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Empowerment: Qualitative Underpinning of a New Clinical Genetics-Specific Patient Reported Outcome

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1 Title: Empowerment: Qualitative Underpinning of a New Clinical Genetics-Specific
2 Patient Reported Outcome.

3

4 Short running title: Empowerment: A New Patient Reported Outcome

5

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19

1 **ABSTRACT**

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

22

23 **KEYWORDS:** Patient Reported Outcomes, empowerment, qualitative research,
24 emotional regulation, grounded theory

25

1 **INTRODUCTION**

2 Evaluating interventions in clinical genetics is difficult because there is lack of clarity
3 about which are the best outcome domains to measure¹⁻³, although efforts are underway
4 to define and achieve consensus on outcomes^{3,4}. Traditional measures of health status
5 e.g. EQ-5D or SF36 may not be appropriate because many genetic conditions can be
6 neither treated nor cured⁵. Often, the interventions offered in clinical genetics relate to
7 provision of information about a diagnosis, or a genetic test result, or empiric risk
8 information. One approach has been to measure patient knowledge, or information
9 recall, but this approach has been criticized because of the substantial assumptions that
10 specific items of knowledge are either valued by patients, or contribute to effective
11 decision-making. In effect, these studies are tests of memory and understanding, not
12 evaluations of patient benefit. Another approach has been to use generic measures of
13 psychological constructs, e.g. anxiety, but these approaches have not been shown to
14 discriminate effectively between different models of service delivery in clinical trials⁶.
15 A third, more recent approach is to measure effectiveness of decisions using measures
16 of informed decision-making or decisional conflict. These approaches are limited to
17 evaluating effectiveness of a single decision, and may not be relevant for evaluating
18 clinical genetics services, where outcomes relate to the capacity to make many
19 decisions⁷. Measures of satisfaction have also been used to evaluate clinical genetics⁸⁻¹⁰.
20 Although an important component in assessing healthcare quality, patient satisfaction is
21 not sufficient to capture all the important patient benefits, and furthermore, is influenced
22 by expectations¹¹.
23 Our recent systematic review of validated outcome measures used in clinical genetics
24 identified that 67 different outcome measures have been used to evaluate clinical

1 genetics. 46 of those measures were used in just one study each⁸, highlighting the
2 confusion about appropriate measures. Outcome domains captured by the measures
3 ranged from health status through knowledge to quality of life. The review concluded
4 that for clinical genetics services (a) there was no single validated outcome measure
5 available to capture all potential patient benefits (b) some important aspects of patient
6 benefit are not captured by available measures and (c) there is scope for new measures.
7 Our previous qualitative research^{6,7,12,13} suggests that patient benefits from clinical
8 genetics services could be summarised using a new construct labelled Empowerment⁷.
9 Empowerment was defined as a set of beliefs that enable a person from a family
10 affected by a genetic condition, to feel that they have some control over and hope for
11 the future. Empowerment included four dimensions: the beliefs that one (1) can make
12 important life decisions in an informed way (Decision-Making), (2) has sufficient
13 information about the condition, including risks to oneself and one's relatives, and any
14 treatment, prevention and support available (Knowledge and Understanding), (3) can
15 make effective use of the health and social care systems for the benefit of the whole
16 family (Instrumentality) and (4) can look to the future with hope for a fulfilling family
17 life, for oneself, one's family, and/or one's future descendants (Future Orientation)⁷.
18 When compared with outcome measures used in evaluations of clinical genetics,
19 Empowerment was found to be conceptually most similar to the construct captured by
20 the Perceived Personal Control (PPC) measure¹⁴, which includes three dimensions:
21 Cognitive Control, Decisional Control and Behavioural Control. However, the PPC
22 questionnaire does not capture all the important patient benefits summarised in the
23 Empowerment construct e.g. benefits to other relatives and to future generations.

1 There is scope to develop a new Patient Reported Outcome Measure (PROM) to capture
2 the Empowerment construct. PROMs are short validated questionnaires completed by
3 patients to evaluate health interventions. A PROM of Empowerment could be useful in
4 evaluating new interventions in clinical genetics, thus contributing to establishing a
5 robust evidence base for practice. However, PROMs development is difficult and time-
6 consuming¹⁵⁻¹⁷, and justifiable only if there is evidence that the new measure will
7 capture a Patient Reported Outcome (PRO) that is novel, useful, relevant and important.
8 The aims in the present study were to (1) explore the validity, relevance and importance
9 of the Empowerment construct and refine the construct as necessary and (2) inform the
10 decision whether or not to develop a clinical genetics-specific PROM of Empowerment.

11 **METHODS**

12 To meet the research objectives, further qualitative data were collected. Qualitative
13 methods are suitable for exploratory research in areas that are not well understood, and
14 where the relevant variables have not been fully identified¹⁸. Since one purpose of this
15 research was to explore whether the Empowerment model includes all relevant facets of
16 patient benefit from using clinical genetics services, a qualitative approach was
17 appropriate. Patients were recruited through Genetic Medicine, St Mary's Hospital,
18 Manchester, UK. The term 'patient' is used here to refer to all users of a clinical
19 genetics service, including unaffected parents and unaffected at risk families members.
20 Patients were asked to participate in a focus group, but also had the option to be
21 interviewed. Genetics clinicians were recruited through professional networks, and were
22 only asked to participate in a focus group. Previous research indicated that focus groups
23 work well with genetics clinicians, whereas a combined focus group / interview
24 approach works best with patients of clinical genetics services^{4 5 11 12}. Commissioners

1 (who purchase clinical genetics services on behalf of primary care) and patient
2 representatives (members of patient support groups) were recruited through UK
3 professional networks, to participate in an interview only because of geographical
4 spread. Focus groups were conducted in Nowgen, Manchester. Interviews were
5 conducted in person or by telephone, according to participant preference. Ethical
6 approval was obtained from the NHS National Research Ethics Service.

7 Participants were asked to describe their views about the benefits and harms of using
8 clinical genetics services, and then given a page describing the Empowerment construct,
9 with its four dimensions, and the dimension definitions⁷. Participants were asked to read
10 and critique this, in particular to identify any benefits / harms of using a clinical
11 genetics service that were missing. Focus groups and interviews were transcribed in
12 full, and analysed using tools of grounded theory methodology^{19,20} including: (1) open
13 coding: labels (codes) significant pieces of text as important (2) constant comparison:
14 each new instance of a code is compared with every other instance of that code, and the
15 concept definition revised accordingly and (3) questioning the data in relation to the
16 research question. Glaser and Strauss, the developers of grounded theory methodology,
17 recommend that any emerging grounded theory (model) should be “checked out” for
18 usefulness with “experts” familiar with the substantive area under investigation^{19,21}. In
19 this study, the emerging construct (model) of Empowerment is checked out with
20 patients, patient representatives, clinicians, and commissioners of clinical genetics
21 services to clarify whether the construct provides a plausible and useful summary of
22 patient benefit. Feedback of this kind can indicate whether or not the construct (model)
23 has “grab”²¹ i.e. whether it provides a novel and useful framework.

24 **RESULTS**

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

10 Participants' views about the 'Empowerment' construct

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

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

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

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

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

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

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

15 “... the patients [...] have the choice to consider their own lifestyles, their own social
16 decisions, the future of their own children and that chance of things if they wish to
17 [...] carry on with the pregnancy if they've already conceived, or to terminate or to not
18 conceive in future, to adopt, to go through PGD (preimplantation genetic diagnosis)
19 whatever, their decision, it is the parents decision.” (service commissioner)

20 One participant, although she validated the importance of the four dimensions of
21 Empowerment, was unsure about the appropriateness of the word “Empowerment”:

22 “I have slight reservations about the word but certainly in terms of those - what you've
23 laid out there as the ... what underpins that I think all are extremely valid, you know, I
24 think that would [...] definitely [be] a useful way of measuring (outcomes) [...]

1 'empowerment' suggests that you are able to be proactive and that I don't think is
2 always going to be the way or would be a potential outcome of a clinical genetic
3 service, I don't think that's feasible. (patient representative, charity supporting couples
4 who have terminated pregnancies for fetal abnormalities)

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

9 Refinement of the Empowerment construct

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

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

1 It appeared that labeling symptoms with a scientific name had the quality of a “latin
2 incantation” which reduced uncertainty/anxiety, and validated the illness experience.
3 Similarly, the data confirmed that Decision-Making, as conceptualised in
4 Empowerment, is more like “Decisional Control”, a dimension of PPC, than to
5 effectiveness of any single decision. Finally, Instrumentality, as conceptualised in
6 Empowerment, is more similar to “Behavioural Control”, a dimension of PPC, than to
7 any specific action taken. Understanding the implications of the genetic condition to
8 oneself, one’s relatives, and future generations, and knowing about available medical
9 interventions and support enables decisions to be made, and actions to be taken:
10 “...it's the information that enables you to make the decisions, whether to go ahead
11 and have another baby, or whether to have more children, and what sort of tests to
12 have.” (patient, baby affected by chromosome abnormality)

13 For these reasons, the PPC terminology of Cognitive, Decisional and Behavioural
14 Control was adopted for the Empowerment dimensions previously known as
15 Knowledge and Understanding, Decision-making and Instrumentality respectively.
16 Future-Orientation was also endorsed by all participants in this study as relevant and
17 important to good patient outcomes. Understanding the genetic condition, and knowing
18 about services and interventions available to help manage the effects of the condition
19 enables families to orient themselves towards the future with more confidence:
20 “... it comes back to this control isn't it that you are able to map out your future life
21 you know, it's not that much of an unknown [...] (and) feeling comfortable about
22 one's children's future” (patient representative, child affected by an unbalanced
23 chromosome translocation)

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

8 Further development of the Empowerment construct

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

19 “... for families to have emotional support around a point of diagnosis [...] I would
20 say that everybody is depressed around the time of using the service”. (patient
21 representative, charity supporting families affected by Batters disease)

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

1 “... when you're first given a diagnosis it's like your whole life is spinning out of
2 control you don't know where you're going, you don't know what's going to happen to
3 you or your family, what the future holds, it's incredibly frightening having been
4 through it myself [...] a very important outcome (is) that the emotions are dealt with.”
5 (patient group representative, child has unbalanced chromosome translocation)
6 Genetic counsellors commented that a large part of their work focuses on offering
7 emotional support to patients, and so they appeared to recognise the importance to
8 families of addressing and dealing with difficult emotions in genetic counselling.
9 No other new issues were identified in the final 5 interviews, and so theoretical
10 saturation was assumed, and recruitment stopped.

11 The new Empowerment construct

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

18 **DISCUSSION**

19 This research has validated and further developed and refined the Empowerment
20 construct, to summarise the patient benefits from using clinical genetics services⁷. A
21 sample of patients and patient representatives, clinicians and commissioners of clinical
22 genetics services in the UK endorsed use of the term “Empowerment” to summarise the
23 patient benefits, and identified one further dimension of Empowerment, Emotional
24 Regulation, that was missing from the four-dimensional model. Furthermore, the data

1 have enabled the meaning of the previously identified four dimensions of Knowledge
2 and Understanding, Decision-Making, Instrumentality and Future-Orientation to be
3 refined. The revised construct is composed of 5 dimensions: Cognitive Control,
4 Decisional Control, Behavioural Control, Hope and Emotional Regulation, and has
5 clarified the relationship between Empowerment, and the concept of Perceived Personal
6 Control (PPC) ^{7,12,20}. Findings support the importance of PPC as a PRO, and identified
7 that Empowerment includes the three PPC dimensions (Cognitive, Decisional and
8 Behavioural Control), and two further dimensions: Hope and Emotional Regulation.
9 In contrast to other research, for example exploring autism in France²², neither this nor
10 our previous research^{6,7,12,13} identified any differences between the views of healthy
11 parents of a child affected by a condition, patients affected by a condition, and those at
12 risk for developing a condition, or between adults at different stages of development.
13 There is scope to explore this further in future research.
14 The Empowerment PRO, if operationalised as a PROM, could be used for evaluation of
15 whether some of the criteria in the recently developed EuroGentest “Instrument for
16 quality self-assessment in provision of genetic counselling”
17 (http://www.eurogentest.org/professionals/assessment_tool/assessment_tool/form.xhtm)
18 have been met. For example, the EuroGentest instrument includes the criteria (1)
19 “Counselees are provided with decision-making support”, which could provide
20 counselees with improved decisional control and (2) “Counselees understand what
21 they are being told” which could provide counselees with improved cognitive control.
22 A PROM of Empowerment would enable these criteria to be measured by patient self-
23 report, rather than by service self-assessment.

1 The constituent dimensions of Empowerment identified in the present study also
2 overlap with some of the outcomes identified in a consensus study recently reported by
3 the US Western States Genetics Services Collaborative (WSGSC)⁴, and the present
4 study provides further support for the relevance and importance of these initiatives.
5 For example, the WSGSC outcome “Individuals and families feel supported in
6 managing grief, stress, and emotional challenges of living with a genetic condition”
7 could be interpreted as Emotional Regulation. The WSGSC outcome “Individuals and
8 families are able to make informed health and life decisions based upon diagnosis”
9 could be interpreted as Decisional Control. However, the present study goes further than
10 identifying patient outcomes by integrating 5 different patient outcomes in the
11 overarching patient-centred construct of Empowerment. The present study provides
12 sufficient evidence to suggest that Empowerment is a novel PRO for clinical genetics
13 services.

14 Identification of Emotional Regulation as an additional dimension of Empowerment,
15 and as an important outcome from using clinical genetics services, is not surprising.
16 Good patient outcomes from using healthcare services could be conceptualised as
17 alleviation of those disease effects experienced as problematic by patients and their
18 families. From this perspective, the finding that Emotional Regulation is an important
19 outcome in clinical genetics is consistent with research evidence that the emotional
20 effects of genetic diseases are problematic for families¹². These emotional effects may
21 include feelings of powerlessness and a threatened sense of security and identity,
22 previously described in relation to diagnosis of long-term chronic conditions²³. This
23 finding also supports models of genetic counselling practice, which emphasise a
24 psycho-educational approach²⁴, rather than a purely educational model²⁵. The

1 educational model emphasises ‘educating’ the patient, and is based on the assumption
2 that patients come to clinical genetics services for information only. In contrast, the
3 psycho-educational approach assumes that patients are seeking a therapeutic
4 relationship in which they are supported in using genetic information in a personally
5 meaningful way that minimizes psychological distress and increases personal control.
6 Hope has not previously been identified as a valued outcome from clinical genetics
7 services, and its inclusion in the model is novel. On a theoretical level, Emotional
8 Regulation is likely to interact with Hope, and with Perceived Personal Control, and it’s
9 three dimensions of Cognitive, Decisional and Behavioural Control. A genetic
10 diagnosis, and a scientific explanation for the previously unexplained death of her child,
11 information about risks to future pregnancies, and options for prenatal diagnosis could
12 help a bereaved mother to (1) understand why her child died (Cognitive Control) (2)
13 make decisions about whether to have further children (Decisional Control) (3) use
14 prenatal testing in future pregnancies to avoid recurrence (Behavioural Control) (4) feel
15 that it is possible to have healthy children and a rewarding family life in the future
16 (Hope) and (5) alleviate feelings of guilt that she had done something in pregnancy to
17 cause her child’s condition (Emotional Regulation). Clearly, it may not be possible to
18 bring about positive change in all 5 dimensions for every patient who attends a clinical
19 genetics service. However it is plausible to hypothesise that the 5 dimensions of
20 Empowerment are facets of a single overarching construct.

21 The main weakness of this research is that it is limited to the perspectives of families
22 living in the UK, who speak English. Empowerment is likely to be influenced by
23 culture⁷, and so Empowerment would benefit from validation in other cultures and
24 languages. A further weakness is that patient participation rates were low. However,

1 because this work builds on extensive previous qualitative research^{4 5 11 12}, and because
2 care was taken to reach data saturation, it is unlikely that significant issues were missed.
3 The main strength of this research is that the new PRO identified – Empowerment - has
4 been developed through consultation with (1) people struggling with the challenges of
5 having a genetic condition in their family and (2) clinicians with experience of helping
6 these families. They are the “experts” consulted in this research to identify the patient
7 benefits from using clinical genetics services. The traditional approach to PROMs
8 development is to use theoretical frameworks to identify PROs, and this approach was
9 used in identifying PPC as a patient outcome from genetic counselling^{14,26}. This
10 approach is useful, but as the present study shows, consulting patients and clinicians can
11 enable new constructs to be developed, and existing constructs to be extended, to ensure
12 that PROs include all benefits valued by patients.

13 A measure of PPC was developed and evaluated in Hebrew¹⁴. When the paper reporting
14 development of the PPC questionnaire was published in a US journal, the PPC was
15 translated into English¹⁴. Whilst the English translation of the PPC has been used with
16 English speaking samples²⁷, it has not yet been validated for use with a UK population.
17 A study is underway to operationalise Empowerment by developing a new genetics-
18 specific PROM of Empowerment, using the PPC measure and the qualitative data
19 collected in this and previous studies^{4 5 11 12}. Both PROMs and patient empowerment
20 have been identified as key aspects of current UK health policy²⁸. In addition, there was
21 a recent call for better measures to capture aspects of Health Related Quality of Life
22 (HRQoL) in clinical trials in medical genetics²⁹. These developments strengthen the
23 argument in support of investing the time and resources needed to develop a new
24 genetics-specific PROM to capture Empowerment. This PROM would then be available

1 to capture HRQoL outcomes in clinical trials, and in clinical practice to generate data to
2 support continuous quality improvement³⁰. As part of PROM development¹⁶, factor
3 analysis will investigate the dimensional structure of Empowerment to confirm whether
4 or not Empowerment is a single multidimensional construct.

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10 **CONFLICT OF INTEREST:**

11 The authors declare no conflict of interest.

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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 & 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 ^I	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	<ul style="list-style-type: none"> • 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 	<ul style="list-style-type: none"> • 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 			

^lThis group is made up of 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 & focus groups) n = 27
*Patient advocate	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

*Not from a family affected by a genetic condition