factor rotation capacity, by using the

Kaiser-Meyer-Olkin (KMO) coefficient and the Bartlett test of sphericity. KMO values > 0.50 have been obtained (0.57 ≤ KMO of all the dimensions ≤ 0.86) therefore showing an adequacy of the sample for the principal component analysis in all the dimensions. A <0.001 value of the test of sphericity has been obtained in all the dimensions, showing a good correlation between the items in each dimension.[33]

The principal component analysis of each dimension has been carried out, involving an exploratory analysis with no pre-set number of factors. This technique was used in order to explore the inter-relationships between variable data and whether these have the same underlying concept/factor. Oblimin rotation with Kaiser normalisation has been used (Appendix 2: https://www.actamedicaportuguesa.com/revista/index.php/amp/article/view/9997/5455). The fact that each dimension had correlated factors was also taken into consideration, which seems more characteristic within the area of Psychology, given the interdependence of psychological dimensions.[32]

Therefore, principal component analysis in factors with a value > 1 and saturation values ≥0.40 has been considered in order to confirm the structure of the scale and to reduce the number of items (Appendix 2).[34] Inter-item correlation analysis has also been used, as well as theoretical and practical relevance of each item.

Internal consistency of each dimension was shown by Cronbach's alpha coefficient. All the dimensions in the initial 74-item version of the scale showed good internal consistency (Cronbach's alpha 0.85-0.96) and good results in dimensional analysis (values of cumulative explained variance 67.11% - 78.81%); nevertheless, the number of items has been reduced in order to obtain a better practical functionality of the scale. A factor saturation >0.40 has been found in all the items, ensuring a good communality of statements.[34,35] The principal component analysis has led

to the exclusion of two items as these saturated in two different factors. Additionally, another 22 items were removed (24 items were removed in total), through the analysis of the correlations and the theoretical relevance.

Final version of the scale and its rating

A 50-item short-version of the scale has been considered (Appendix 3: https://www.actamedicaportuguesa.com/revista/index.php/amp/article/view/9997/5456), including five dimensions that have shown good values of Cronbach's alpha, ranging between 0.83 and 0.92, corresponding to a good internal consistency of this instrument (Fig. 2). The factors of each dimension have explained for a considerable cumulative variance ranging between 59.50 and 76.77%, therefore ensuring good construct validity.

The suggested rating for the analysis of the responses to the scale is based on the statistical analysis of the scores within each dimension. Therefore, the following calculation procedure has been defined: 1) a median value for each factor within each dimension has been obtained (as mean values were largely affected by the extreme scores); 2) the multiplication of the median of each factor by its percentage of explained variance in the principal component analysis (in order to consider the weight of each factor within each dimension); 3) the normalisation of the percentage to 100% (as a 100% of cumulative explained variance is never obtained in each dimension); 4) the sum of these normalised percentages in order to obtain the total percentage of each dimension. Final scores were rated as follows: scores <20% were considered as a poor assessment of practice quality regarding the aspects that were included in that dimension; scores 21-40% were considered as a suboptimal assessment; scores 41-60% as an optimal assessment, 61-80% as good and those >81% as a very good assessment of genetic counselling practice throughout the session.
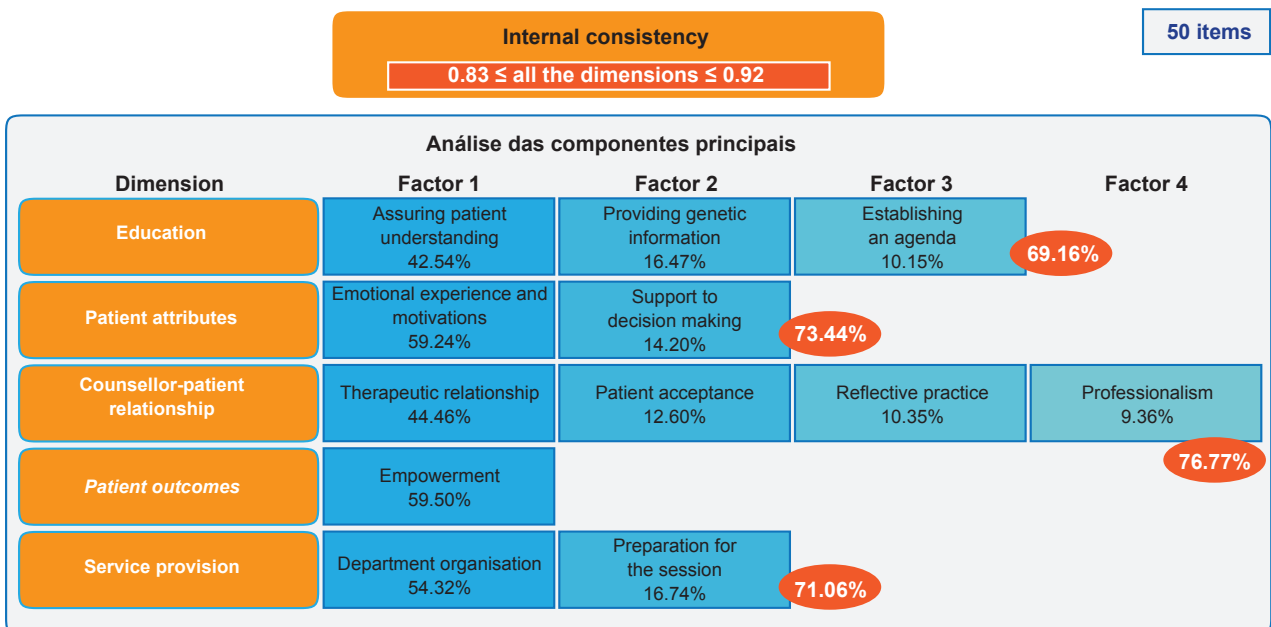

Figure 2 – Scale proposal - psychometric properties of the final version

Scores obtained within the process of psychometric validity of this national study could serve as a reference and are shown in Table 1. A baseline calculation formula has been developed in order to make rating easier for users.

DISCUSSION

This is a well-grounded scale due to its theoretical rationale as to the search for a conceptualisation of dimensions and sub-dimensions, keeping the underlying empirical, theoretical and practical aspects with the national research carried out by the authors.[11,12,21,25] The instrument that was developed in this research has shown appropriate psychometric properties, allowing for its immediate use within the clinical context and in research. Based on the results, the scale that was the result of this study is a suitable instrument to be used by healthcare professionals. The scale is different from other patient-centred instruments designed for the assessment of the quality of genetic counselling[18,19] focused on specific aspects of the process and/or its effects.[16-20]

It is worth mentioning that rating of the different dimensions of the scale that were obtained by the participants in this study could serve as reference values for future national studies regarding relevant components of healthcare including (i) department organisation, (ii) the presence of a reflective practice by professionals, (iii) quality of information support and assured patient's understanding on provided medical information, (iv) identification of patient's needs and recognition of the relevance of patient's individual characteristics to an adequate consultation process, (v) support to decision making and patient's empowerment, (vi) anticipation of patient outcomes and the presence of a strong therapeutic relationship, empathetic and respectful of the patient's autonomy.

Some limitations were identified. The small number of specialists currently active in Portugal is worth mentioning, which was a limitation to the statistical analysis of these data. This was aggravated by the fact that the three questionnaires were not completed by all the participating experts, therefore reducing the final number of eligible questionnaires. A total of 30 participants (52%) from the initial 58 have been obtained and, such as described, the positive feedback for the development of the scale reflected the perception regarding its usefulness for the departments and explaining for the receptiveness of the study by the major departments in Portugal.

The need for the completion of the scale upon three different sessions prevented from immediately collecting all the individual responses and this fact may have reduced the number of participants. In the future, studies of confirmatory factorial analysis suggesting for a new item organisation, as well as studies of metric invariance for the assessment of the scale structure stability, among others, could be included, in order to remedy the limitation related to the number of participants and improve the psychometric validity.

It is worth mentioning that the instrument had a satisfactory preliminary validity and that further empirical evidences will allow for better refinement of the current scale. Therefore, further studies will be required in order to improve the characteristics of the scale and the replication of this analysis applied to a larger sample of participants would be relevant in order to allow for the confirmation of the results and the reduction of possible disparities. This scale may also be applied to professionals from other countries in order to check for the validity and international applicability. In fact, interest has been already shown by professionals in genetic counselling from Norway, Spain and France.[36]

CONCLUSION

This proposed scale, unique in its kind, was aimed at leading genetic counselling professionals to a reflection regarding clinical practice and working as a practice guide. This is a multidimensional instrument bringing some important contributions to genetic counselling practice in Portugal. It seems quite useful in the reflective practice of professionals and in the assessment of genetic counselling practice.

Relevant indicators of counsellor-patient relationship

Table 1 – Reference scores of dimensions and scale factors

Median					
	Dimension 1	**Dimension 2**	**Dimension 3**	**Dimension 4**	**Dimension 5**
Factor 1	Items 1.5, 1.6, 1.9, 1.10 and 1.11 **3.00**	Items 2.1 to 2.7 **4.00**	Items 3.1, 3.2, 3.3, 3.7 and 3.8 **4.00**	Items 4.1 to 4.8 **5.00**	Items 5.1, 5.3, 5.5, 5.6 e 5.7 **4.50**
Factor 2	Items 1.2, 1.3, 1.4, 1.7, 1.8 and 1.12 **5.00**	Items 2.8 to 2.11 **5.00**	Items 3.3 to 3.5 **5.00**		Items 5.2, 5.3, 5.4 and 5.8 **4.50**
Factor 3	Items 1.1 and 1.12 **4.50**		Items 3.10 and 3.11 **3.00**		
Factor 4			Items 3.6 and 3.9 **4.50**		
Percentage of each dimension (%)					
	73.93	**83.87**	**81.80**	**100.00**	**90.00**

were assessed by the proposed scale, in order to respond to medical, educational and psycho-social issues involved in the process of genetic counselling and decision making in healthcare. Even though it is only focused on professionals, it allows for a check-list of whether the objectives that were established for each session were met and this is a measure of the usefulness of the instrument. Therefore, it may become relevant in the quality assessment, including the identification of the less developed aspects and allowing for the development of helpful intervention programmes.

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The authors declare that the followed procedures were according to regulations established by the Ethics and Clinical Research Committee of the ARS LVT (reference 9136/CES/2016) and according to the Helsinki Declaration of the World Medical Association.

DATA CONFIDENTIALITY

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

CONFLICTS OF INTEREST

The authors declare that there were no conflicts of interest in writing this manuscript.

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