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ARTICLE

Empowerment: qualitative underpinning of a new clinical genetics-specific patient-reported outcome

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Recent qualitative research developed a new construct labelled Empowerment describing a new patient outcome from using clinical genetics services that included four dimensions: Knowledge and Understanding, Decision-Making, Instrumentality and Future-Orientation. The aim of this study was to explore the validity, relevance and importance of the Empowerment construct for use as a patient-reported outcome (PRO) for clinical genetics services, and to refine the construct if necessary. Qualitative research (interviews and focus groups) was conducted in the UK with 12 patients, 15 representatives from patient support groups, 10 genetics clinicians and 4 service commissioners. Participants were asked to (1) describe what they think are the patient benefits from using clinical genetics services and (2) critique the Empowerment construct as a PRO. Interviews and focus groups were transcribed in full and analyzed using grounded theory. Findings confirmed the relevance and importance of the Empowerment construct, and identified Emotional Regulation as a further dimension of Empowerment. Data analysis also resulted in refinement of the construct, by renaming the other four dimensions to be Cognitive Control, Decisional Control, Behavioural Control and Hope. Empowerment has the potential to be a useful PRO to evaluate interventions in clinical genetics, and for use in clinical practice to generate data for continuous quality improvement. A study is underway to operationalise Empowerment by developing a psychometrically sound PRO measure that will take the form of a short questionnaire.
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INTRODUCTION

Evaluating interventions in clinical genetics is difficult because there is lack of clarity about which are the best outcome domains to measure,[1–3] although efforts are underway to define and achieve consensus on outcomes.[3,4] Traditional measures of health status, eg, EQ-5D or SF36 may not be appropriate because many genetic conditions can be neither treated nor cured.[5] Often, the interventions offered in clinical genetics relate to provision of information about a diagnosis, or a genetic test result, or empiric risk information. One approach has been to measure patient knowledge, or information recall, but this approach has been criticized because of the substantial assumptions that specific items of knowledge are either valued by patients, or contribute to effective decision-making. In effect, these studies are tests of memory and understanding, not evaluations of patient benefit. Another approach has been to use generic measures of psychological constructs, eg, anxiety, but these approaches have not been shown to discriminate effectively between different models of service delivery in clinical trials.[6] A third, more recent approach is to measure effectiveness of decisions using measures of informed decision-making or decisional conflict. These approaches are limited to evaluating effectiveness of a single decision, and may not be relevant for evaluating clinical genetics services, where outcomes relate to the capacity to make many decisions.[7] Measures of satisfaction have also been used to evaluate clinical genetics.[8–10] Although an important component in assessing healthcare quality, patient satisfaction is not sufficient to capture all the important patient benefits and, furthermore, is influenced by expectations.[11]

Our recent systematic review of validated outcome measures used in clinical genetics identified that 67 different outcome measures have been used to evaluate clinical genetics. A total of 46 of those measures were used in just one study each,[8] highlighting the confusion about appropriate measures. Outcome domains captured by the measures ranged from health status through knowledge to quality of life. The review concluded that for clinical genetics services, (a) there was no single validated outcome measure available to capture all potential patient benefits (b) some important aspects of patient benefit are not captured by available measures and (c) there is scope for new measures.

Our previous qualitative research[6,7,12,13] suggests that patient benefits from clinical genetics services could be summarised using a new construct labelled Empowerment.[7] Empowerment was defined as a set of beliefs that enable a person from a family affected by a genetic condition to feel that they have some control over and hope for the future. Empowerment included four dimensions: the beliefs that one (1) can make important life decisions in an informed way (Decision-Making), (2) has sufficient information about the condition, including risks to oneself and one's relatives, and any treatment, prevention and support available (Knowledge And Understanding), (3) can make effective use of the health and social care systems for the benefit of the whole family (Instrumentality) and (4) can look to the future with

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hope for a fulfilling family life, for oneself, one's family and/or one's future descendents (Future Orientation).[7]

When compared with outcome measures used in evaluations of clinical genetics, Empowerment was found to be conceptually most similar to the construct captured by the Perceived Personal Control (PPC) measure,[14] which includes three dimensions: Cognitive Control, Decisional Control and Behavioural Control. However, the PPC questionnaire does not capture all the important patient benefits summarised in the Empowerment construct, eg, benefits to other relatives and to future generations.

There is scope to develop a new patient reported outcome measure (PROM) to capture the Empowerment construct. PROMs are short validated questionnaires completed by patients to evaluate health interventions. A PROM of Empowerment could be useful in evaluating new interventions in clinical genetics, thus contributing to establishing a robust evidence base for practice. However, development of PROMs is difficult and time consuming,[15–17] and justifiable only if there is evidence that the new measure will capture a PRO that is novel, useful, relevant and important. The aims in the present study were to (1) explore the validity, relevance and importance of the Empowerment construct and refine the construct as necessary and (2) inform the decision whether or not to develop a clinical genetics-specific PROM of Empowerment.

METHODS

To meet the research objectives, further qualitative data were collected. Qualitative methods are suitable for exploratory research in areas that are not well understood, and where the relevant variables have not been fully identified.[18] As one purpose of this research was to explore whether the Empowerment construct includes all relevant facets of patient benefit from using clinical genetics services, a qualitative approach was appropriate. Patients were recruited through Genetic Medicine, St Mary's Hospital, Manchester, UK. The term 'patient' is used here to refer to all users of a clinical genetics service, including unaffected parents and unaffected at-risk families members. Patients were asked to participate in a focus group, but also had the option to be interviewed. Genetics clinicians were recruited through professional networks, and were only asked to participate in a focus group. Previous research indicated that focus groups work well with genetics clinicians, whereas a combined focus group/interview approach works best with patients of clinical genetics services.[4,5,11,12] Commissioners (who purchase clinical genetics services on behalf of primary care) and patient representatives (members of patient support groups) were recruited through UK professional networks to participate in an interview only because of geographical spread. Focus groups were conducted in Nowgen, Manchester, UK. Interviews were conducted in person or by telephone, according to participant preference. Ethical approval was obtained from the NHS National Research Ethics Service.

Participants were asked to describe their views about the benefits and harms of using clinical genetics services, and then given a page describing the Empowerment construct, with its four dimensions, and the dimension definitions.[7] Participants were asked to read and critique this, in particular to identify any benefits/harms of using a clinical genetics service that were missing. Focus groups and interviews were transcribed in full, and analyzed using tools of grounded theory methodology[19,20] including: (1) open coding: labels (codes) significant pieces of text as important (2) constant comparison: each new instance of a code is compared with every other instance of that code, and the concept definition revised accordingly and (3) questioning the data in relation to the research question. Glaser and Strauss, the developers of grounded theory methodology, recommend that any emerging grounded theory (model) should be 'checked out' for usefulness with 'experts' familiar with the substantive area under investigation.[19,21] In this study, the emerging construct (model) of Empowerment is checked out with patients, patient representatives, clinicians and commissioners of clinical genetics services to clarify whether the construct provides a plausible and useful summary of patient benefit. Feedback of this kind can indicate whether or not the construct (model) has 'grab',[21] ie, whether it provides a novel and useful framework.

RESULTS

A total of 138 people were approached and asked to participate. In all, 54 (39.2%) agreed to participate and 18 (13%) declined. Of the 54 who agreed to participate, 13 (9.4%) were excluded because they could not be accommodated in a focus group because of scheduling difficulties, and did not wish to be interviewed instead. The final sample was composed of 41 participants (29.8%) (Tables 1 and 2). Owing to ethical constraints, those approached were not asked to give reasons for their decision, so reasons for the low response rate amongst patients are not known. Three focus groups were conducted, two with genetics clinicians (n=5 in both) and one with patients (n=4). A total of 27 interviews were conducted, 4 with service commissioners, 8 with patients and 15 with patient representatives.

Participants' views about the 'Empowerment' construct

The qualitative study generated data confirming the importance of the Empowerment construct, and the previously identified four dimensions of Empowerment (Knowledge and Understanding, Decision-Making, Instrumentality and Future-Orientation). All 41 participants, except for one patient representative, endorsed use of the word 'Empowerment' as a good overarching term to summarise patient benefits, and clarified further what Empowerment means in the context of clinical genetics:

> 'I think that's the best word you could think of, yes it is, having empowerment -having that knowledge is the biggest benefit that you can gain. And yes I would recommend a genetic service to anybody, well I do when they have a new baby and they're all sort of all bewildered […] 'empowerment,' yes that is […] a very good word to describe it' (patient, Congenital adrenal hyperplasia).

Results suggest that genetic diagnosis may engender a state of disempowerment, which throws families into a new state, perhaps best described as social alienation, which can be addressed in genetic counseling, as illustrated by this participant:

> 'I just feel though that in a way the system of the diagnosis is what has disempowered parents in the first place, so it [is] actually about re-establishing confidence rather than just giving it [to] them for the first time' (patient representative, D/deaf charity).

A good outcome from using a clinical genetics service could then be seen to involve learning about their new situation, and regaining some confidence and control over their lives, so that they 'know what they are doing', which is experienced as empowering, expressed as follows:

> '…the information empowers you to make decisions, and provided us with the opportunity to have a family, it gave us the confidence to try for another baby, which we would not have done otherwise' (patient, previously had a late termination at 23 weeks gestation of a pregnancy affected by Edwards syndrome).

The views expressed by patients, and patient representatives, were endorsed by the service commissioner participants who confirmed that they want to commission clinical genetics services that can offer choice to patients:

> '… the patients […] have the choice to consider their own lifestyles, their own social decisions, the future of their own children and that chance of things if they wish to […] carry on with the pregnancy if they've already conceived, or to terminate or to not conceive in future, to adopt, to go through PGD

Table 1 Participation rates and sample composition

	Patients (choice of focus group or interview), n *(%)*	*Patient representatives (interviews only),* n *(%)*	*Genetics clinicians (focus groups only),* n *(%)*	*Commissioners (interviews only),* n *(%)*	*Total (interviews and focus groups),* n *(%)*
Approached	79 (100)	20 (100)	32 (100)	7 (100)	138 (100)
No response	47 (59.5)	5 (25)	12 (37.5)	2 (28.6)	66 (47.8)
Declined	17 (21.5)	0 (0)	0 (0)	1 (14.3)	18 (13)
Agreed to participate	15 (19)	15 (75)	20 (62.5)	4 (57.1)	54 (39.2)
Subsequently unable to participate[a]	3 (3.8)	0	10 (31.25)	0	13 (9.4)
Participated	12 (15.2)	15 (75)	10 (31.25)	4 (57.1)	41 (29.8)
Genetic conditions represented	• Hereditary cardiomyopathy • Ehlers Danlos syndrome • Congenital adrenal hyperplasia • BRCA2 • Zellweger syndrome • Chromosome abnormalities • Retinitis pigmentosa • Marfan syndrome • Neurofibromatosis type 1 • Fragile X syndrome • Undiagnosed rare conditions	• Chromosome abnormalities • Hereditary deafness • Sickle cell disease • Alpha-1 antitrypsin deficiency • Cystic fibrosis • Laurence Moon Bardet Biedl syndrome • Muscular dystrophy • Porphyria • Pregnancy termination for fetal abnormalities • Charcot Marie tooth disease • Battens disease • Ectodermal dysplasia			

[a]This group comprises individuals who wanted to participate but could not be accommodated because of the logistical difficulties associated with trying to find a suitable time to meet.

Table 2 Sample characteristics: patients and patient representatives

Genetic status	*Patients (choice of focus group or interview)* n=*12*	*Patient representatives (interviews only)* n=*15*	*Total (interviews and focus groups)* n=*27*
Patient advocate[a]	0	3	3
Diagnosed with a genetic condition	4	7	11
At risk for developing a genetic condition	1	0	1
Parent/grandparent of an affected child	7	5	12

[a]Not from a family affected by a genetic condition.

(preimplantation genetic diagnosis) whatever, their decision, it is the parents decision' (service commissioner).

One participant, although she validated the importance of the four dimensions of Empowerment, was unsure about the appropriateness of the word 'Empowerment':

'I have slight reservations about the word but certainly in terms of those – what you've laid out there as the … what underpins that I think all are extremely valid, you know, I think that would […] definitely [be] a useful way of measuring (outcomes) […] 'empowerment' suggests that you are able to be proactive and that I don't think is always going to be the way or would be a potential outcome of a clinical genetic service, I don't think that's feasible. (patient representative, charity supporting couples who have terminated pregnancies for fetal abnormalities).

Because all 41 participants, except for this one, endorsed use of the word 'Empowerment' to summarise patient benefits from using clinical genetics services, and some did so strongly, it was decided to retain the term 'Empowerment' as an overarching term to describe the model.

Refinement of the Empowerment construct

All participants endorsed the relevance and importance of the four dimensions of Empowerment when shown the definitions, but the analysis resulted in renaming three of these dimensions to further clarify the Empowerment construct. Data analysis confirmed that Knowledge and Understanding, as conceptualised in Empowerment, is more akin to 'Cognitive Control', a dimension of PPC, than to information recall. Knowledge about the condition is valued for its own sake because it provides an explanation for previously unexplained, unexpected and distressing events (recurrence of disease in the family, loss of a child, disease symptoms), and this in itself is a benefit. Simply having a diagnosis provides reassurance and enables families to prepare for the future:

'[…] The benefits to me of having the diagnosis confirmed that what I was experiencing that it actually had a name, and despite the fact that there was very little research done in to the condition twelve years ago it enabled us to do some research ourselves and to keep up with the latest what was going on. So we could put a focus on it, you know…' (patient representative, affected by Porphyria).

It appeared that labeling symptoms with a scientific name had the quality of a 'latin incantation', which reduced uncertainty/anxiety and validated the illness experience.

Similarly, the data confirmed that Decision-Making, as conceptualised in Empowerment, is more like 'Decisional Control', a dimension of PPC, than to effectiveness of any single decision. Finally, Instrumentality, as conceptualised in Empowerment, is more similar to 'Behavioural Control', a dimension of PPC, than to any specific action taken. Understanding the implications of the genetic condition to oneself, one's relatives and future generations, and knowing about available medical interventions and support enables decisions to be made and actions to be taken:

> '…it's the information that enables you to make the decisions, whether to go ahead and have another baby, or whether to have more children, and what sort of tests to have' (patient, baby affected by chromosome abnormality).

For these reasons, the PPC terminology of Cognitive, Decisional and Behavioural Control was adopted for the Empowerment dimensions previously known as Knowledge and Understanding, Decision-making and Instrumentality, respectively.

Future-Orientation was also endorsed by all participants in this study as relevant and important to good patient outcomes. Understanding the genetic condition and knowing about services and interventions available to help manage the effects of the condition enables families to orient themselves towards the future with more confidence:

> '… it comes back to this control isn't it that you are able to map out your future life you know, it's not that much of an unknown […] (and) feeling comfortable about one's children's future' (patient representative, child affected by an unbalanced chromosome translocation).

One participant suggested that the Future-Orientation dimension be renamed 'Hope' because she felt that providing Hope was an important part of any diagnosis of a life-limiting condition. The word Hope came up a few times in interviews as an important benefit from using clinical genetics, and the decision was made to change the name of this dimension from Future-Orientation to Hope, partly because the term Future-Orientation required some explanation to participants, whereas the word Hope is more widely understood.

Further development of the Empowerment construct

The qualitative analysis resulted in the identification of one further dimension of Empowerment: Emotional Regulation. Genetics clinicians in one focus group and a number of patient representatives, some of whom were affected by genetic conditions and some of whom were parents of affected people, commented in interviews that the model omitted any mention of emotions, which they felt were important. No other omissions were identified. Patient representatives, all of whom had a genetic condition in their own family and who were also able to provide an overview of the variability in experiences of their patient group members, described how a genetic diagnosis throws the family into emotional turmoil and highlighted the need for families to be given emotional support:

> '… for families to have emotional support around a point of diagnosis […] I would say that everybody is depressed around the time of using the service' (patient representative, charity supporting families affected by Battens disease).

They felt strongly that clinical genetics services should be instrumental in addressing these difficult emotions and in helping families to reach a new equilibrium:

> '… when you're first given a diagnosis it's like your whole life is spinning out of control you don't know where you're going, you don't know what's going to happen to you or your family, what the future holds, it's incredibly frightening having been through it myself […] a very important outcome (is) that the emotions are dealt with' (patient group representative, child has unbalanced chromosome translocation).

Genetic counsellors commented that a large part of their work focuses on offering emotional support to patients, and so they appeared to recognise the importance to families of addressing and dealing with difficult emotions in genetic counselling.

No other new issues were identified in the final five interviews and so theoretical saturation was assumed and recruitment was stopped.

The new Empowerment construct

In summary, the qualitative analysis endorsed the Empowerment construct and refined the construct by (a) adopting the PPC terminology of Cognitive, Decisional and Behavioural Control for the dimensions previously labeled Knowledge and Understanding, Decision-Making and Instrumentality, respectively, (2) adopting the term 'Hope' for the dimension previously labeled 'Future-Orientation' and (3) extending the model to include a fifth dimension: Emotional Regulation.

DISCUSSION

This research has validated and further developed and refined the Empowerment construct, to summarise the patient benefits from using clinical genetics services.[7] A sample of patients and patient representatives, clinicians and commissioners of clinical genetics services in the UK endorsed use of the term 'Empowerment' to summarise the patient benefits, and identified one further dimension of Empowerment, Emotional Regulation, that was missing from the four-dimensional model. Furthermore, the data have enabled the meaning of the previously identified four dimensions of Knowledge and Understanding, Decision-Making, Instrumentality and Future-Orientation to be refined. The revised construct is composed of five dimensions: Cognitive Control, Decisional Control, Behavioural Control, Hope and Emotional Regulation, and has clarified the relationship between Empowerment and the concept of PPC.[7,12,20] Findings support the importance of PPC as a PRO, and identified that Empowerment includes the three PPC dimensions (Cognitive, Decisional and Behavioural Controls), and two further dimensions: Hope and Emotional Regulation.

In contrast to other research, eg, exploring autism in France,[22] neither this nor our previous research[6,7,12,13] identified any differences between the views of healthy parents of a child affected by a condition, patients affected by a condition and those at risk for developing a condition, or between adults at different stages of development. There is scope to explore this further in future research.

The Empowerment PRO, if operationalised as a PROM, could be used for evaluation of whether some of the criteria in the recently developed EuroGentest 'Instrument for quality self-assessment in provision of genetic counselling' (http://www.eurogentest.org/professionals/assessment_tool/index.xhtml) have been met. For example, the EuroGentest instrument includes the criteria (1) 'Counsellees are provided with Decision-Making support', which

could provide counsellees with improved Decisional Control and (2) 'Counsellees understand what they are being told' which could provide counsellees with improved Cognitive Control. A PROM of Empowerment would enable these criteria to be measured by patient self-report, rather than by service self-assessment.

The constituent dimensions of Empowerment identified in the present study also overlap with some of the outcomes identified in a consensus study recently reported by the US Western States Genetics Services Collaborative (WSGSC),[4] and the present study provides further support for the relevance and importance of these initiatives.

For example, the WSGSC outcome 'Individuals and families feel supported in managing grief, stress, and emotional challenges of living with a genetic condition' could be interpreted as Emotional Regulation. The WSGSC outcome 'Individuals and families are able to make informed health and life decisions based upon diagnosis' could be interpreted as Decisional Control. However, the present study goes further than identifying patient outcomes by integrating five different patient outcomes in the overarching patient-centred construct of Empowerment. The present study provides sufficient evidence to suggest that Empowerment is a novel PRO for clinical genetics services.

Identification of Emotional Regulation as an additional dimension of Empowerment, and as an important outcome from using clinical genetics services, is not surprising. Good patient outcomes from using healthcare services could be conceptualised as alleviation of those disease effects experienced as problematic by patients and their families. From this perspective, the finding that Emotional Regulation is an important outcome in clinical genetics is consistent with research evidence that the emotional effects of genetic diseases are problematic for families.[12] These emotional effects may include feelings of powerlessness and a threatened sense of security and identity, previously described in relation to diagnosis of long-term chronic conditions.[23] This finding also supports models of genetic counselling practice, which emphasise a psycho-educational approach,[24] rather than a purely educational model.[25] The educational model emphasises 'educating' the patient and is based on the assumption that patients come to clinical genetics services for information only. In contrast, the psycho-educational approach assumes that patients are seeking a therapeutic relationship in which they are supported in using genetic information in a personally meaningful way that minimizes psychological distress and increases personal control.

Hope has not previously been identified as a valued outcome from clinical genetics services, and its inclusion in the model is novel. On a theoretical level, Emotional Regulation is likely to interact with Hope, and with PPC and it's three dimensions of Cognitive, Decisional and Behavioural Control. A genetic diagnosis, and a scientific explanation for the previously unexplained death of her child, information about risks to future pregnancies, and options for prenatal diagnosis could help a bereaved mother to (1) understand why her child died (Cognitive Control), (2) make decisions about whether to have further children (Decisional Control), (3) use prenatal testing in future pregnancies to avoid recurrence (Behavioural Control), (4) feel that it is possible to have healthy children and a rewarding family life in the future (Hope) and (5) alleviate feelings of guilt that she had done something in pregnancy to cause her child's condition (Emotional Regulation). Clearly, it may not be possible to bring about positive change in all five dimensions for every patient who attends a clinical genetics service. However, it is plausible to hypothesise that the five dimensions of Empowerment are facets of a single overarching construct.

The main weakness of this research is that it is limited to the perspectives of families living in the UK, who speak English. Empowerment is likely to be influenced by culture,[7] and so Empowerment would benefit from validation in other cultures and languages. A further weakness is that patient participation rates were low. However, because this work builds on extensive previous qualitative research,[4,5,11,12] and because care was taken to reach data saturation, it is unlikely that significant issues were missed.

The main strength of this research is that the new PRO identified – Empowerment – has been developed through consultation with (1) people struggling with the challenges of having a genetic condition in their family and (2) clinicians with experience of helping these families. They are the 'experts' consulted in this research to identify the patient benefits from using clinical genetics services. The traditional approach to the development of PROMs is to use theoretical frameworks to identify PROs, and this approach was used in identifying PPC as a patient outcome from genetic counselling.[14,26] This approach is useful, but as the present study shows, consulting patients and clinicians can enable new constructs to be developed, and existing constructs to be extended, to ensure that PROs include all benefits valued by patients.

A measure of PPC was developed and evaluated in Hebrew.[14] When the paper reporting development of the PPC questionnaire was published in a US journal, the PPC was translated into English.[14] Although the English translation of the PPC has been used with English speaking samples,[27] it has not yet been validated for use with a UK population. A study is underway to operationalise Empowerment by developing a new genetics-specific PROM of Empowerment, using the PPC measure and the qualitative data collected in this and previous studies.[4,5,11,12] Both PROMs and patient empowerment have been identified as key aspects of current UK health policy.[28] In addition, there was a recent call for better measures to capture aspects of health-related quality of life (HRQoL) in clinical trials in medical genetics.[29] These developments strengthen the argument in support of investing the time and resources needed to develop a new genetics-specific PROM to capture Empowerment. This PROM would then be available to capture HRQoL outcomes in clinical trials and in clinical practice to generate data to support continuous quality improvement.[30] As part of PROM development,[16] factor analysis will investigate the dimensional structure of Empowerment to confirm whether or not Empowerment is a single multidimensional construct.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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