

Presenting life with cystic fibrosis: a Q-methodological approach to developing balanced, experience-based prenatal screening information

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Abstract

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Background Cystic fibrosis (CF) is one of the most common life-threatening genetically inherited conditions and prenatal screening for CF is available in many countries. Genetic counsellors and other health professionals are expected to provide information about the condition in a way that facilitates personal decision making. Knowing what information to deliver about complex genetic conditions to support informed screening decisions can be challenging for health professionals.

Objective To solicit views from those with personal experience with CF on which aspects of the condition they consider most important to include in prenatal screening materials.

Methods Q-methodology; an approach to systematically explore variations in viewpoint that combines factor analytic techniques with qualitative approaches to pattern interpretation. Setting and Participants: Twelve adults with CF and 18 parents of affected children were recruited from a regional centre in the UK.

Results Five distinct viewpoints on the items most and least important to include in screening information were identified: Factor 1 the normality of life with CF and increasing life expectancy; Factor 2 the hardships and reduced lifespan. Factor 3 medical interventions and the importance of societal support. Factor 4 longer-term consequences of CF. Factor 5 the ability to adjust to the condition.

Discussion The identification of five different views on what represented the most and least important information to include about CF highlights the challenge of portraying a complex genetic condition in a balanced and accurate manner. Novel ways in which Q-methodology findings can be used to meet this challenge are presented.

Introduction

Information provided via prenatal or genetic testing services is intended to help pregnant women, and their partners make informed decisions about their testing options.^{1,2} Many prospective parents have had little personal experience with the conditions for which testing is available, and health-care services are frequently the sources of information on these conditions. However, it has been recognized that delivering balanced information about complex genetic conditions such as cystic fibrosis (CF) to facilitate informed decision making is challenging for health professionals.³

Cystic fibrosis is one of the most common life-threatening genetically inherited conditions affecting Caucasians: around 1 : 25 people are carriers of a CF gene mutation, and there is an incidence of 1/2500 births.⁴ CF is a chronic, multiorgan disease characterized by frequent respiratory infections, bronchiectasis, male infertility and pancreatic malabsorption. Life expectancy is significantly reduced, and current median predicted survival is approximately 34 years for those living in countries with access to appropriate health care.⁵ Survival continues to improve, however, and babies born today with CF have a life expectancy over 50 years.^{6,7} Routine newborn screening for CF has played an important role in earlier access to treatment and management, but whether this directly translates into a reduction in child deaths is uncertain.⁸ New approaches to treatment for CF, for example stem-cell and gene therapies, continue to be sought but currently do not impact on life expectancy assessments.⁹ However, there is much promise of treatments that are specific to the gene mutation, which do address the underlying genetic defects.¹⁰ Despite such progress, for many people with CF survival is still dependent on frequent hospitalization, and a lifetime regimen of medication and therapies that are time-consuming and arduous.

Prenatal testing for CF takes a variety of forms. Carrier screening for CF (testing asymptomatic individuals to see if they carry a copy

of a known gene mutation) is available in many countries although health policies vary on how and to whom it is offered.¹¹ In the United States, for example, practice standards state that all Caucasian couples be offered CF carrier testing as part of pre-conceptual or prenatal care.¹² In contrast, the UK does not routinely screen for CF carriers during pregnancy unless there is a history of CF or there has been identification of risk factors, such as echogenic bowel via ultrasound scanning. CF is a recessive genetic condition and when carrier testing reveals that both parents carry a known mutation the mother is offered invasive diagnostic testing. Following a positive diagnosis (and in those countries where termination of pregnancy is permitted), parents are offered a choice whether to continue the affected pregnancy. Up-to-date and balanced information about CF, for example, clinical features, medical management and life expectancy is important to understand when making an informed choice about prenatal testing.¹³

Parents value and use information about the impact of a genetic condition on quality of life (QoL) in their prenatal testing decisions.^{14–16} Parents without direct knowledge of a condition will consider second-hand experiential information in their prenatal testing decisions, yet such information, including that from providers, may be biased or incomplete.¹⁷ Research shows that knowledge about QoL in relation to CF is frequently low in couples undergoing prenatal carrier screening for the condition¹¹ and that increasing such knowledge affects attitudes towards testing.¹⁷ Providing balanced and accurate information is therefore important for informed test decision making, but there are a number of specific challenges associated with developing balanced information about a complex condition such as CF. One is that variations in genotype and the role of other modifier genes lead to wide phenotypic variability. In addition, environmental factors and adherence to treatment impact on the severity of the condition. Quality of life for people with CF is very difficult to predict as perceptions vary not only with disease stage

and severity, but also by individual factors such as life stage and gender.¹⁸

One study that considered the issue of balanced information provision in CF carrier-testing leaflets identified that in comparison with leaflets aimed at screening a non-pregnant population, information about the condition in prenatal testing information was more negative in tone.¹⁹ The authors suggested that a 'relatively equitable proportion' of 'negative', 'neutral' and 'positive' information statements should be used to construct a balanced account of CF in prenatal testing materials. For example, (negative) information about reduced life-span should be balanced with (positive) information about treatment developments for CF. However, it is argued that balanced information is not simply that which presents equal amounts of 'bad' and 'good' news. A different approach has been influenced by calls for prenatal testing information to incorporate knowledge from those with direct experience of living with the condition as a way of counterbalancing the (more negative) medical viewpoint.^{20,21}

Incorporating experiential knowledge into all types of prenatal testing information introduces its own challenges as perceptions of what is a balanced account of a condition vary.²² Health professionals delivering prenatal testing in the UK for example are situated in a position of inherent tension as they are required to provide information that both supports reproductive autonomy *and* represents disability in a way that does not negatively impact on the status of people who have the conditions.²³ It has been argued that prenatal testing and abortion is a problematic way for society to deal with those who have an impairment,^{24,25} and some adults with CF have taken issue with the way the condition is presented as an automatic candidate for screening.²⁶ Commentators taking a disability rights perspective have asserted the need to provide more realistic accounts of life with a condition provided by those people most closely affected by them.²⁷

It is increasingly recognized that prenatal testing information must cover the issues that people with genetic conditions and their fami-

lies believe to be important²⁸; however, incorporating personal experience into information materials does not necessarily mean that the end result is 'balanced'. One web resource developed specifically to put the narratives of those affected by tested-for conditions at the centre of prenatal testing information material was evaluated by a range of stakeholders including those with personal experiences of the featured conditions.²² In the case of CF, a group of mothers of affected children did not feel the personal testimony of the adult with CF included in the resource captured the variation in severity of the condition and as such was unbalanced. The conclusion was that these narratives counter-balanced the medical tragedy accounts of genetic conditions, but in themselves were not balanced.²² This is of concern because first-person patient narratives are popular with people making difficult health-care choices and are increasingly used in health information, yet there is some evidence that this direct use of personal experience may inadvertently bias patient decision making in unpredictable ways.²⁹

To date, development of patient information using experiential knowledge has used conventional qualitative approaches to capture personal narrative.^{30,31} In contrast, we present a study that used Q-methodology to develop balanced information about CF for use in screening information. Q-methodology offers a promising alternative to developing balanced information based on experiential knowledge as it is designed to identify competing viewpoints on a topic, for example 'life with CF'.³² Allowing participants to respond to things others have written or said about CF provides a way of 'depersonalizing' personal accounts. This may be a useful alternative to the use of first-person narratives, especially as the latter may be more associated with persuasive effects.^{29,30}

Research aims

In this study, we aimed to utilize the experiential knowledge of adults with CF and parents of affected children to identify competing

viewpoints on which aspects of the condition they considered most important to include in prenatal screening information.

Method

The study received ethical approval from a UK National Health Service Research Ethics Committee (REC reference number 08/H1306/18).

Designing the Q-set

Q-methodology combines quantitative research techniques and analysis with qualitative approaches to sampling and pattern interpretation.³³ The method requires participants to consider and respond to a set of statements (the Q-set) extracted from the 'field of shared knowledge and meaning' on the topic under investigation.³⁴ To do this, participants use a ranking technique (the Q-sort) to allow them to express their view towards a diverse range of things already 'written or said' about the topic.³² In this study, a search of the academic literature was conducted using search terms initially based on key phrases and words in literature already identified. The search terms were refined throughout the search process as potential new key terms emerged. Electronic databases, including Psych-Info and Medline were the main sources of academic literature and the reference section of relevant papers, were also hand-searched. Statements were extracted from papers that addressed personal experiences of CF, for example, from a QoL perspective. The search for articles continued until no new statement themes emerged. A search of non-academic sources (support group websites, magazines, television programmes and newspapers) was also conducted. It was not our intention to conduct a comprehensive search of these sources but just to identify recent and topical examples of a non-academic perspective on life with CF. For example, an interview in a women's magazine with a woman with CF was found, and the lead researcher watched a television programme about a young English man with CF. Newspaper articles were searched for

articles on the then Prime Minister of Great Britain (Gordon Brown), whose son had recently been revealed to have CF.

In this study, two interviews were conducted with individuals who had long-term experiences of CF. The interviews were designed to provide additional material on themes underrepresented in the literature, for example, views on the impact of the condition on schooling and employment. The first interviewee was a parent of two teenage children with CF who attended the outpatient service where the study was conducted. The second was an adult with CF who volunteered after hearing about the study. The interviews focused on exploring how CF affected the quality of life of individuals and family members at key life stages.

From these different sources, 154 different statements about CF were extracted. Each statement was given a primary category under themes identified as important during an earlier review of the literature: lifespan, manageability, QoL, impact on society.³⁵ Each statement was given secondary classifications: (i) according to whether it broadly represented a medical, psychosocial or social model position on disability, (ii) whether it had been identified in academic or popular literature or an interview, and (iii) whether it conveyed a positive or negative message about CF. For example, the statement 'Many parents believe CF has made their family stronger' was classified as belonging to the primary category QoL and to secondary categories of: (i) psychosocial model, (ii) academic literature, and (iii) positive message. A range of further categories was identified which emerged from this process, and these were used to help refine the 154 items to a short list of 52 for a preliminary Q-set. Three researchers, one with expertise in Q-methodology, two medical specialists in CF and a consultant psychologist reviewed the Q-set. A pilot study was conducted with six individuals, one of whom was an adult with CF. Following this, some items were removed from the Q-set and some wording was changed to improve clarity. A final Q-set of 40 items (see Table 1) representing the original categories was selected.

Table 1 Factor arrays (item by ranked position)

		F1	F2	F3	F4	F5
1	At first, most parents think they won't be able to cope with a child with cystic fibrosis (CF) – but time and again, they find they can	+4	0	-2	0	+3
2	Nearly all men with CF are infertile	-3	-3	-3	0	0
3	People can still die very young from CF	0	+5	-2	+5	-5
4	Modern equipment means that children with CF have a lot more freedom than they used to and live a more normal life, e.g. they can go on sleepovers	+3	0	+3	+2	+4
5	The typical person with CF is now a fit individual with minimal if any symptoms, who happens to have a problem called CF	+3	-2	0	-2	+5
6	Following the introduction of new treatments and better disease management, quality of life has dramatically improved for people with CF	+3	+1	+5	+4	+1
7	CF can be isolating because of the risk of cross infection from other people with the condition	0	0	+3	-1	-1
8	Children with CF undergo numerous treatments, face frequent hospital visits and some have repeated medical procedures.	+1	+4	0	0	+2
9	Treatment for CF often includes physiotherapy for 20 minutes once or twice a day at home	+2	+2	+2	+1	+1
10	You can't know how badly a child will be affected with CF before it is born	+1	+4	-1	+3	+4
11	Women with CF who get pregnant can experience more problems with their health than other women	-2	-1	-2	+2	+1
12	Women with CF have greater difficulty getting pregnant	-3	-4	-3	-1	-3
13	Research suggests mothers of children with CF are more prone to decreased marital satisfaction and sexual intimacy	-4	-1	-4	-3	-2
14	Many couples whose first born has CF go on to have other children	+2	0	-3	-3	-4
15	Many adults with CF enjoy a good quality of life	+4	+2	0	+3	+1
16	It can be difficult for a woman to have career and raise a child with CF	-1	+1	0	-1	-2
17	More than a third of adults with CF are married or living with a partner	+2	-2	0	-1	-2
18	People with CF may have to stay in hospital to be given antibiotics intravenously	0	+3	+2	-1	-1
19	Ultimately lung transplant is often necessary as CF worsens	-2	+3	0	+1	+1
20	Many parents believe CF has made their family stronger	+2	0	-4	-3	-1
21	Many adults with CF have jobs or study, and are involved in a wide range of professions and occupations	+2	-2	+1	+1	-3
22	If people with CF have children, there is a risk that they will be left without a mother or father	-2	-1	-1	0	0
23	Negative attitudes from others can create extra difficulties for people with CF	-3	-4	-2	-3	0
24	As adults with CF get older, they are prone to complications such as diabetes, liver disease, pneumothorax, osteoporosis, and incontinence	0	+2	-3	+2	+2
25	The time and effort spent on the treatment regimen for a child with CF can have an impact on other family relationships	+1	+3	-2	-2	0
26	The brothers and sisters of children with CF often feel they are missing out	-1	+3	+1	-2	+3
27	The improvement in outlook for people with CF has been achieved by increasingly complex and demanding treatments	0	0	+4	0	+2
28	Up to half of patients with CF currently waiting for a lung transplant will die before a donor becomes available.	-3	+2	+2	+1	-1
29	Adolescents with CF whose condition is not severe say they have a good quality of life	+3	-1	-2	+2	0
30	Many people with CF believe it has made them stronger people, and given added meaning to their lives	+1	-3	+1	-4	0
31	The improvement in outlook for people with CF has only been achieved by increasingly expensive treatments, which the NHS is expected to pay for	-4	-3	3	-5	-2
32	One challenge for people with CF is managing variability in their health on a daily basis	0	-2	+1	+3	+2
33	Gene therapy for CF does not appear to be a realistic option in the near future	-2	+1	+3	-2	-3
34	Half of patients with CF who have a lung transplant are still alive ten years later	-1	+1	+1	+3	+2
35	Increasing numbers of women with CF are having children	-1	-3	-2	+2	-4

Table 1. Continued

		F1	F2	F3	F4	F5
36	Research shows children and young people with chronic conditions like CF are much more likely to develop emotional and behavioural difficulties.	-1	-2	-1	-2	-4
37	Many treatments, that either target the underlying defect in CF or its direct consequences, are currently being developed	+1	+1	+3	+1	+3
38	People with CF have to pay just over £100 a year for prescriptions	-5	-5	+2	-4	-1
39	Children born with CF now are likely to live well beyond 40	+5	+2	+1	+4	+3
40	Because many people with CF are living longer, they may increasingly be cared for by their children or partners, rather than their parents	-2	-1	-5	0	-3

Participants

Potential participants (adults with CF or parents of affected children) who were attending routine outpatient appointment at the regional CF centre were invited to participate by staff. Recruitment was carried out over 4 months in 2009 via convenience sampling, that is, during the time period we aimed to get as many patients who met the eligibility criteria as possible. Patients who were significantly unwell were not approached. Response rates were not recorded but out of those directly approached, agreement to participate in the research was high and, subjectively, there did not appear to be a pattern in terms of who declined, for example, in terms of socio-economic status. Sample characteristics were periodically reviewed to ensure diversity in education, age, family circumstance and experience of CF. For example, the parent participants included a bereaved father, the parent of a teenager who would need a liver transplant and a mother whose daughter had such mild symptoms that she was not diagnosed until she was ten. One participant with CF had undergone a lung transplant, and another frequently spent time in hospital having intravenous antibiotics. In contrast, one woman had rarely been unwell, and another had such mild symptoms she had remained undiagnosed until her mid forties.

Thirty participants carried out a Q-sort: 12 were adults with CF and 18 were parents of children with CF. Of the parent group, 17 were mothers with one father. One parent was the

mother of an adult participant and was recruited via him. Family size ranged from one to three children; two parents had two children with CF; several parents had terminated pregnancies where the foetus had a diagnosis of CF. For parents recruited via the children's service affected children ranged in age from 8 months to 14 years. Ten parents reported having left formal education at 18 years or younger and eight had gone on to higher education. Adults with CF (six men, six women) ranged in age from 23 to 47 years. Nine reported having left formal education at 18 years or younger, and three had gone on to higher education. One participant was of South Asian heritage, and one was of mixed African–Caribbean heritage. The remainder of the participants were White British.

Procedure

Q-sorting took place at the participants' homes. Informed consent was sought from all participants. Participants were given a shuffled set of cards (one item per card), which they sorted according to a verbal instruction to consider which items are most and least important for pregnant women to take into account when deciding whether or not to have prenatal screening for CF. In a series of steps, participants ranked the items by placing each card in a cell on a blank grid (see Fig. 1). Each column of the grid represented a response from +5 (most important) to -5 (least important). In this way, a set of ranked data was collected

												32. One challenge for people with CF is managing variability in their health on a daily basis											
												21. Many adults with CF have jobs or study, and are involved in a wide range of professions and occupations	37. Many treatments, that either target the underlying defect in CF or its direct consequences, are currently being developed	27. The improvement in outlook for people with CF has been achieved by increasingly complex and demanding treatments	36. Research shows children and young people with CF are much more likely to develop emotional and behavioural difficulties	40. Because many people with CF are living longer, they may increasingly be cared for by their children or partners, rather than their parents							
												29. Adolescents with CF whose condition is not severe say they have a good quality of life	*20. Many parents believe CF has made their family stronger	30. Many people with CF believe it has made them stronger people, and given added meaning to their lives	24. As adults with CF get older, they are prone to complications such as liver disease, pneumothorax, osteoporosis and incontinence	35. Increasing numbers of women with CF are having children	33. Gene therapy for CF does not appear to be a realistic option in the near future	28. Up to half of patients with CF currently waiting for a lung transplant will die before a donor becomes available					
												new treatments and better disease management, quality of life has dramatically improved for people with CF	than a third of adults with CF are married or living with a partner	and effort spent on the treatment regimen for a child with CF can have an impact on other family relationships	CF may have to stay in hospital to be given antibiotics intravenously	patients with CF who have a lung transplant are still alive ten years later	with CF have children, there is a risk that they will be left without a mother or father	23. Negative attitudes from others can create extra difficulties for people with CF					
												15. Many adults with CF enjoy a good quality of life	5. The typical person with CF is now a fit individual with minimal if any symptoms, who happens to have a problem called CF	*14. Many couples whose first born has CF go on to have other children	10. You can't know how badly a child will be affected with CF before it is born	7. CF can be isolating because of the risk of cross infection from other people with the condition	26. The brothers and sisters of children with CF often feel they are missing out	19. Ultimately lung transplant is often necessary as CF worsens	12. Women with CF have greater difficulty getting pregnant	31. The improvement in outlook for people with CF has only been achieved by expensive treatments, which the NHS have to pay for			
												39. Children born with CF now are likely to live well beyond 40	1. At first, most parents think they won't be able to cope with a child with CF, but time and again, they find they can	4. Modern equipment means that children with CF have a lot more freedom than they used to and live a more normal life, e.g. they can go on sleepovers	9. Treatment for CF often includes physiotherapy for 20 minutes once or twice a day at home	8. Children with CF undergo numerous treatments, face frequent hospital visits and some have repeated medical procedures	3. People can still die very young from CF	16. It can be difficult for a woman to have career and raise a child with CF	11. Women with CF who get pregnant can experience more problems with their health than other women	2. Nearly all men with CF are infertile	13. Research suggests mothers of children with CF are more prone to decreased marital satisfaction and sexual intimacy	38. People with CF have to pay just over £100 a year for prescriptions	
+5	+4	+3	+2	+1	0	-1	-2	-3	-4	-5													
Most important to include in testing information												Least important to include in testing information											

* The placement of items in bold distinguished this factor from all others at $p < 0.01$

Figure 1 Composite Q-sort for Factor 1. Presenting life with cystic fibrosis as a 'normal life'.

for each participant, the pattern of the Q-sort representing their views about the relative importance of the items. Participants were encouraged to make comments on the items, and these were recorded in a booklet.

Analysis and interpretation

The Q-sort data were managed and analysed using a Q-software package PQMethod, V. 2.11.³⁶ Q-Methodology uses factor analysis to

Table 2 Characteristics of participants whose Q-sorts exemplified each factor

	Adults with CF	Parents of children with CF
Factor 1	A woman in her late thirties and a man in his early forties. Both described themselves as being in good health.	One mother with two children with CF. One father with a child with CF and a previous child with CF who had died. Five mothers who had one child with CF and at least one other child without CF
Factor 2		Four mothers, each with one child with CF and one child without CF
Factor 3	A woman in her early twenties who had undergone a lung transplant	A mother of a baby with CF and an older child without CF
Factor 4	Three men and one woman with CF in their twenties	
Factor 5	A woman with CF in her mid thirties	A mother of a 10 year old child with CF and two other children without CF.

identify relationships between individual Q-sorts. Each factor represents a highly inter-correlated cluster of Q-sorts sorted in a statistically similar way. Each cluster is considered to represent a distinct point of view on the given topic. Using the strategy described by Watts and Stenner,³³ and using PCA and Varimax rotation, a five-factor solution was identified as the 'best fit' for the data, that is, the maximum number of interpretable and distinct viewpoints. Each of the factors had at least two Q-sorts loading highly and at $P < 0.01$ on one factor only (known as exemplar sorts). Table 2 provides information on the participants whose Q-sorts were exemplars of each factor. A weighted averaging formula was used to produce an 'average' composite Q-sort from the exemplar sort to represent each factor (see Table 1).³⁷ These composite sorts were reconstructed on grids to support interpretation; for example, Fig. 1 represents the composite Q-sort for Factor 1. During interpretation, particular attention was given to the position of the 'most important' and 'least important' items and the statements identified as statistically distinguishing for each factor at $P < 0.01$.

Findings

A description of each factor is now presented. The comments provided by participants were used to inform, support or challenge factor interpretations, and these are presented where they clarify the interpretation. For each factor,

a label summarizes the central message of the viewpoint.

Factor 1: 'Presenting life with CF as a normal life'

The central message of this factor was that life with CF is essentially a normal one. Participants considered Item 39 (children born with CF now are likely to live well beyond 40 years) as the most important thing that prospective parents should know. Other items considered important were those that emphasized the probability of a good quality of life for the person with CF and their parents, the ability of parents to cope and normal life experiences including relationships and children for those with CF. The placement of Items 14, 17 and 20 (see Table 1) statistically distinguished this factor from all others. Unlike any other factor, Factor 1 participants agreed it is important to convey that having a child with CF can strengthen a family. A mother with two children with CF said, '*I think it's important for people to know it's not all doom and gloom. A lot of it is down to attitude and being positive and not always fearing the worst*'.

Participants gave a low priority to information about the possible negative long-term consequences of CF, for example infertility or the potential need for a lung transplant. Nevertheless, they agreed that information about the 'burden' of treatments was still important (Items 8, 9 and 25). The financial costs of CF to the individual and the UK National Health

Service were considered least important to include along with information that mothers of affected children may experience lowered marital satisfaction.

Factor 2: 'Presenting CF as a serious threat to health, happiness and a normal life'

The central message of Factor 2 was that CF is a serious life-threatening health condition with major implications for the individual and their family. As in Factor 1, participants considered information about lifespan as the most important to convey to prospective parents but in contrast selected Item 3 (people can still die very young from CF) as the 'most important'. As one mother said, *'That's the first thing you think about. You definitely need to know that'*. The potential for living beyond 40 years was considered to be lower priority (Item 39) for this factor.

Other important items were those confirming the difficulty of predicting severity and that the burden of treatments on the individual and their family can be significant. A mother of a young child with CF said, *'Treatment takes so much time up.... Hospital takes over your life: it's majorly important'*. In this view, it is more important to present information about the immediate life difficulties of the child rather than the (potential) future life as a teenager or adult. The placement of Items 16 and 25 distinguished this factor statistically demonstrating the view that the impact of CF on the family, and especially the mother, should be taken into consideration by those considering screening. Participants did not agree that CF makes people or families stronger. Items that portray life with CF as difficult were prioritized over positive aspects. As the mother of a young child with CF said, *'You don't just want to put all the good points as most important, because otherwise you're encouraging people to have kids with CF when they don't know how bad it is'*.

Factor 3: 'Presenting quality of life with CF as dependent on medical intervention'

The main message of Factor 3 was that for people with CF, a 'normal' life was likely to be

one highly dependent on a regimen of medications and therapies and that health services had a very important role in upholding the QoL of an affected individual. Participants placed little importance on information about family adjustment and coping, impact on personal relationships or the long-term negative consequences of CF.

The placement of Items 31 and 38 statistically distinguished this factor and emphasized the possible financial implications of CF, for example, prescription charges. The woman with CF said, *'very important, because people can't really afford it'*. Some difficulties, such as women trying to have a career while raising an affected child, were also seen as being due to a lack of social support. For example, in response to Item 16, the mother of the baby with CF said, *'True. I was going back to work, but now it's absolutely impossible, you can't work round it. It's very important [to know], because I feel there's no help or support'*.

Factor 4: 'Presenting CF from the life-course perspective'

Exemplars of this factor were all adults with CF and most strongly emphasized the need for information to convey the longer-term view of life with the condition. Items considered important to include balanced the possibility of an early death with the potential for a longer, good quality life and possibilities for parenthood. The need to convey information about the range in severity and variability of health was also emphasized.

This factor was distinguished from all the others by the high rating of information about the fertility and parenthood prospects of the person with CF (Items 2, 11, 12, 35, 40). Comments suggested that participants wanted information to capture a 'life' with CF, not just discrete problems. One man with CF said, *'It's like anything, there are good times and bad times'*. Assumptions about the burden of treatment were not always supported. One man said, *'I quite like going into hospital. It's time off work and a nice holiday. I like peace and quiet – they leave me alone'*. Factor 4 most

strongly rejected the idea that parents should be told CF makes people stronger or adds meaning to their lives.

Factor 5: 'Presenting life with CF as one of adjustment and living in the present'

The central message was that while the severity of CF cannot always be known in advance and health will vary over time, most parents and affected individuals adjust and cope (Items 10, 4 and 5). The woman participant with CF said, '*If you decide to have a child, you take what comes*'. Higher priority was given to presenting positive information and to demonstrating that people with CF can be healthy. This factor emphasized the importance of living in the present and the fact that some people affected with CF can still die very young was considered least important to convey. One mother said, '*I don't know [what will happen in the future] and I don't want to think about it*'.

Including information about the potential for people with CF to have a partner, a job and children was seen as a low priority. Low priority was also given to the possible impact on a mother's career and the fact that many couples with an affected child will subsequently have non-affected children.

Discussion

This study aimed to identify competing viewpoints on the aspects of CF that those with personal experience of the condition considered most important to include in prenatal screening information. Although the main focus of the task was on information development, the findings reflect previous work showing that those with outwardly similar experiences can have very different perspectives on life with the same condition.³⁸ Personal factors, including CF related experiences, disease severity and family resources may, for example, have influenced the viewpoints expressed and may help explain the conflicting views of Factors 1 and 2, and the concerns about financial and social support in Factor 3. All the participants with CF had survived to

adulthood, which may underlie their more 'future focused' views and the need to 'live in the present' as expressed in Factors 4 and 5. In-depth interviews were not conducted, however, in the initial short background interviews, participants across all factors reported a wide range in problems and experiences. It is unlikely that viewpoints were solely related to specific life experiences or severity of the condition *per se*. For example, the mothers who were exemplars of Factor 2 reported significant health problems in their children, but some Factor 1 exemplars had experienced CF related bereavement and similar difficult health challenges. In support of this view, a review of studies investigating adaptation to living with genetic conditions, including CF, suggested that disease severity is less related to successful adaptation of parents or individuals than effective coping strategies.³⁹ However, the Q-sorts represent a snapshot in time. One parent, an exemplar of Factor 1, said, '*When [son] was diagnosed, they said he'd have a port-a-cath and be in hospital every 3 months. I'd have answered very differently then*'. The severity of CF is therefore not a static characteristic of the disease and ability to cope may also fluctuate over time. In addition, as new treatments are developed the outlook for some children born with CF may dramatically improve. This highlights the need to regularly revisit condition-related information in prenatal testing materials.

This research highlights once more the challenge of portraying a complex genetic condition like CF in a way that can be considered 'balanced' by all interested parties. Opinions on what represented the most and least important information to include in prenatal screening information about CF varied considerably across accounts. Although CF is considered a serious life-shortening condition, some participants expressed conflict or uncertainty when considering where to place some of the 'negative' items of information on the Q-grid. For example, in relation to Item 19, a participant with CF in Factor 1 said, '*They do need to know the full picture [about lung transplantation]...*

but why mention it at this moment in time?' In response to Item 40 a woman with CF said, 'I think that's true [that partners or children may become carers of those with CF] but I wouldn't want it to put people off.' These participants may have considered these items as too abstract or long-term to be relevant at the prenatal screening stage. Alternatively, it may be difficult for those for whom coping with CF means living in the present and 'being positive' to want to include rather negative, long-term aspects of the condition in prenatal screening information.

In contrast to the arguments already presented that chronic illness and disability have been too negatively portrayed in prenatal testing information, it has also been suggested that the use of personal accounts leads to an overly positive representation of serious health problems.²² As already identified, one person's perception of balance is another's bias and so subjective assessments of this kind have limited value. However, for a balance of perspectives to be present, the views of those who consider 'negative' information about a condition to be important need to be valued equally with the views of those who prefer a more 'positive' view. While many parents successfully adapt to having a child with a disability or genetic condition not all do, and this is an important factor to consider when trying to utilize a range of experiences in prenatal testing information development.³⁹

We employed Q-methodology specifically because it helps identify competing viewpoints on complex issues and provides a more objective approach to the selection of experience-based material to include in prenatal screening information. Clearly defining the constituent elements of separate and sometimes conflicting views of CF is important for the development of a balanced portrayal. Nevertheless, from a practical perspective, integrating contradictory viewpoints into health resources presents a challenge: we suggest three ways in which the findings obtained using Q-methodology could be employed in the development of prenatal screening information.

1. *The creation of third-person narratives* One approach to using the findings of a Q-study would be to construct a set of short third-person accounts based on the individual factors. Presenting separate accounts may help parents clarify their own values as they consider which perspectives they most identify with and those they reject.³⁰
2. *Using factor exemplars to select interviewees* Where the use of first-person narratives is required or preferred, an evidence-based rationale for interviewee sampling would be to select individuals who were factor exemplars across the range of views.
3. *Integrating factors to create a balanced summary* Another option would be to create a single integrated account to represent a range of differing views. For example, in this study, if all items ranked +5 and +4 across the different factors were integrated, the summary for CF would read something like this:
'People with CF can have a high quality of life but the degree to which a person will be affected cannot always be predicted before birth. A baby born today with CF is likely to live well into adulthood, but some will die in childhood. Modern treatment means that affected children have more freedom than they used to and live a more normal life. Medical advances mean people with CF can live healthier lives, but children and adults often require ongoing treatments, frequent hospital visits and medical procedures. These treatments can be complicated and are experienced as demanding by some parents and children. Many parents find that they can cope with a child with CF better than they might have expected, but some parents do not.'

The advantage of the third approach would be that conflicting viewpoints are integrated in a way that is familiar to many users of health information. However, the selected content would be underpinned by research evidence and would represent the experiential knowledge of those who have a diverse range of experiences and differing perspectives. Such a

summary could be presented in written format as part of a leaflet or website, or communicated verbally in addition to traditional general facts information based on medical and clinical evidence, which is also valued by prospective parents.³⁰ Other key stakeholders, for example, health-care providers and patient support organizations should therefore be included in the development of test information.

This study had some limitations. Although the experience of participants varied according to disease severity, family economic circumstances and experiences of bereavement, all participants used the same CF services. We did not include children or people with CF who were significantly unwell; their viewpoints may be different from those expressed in this study. Only one father participated; therefore, parental reviews mainly reflected those of mothers. It is also important to recognize that parents vary in their information requirements at different stages of the testing pathway. This study required participants to consider information for prenatal screening only. We acknowledge the important differences between screening and invasive diagnostic tests and that more detailed information about health-related implications, for example, may be of benefit to parents at the diagnostic decision-making stage. Finally, this study focused on prenatal testing, something which is inextricably linked with termination of pregnancy. Pre-conceptual testing for CF carrier status is available, and it may be more appropriate for some couples than testing during pregnancy; it is possible that participants may have prioritized some items differently if the development of pre-conception testing information had been the aim.

Conclusion

There are currently no guidelines on how to develop balanced, condition-related information within prenatal or other genetic testing information. Research suggests that when delivering information about conditions, health professionals select the information to convey;

inevitably therefore, judgments about what constitutes balanced information remains with the individual professional.^{23,40} Where the support of autonomous informed choice is a goal, parents making important reproductive decisions should be able to access information that supports a range of viewpoints. There is a need for more systematic and objective approaches to the development of this kind of information, and we have argued that Q-methodology provides one such approach. In particular, the strength of applying Q-methodology to this problem lies in its original premise that all diverse viewpoints on a socially debated topic are equally valid. Using Q-methodology in the ways we suggest here may help the development of information materials that better support prenatal testing decisions for CF. While it is recognized that the application of Q-methodology to developing health information is novel and needs further refinement, we believe it has the potential to make a significant contribution beyond the prenatal screening context.

References

- 1 Raffle AE. Information about screening: is it to achieve high uptake or to ensure informed choice? *Health Expectations*, 2001; **4**: 92–98.
- 2 NICE. *NICE Clinical Guideline 62. Antenatal Care: Routine Care for the Healthy Pregnant Woman*. London: NHS, 2008.
- 3 Entwistle V. Considering 'balance' in information. *Health Expectations*, 2007; **10**: 307–308.
- 4 Walters S, Mehta A. Epidemiology of cystic fibrosis. In: Hodson M, Geddes D, Bush A (eds) *Cystic Fibrosis*, 3rd edn. London: Hodder Arnold, 2007: 433–441.
- 5 Cystic Fibrosis Trust. *UK CF Registry Annual Data Report 2009*. Cystic Fibrosis Trust: Bromley, 2011.
- 6 Dodge J, Lewis P, Stanton M, Wilsher J. Cystic fibrosis mortality and survival in the UK: 1947–2003. *European Respiratory Journal*, 2007; **29**: 522.
- 7 Anderson P. Emerging therapies in cystic fibrosis. *Therapeutic Advances in Respiratory Disease*, 2010; **4**: 177–185.
- 8 Grosse SD, Rosenfeld M, Devine OJ, Lai HJ, Farrell PM. Potential impact of newborn screening for cystic fibrosis on child survival: a systematic

- review and analysis. *The Journal of Pediatrics*, 2006; **149**: 362–366.
- 9 Conese M, Ascenzioni F, Boyd AC *et al.* Gene and cell therapy for cystic fibrosis: from bench to bedside. *Journal of Cystic Fibrosis*, 2011; **10** (Suppl. 2): S114–S128 [doi: 10.1016/S1569-1993(11)60017-9].
 - 10 Clancy JP, Rowe SM, Accurso FJ *et al.* Results of a phase IIa study of VX-809, an investigational CFTR corrector compound, in subjects with cystic fibrosis homozygous for the F508del-CFTR mutation. *Thorax*, 2012; **67**: 12–18.
 - 11 Green JM, Hewison J, Bekker HL, Bryant LD, Cuckle HS. Psychosocial aspects of genetic screening of pregnant women and newborns: a systematic review. *Health Technology Assessment*, 2004; **8**.
 - 12 American College of Obstetricians and Gynecologists. Update on carrier screening for cystic fibrosis. Committee Opinion No. 486. *Obstet Gynecol*, 2011; **117**: 1028–1031.
 - 13 Langfelder-Schwind E, Kloza E, Sugarman E *et al.* Cystic fibrosis prenatal screening in genetic counseling practice: recommendations of the national society of genetic counselors. *Journal of Genetic Counseling*, 2005; **14**: 1–15.
 - 14 Etchegary H, Potter B, Howley H, Cappelli M, Coyle D, Graham I, *et al.* The influence of experiential knowledge on prenatal screening and testing decisions. *Genetic Testing*, 2008; **12**: 115–124.
 - 15 Ahmed S, Atkin K, Hewison J, Green JM. The influence of faith and religion and the role of religious and community leaders in prenatal decisions for sickle cell disorders and thalassaemia major. *Prenatal Diagnosis*, 2006; **26**: 801–809.
 - 16 France EF, Locock L, Hunt K, Ziebland S, Field K, Wyke S. Imagined futures: how experiential knowledge of disability affects parents' decision making about fetal abnormality. *Health Expectations*, 2011; **15**: 139–156.
 - 17 Wilson J, Ferguson G, Thorn J. Genetic testing likelihood: the impact of abortion views and quality of life information on women's decisions. *Journal of Genetic Counseling*, 2011; **20**: 143–156.
 - 18 Gee L, Abbott J, Conway SP, Etherington C, Webb AK. Quality of life in cystic fibrosis: the impact of gender, general health perceptions and disease severity. *Journal of Cystic Fibrosis*, 2003; **2**: 206–213.
 - 19 Loeben GL, Marteau TM, Wilfond BS. Mixed messages: presentation of information in cystic fibrosis-screening pamphlets. *American Journal of Human Genetics*, 1998; **63**: 1181–1189.
 - 20 Bryant LD, Murray J, Green JM, Hewison J, Sehmi I, Ellis A. Descriptive information about Down syndrome: a content analysis of serum screening leaflets. *Prenatal Diagnosis*, 2001; **21**: 1057–1063.
 - 21 Petersen A. The best experts: the narratives of those who have a genetic condition. *Social Science & Medicine*, 2006; **63**: 32–42.
 - 22 Ahmed S, Bryant LD, Hewison J. 'Balance' is in the eye of the beholder: providing information to support informed choices in antenatal screening via AnSWeR. *Health Expectations*, 2007; **10**: 309–320.
 - 23 Madeo AC, Biesecker BB, Brasington C, Erby LH, Peters KF. The relationship between the genetic counseling profession and the disability community: a commentary. *American Journal of Medical Genetics Part A*, 2011; **155**: 1777–1785.
 - 24 Solberg B. Prenatal screening for Down syndrome: why we shouldn't? In: Kristiansen K, Vehmas S, Shakespeare T (eds) *Arguing about Disability: Philosophical Perspectives*. Abingdon, Oxon: Routledge, 2009: 185–202.
 - 25 Parens E, Asch A. The disability rights critique of prenatal genetic testing: reflections and recommendations. *Mental Retardation and Dev Disabilities Res Reviews*, 2003; **9**: 40–47.
 - 26 Kerr A. Understanding genetic disease in a socio-historical context: a case study of cystic fibrosis. *Sociology of Health & Illness*, 2005; **27**: 873–896.
 - 27 Shakespeare T. Choices and rights: eugenics, genetics and disability equality. *Disability and Society*, 1998; **13**: 665–681.
 - 28 Lewis C, Mehta P, Kent A, Skirton H, Coviello D. An assessment of written patient information provided at the genetic clinic and relating to genetic testing in seven European countries. *European Journal of Human Genetics*, 2007; **15**: 1012–1022.
 - 29 Winterbottom A, Bekker HL, Conner M, Mooney A. Does narrative information bias individual's decision making? A systematic review. *Social Science & Medicine*, 2008; **67**: 2079–2088.
 - 30 Entwistle VA, France EF, Wyke S *et al.* How information about other people's personal experiences can help with healthcare decision-making: a qualitative study. *Patient Education and Counseling*, 2011; **85**: e291–e298.
 - 31 Herxheimer A, McPherson A, Miller R, Shepperd S, Yaphe J, Ziebland S. Database of patients' experiences (DIPEX): a multi-media approach to sharing experiences and information. *The Lancet*, 2000; **355**: 1540–1543.
 - 32 Stainton Rogers R. Q Methodology. In: Smith JA, Van Langenhove L (eds) *Rethinking Methods in Psychology*. London: Sage, 1995: 178–192.

- 33 Watts S, Stenner P. Doing Q methodology: theory, method and interpretation. *Qualitative Research in Psychology*, 2005; **2**: 67–91.
- 34 Watts S, Stenner P. Q Methodology, Quantum Theory, and Psychology. *Operant Subjectivity*, 2003; **26** : 157–175.
- 35 Wright KF. *Which Features of Cystic Fibrosis might Inform Pregnant Women's Prenatal Testing Decisions?* Doctor of Clinical Psychology Thesis. Leeds, UK: University of Leeds, 2009.
- 36 Schmolck P, Atkinson J. PQMETHOD (Version 2.11) (2003). Available at: <http://www.rz.unibw-muenchen.de/~p41bsmk/qmethod/>; University of the Federal Armed Forces, Munich; accessed 20 June 2009.
- 37 Brown SR. *Political Subjectivity: Applications of Q Methodology in Political Science*. New Haven: Yale University Press, 1980.
- 38 Lowton K, Gabe J. Life on a slippery slope: perceptions of health in adults with cystic fibrosis. *Sociology of Health & Illness*, 2003; **25**: 289–319.
- 39 Biesecker BB, Erby L. Adaptation to living with a genetic condition or risk: a mini-review. *Clinical Genetics*, 2008; **74**: 401–407.
- 40 Sheets KB, Best RG, Brasington CK, Will MC. Balanced information about Down syndrome: what is essential? *American Journal of Medical Genetics Part A*, 2011; **155**: 1246–1257.