

•

Consultation with Individuals with Down syndrome about Non Invasive Prenatal Testing (NIPT)

1 March 2017

Dr Barbara Barter (Lead Facilitator and Clinical Psychologist)

**Professor Richard Hastings (Centre for Educational Development,
Appraisal and Research; University of Warwick).**

Facilitated by The Royal Mencap Society



In August 2016 The Nuffield Council on Bioethics commissioned Mencap to lead on gaining the views of individuals with genetic variations and a learning disability in order to inform the Council's project on non-invasive prenatal testing. Mencap asked Dr Barbara Barter to act as lead facilitator for this piece of work, in light of her previous research in this field (Barter, Hastings, Williams and Huw, 2016). Any views expressed in this report are the authors' own and not those of the Nuffield Council on Bioethics or Mencap.

With thanks to Dan Scorer and James Robinson (Mencap) for their support in facilitating the consultation.

Executive Summary

It is proposed that Non-Invasive Prenatal Testing will be offered via NHS services to women who are deemed at 'higher risk' following the current primary screen for trisomy (Down syndrome, Edward syndrome and Patau syndrome). The Department of Health issued a statement in November 2016 that the roll out of NIPT should be offered as a choice and it is likely to begin in 2018 or 2019 (Department of Health, 2016). In April 2016, The Nuffield Council on Bioethics set up a working group to consider the wider ethical implications of this proposed change in prenatal screening.

In August 2016, The Nuffield Council on Bioethics invited the Royal Mencap Society to lead on gaining the views of individuals with genetic differences and associated learning disabilities. Mencap invited Dr Barbara Barter (Clinical Psychologist) to lead this project in light of previous research.

The deadline of the Nuffield Council on Bioethics project meant that it was not possible to carry out a large scale consultation with individuals with learning disabilities. All too often, individuals with learning disabilities are omitted from influencing policy which may directly affect their lives. As such, it was considered that a small scale consultation could be completed to ensure that the voice of individuals with Down syndrome was not entirely excluded. It was agreed that the consultation would inform the larger project as completed by the Nuffield Council on Bioethics. It was agreed that a separate report would be written to outline the consultation with individuals with Down syndrome, placing any findings reported by the Nuffield Council on Bioethics in context.

Following ethical review by the Nuffield Council on Bioethics and an independent ethical review completed for Mencap, recruitment to the current consultation began in October 2016. Mencap made contact with a number of organisations including The 'Down Syndrome Association', 'Don't Screen Us Out', the 'Downs Syndrome Research Foundation' and a wide pool of Mencap groups. Invitations were posted on Mencap's social media channels to two information sessions. It is estimated that these invitations reached a potential 65,000 people on Facebook and a potential 186,000 on Twitter. Nine individuals with Down syndrome attended five information sessions whereby they met with Dr Barter (Lead Facilitator) and members of Mencap, and had the opportunity to discuss the consultation process, and understand or clarify information. Seven individuals took part in individual interviews in November and December 2016. The interviews were guided by particular questions and tailored to individual comfort

with the topic. Interviews from six participants were transcribed verbatim, and a process of thematic analysis was employed to analyse the data.

Summary of findings

The following themes and subthemes were identified and are summarised in the table below.

Current Consultation	Thematic Analysis (TA) What does Down syndrome mean? <ul style="list-style-type: none"> - Definition and understanding - Disability and difference - Self-Identity - Others' perception of Down syndrome. 	TA A test for Down syndrome <ul style="list-style-type: none"> - Previous knowledge - Old and new test - View of testing - Decision following testing. 	TA What does testing mean to me? <ul style="list-style-type: none"> - Personal impact - Impact on Down syndrome community. 	TA Balanced information <ul style="list-style-type: none"> - Balanced information and informed choice.
----------------------	--	---	---	--

Participants spoke of negative discourses about disability in society and a lack of understanding and fear of Down syndrome. This stood in stark contrast to their positive life experience and the life possibilities for anyone born with Down syndrome or disability.

In general, participants agreed with testing as a means of preparation and as an opportunity for prospective parents to gain information. Participants' views of termination varied from disagreeing to feeling unsure. Overall, participants were saddened and expressed disappointment with termination following testing, which could be understood in the context of their view of disability and their experience of living with Down syndrome. Although the participants were saddened and disappointed by decisions to terminate following testing, the majority also respected women's right to choose.

The negative discourses surrounding disability, decisions to test, and decisions taken following testing may have a direct or indirect impact on individuals on an emotional and practical level.

There was a consistent and strong call for 'balanced' information, including the possibility to meet people with Down syndrome, as a means of supporting educated or informed choice for women and families.

Authors' Recommendations

The findings from the current consultation and previous research by Barter, Hastings, Williams and Huws (2016) were triangulated leading to the following recommendations:

1. Because both studies were small and not representative of people with Down syndrome, further larger scale consultation is required to continue to build on this initial work.
2. People with Down syndrome and other disabilities should not be excluded from discussion about NIPT and similar issues.
3. Discussion with people with learning disabilities about complex and sensitive topics (such as NIPT) must be carried out in a thoughtful manner with preparation and support available, and adequate time for meaningful participation.
4. Individuals with Down syndrome may be aware of the issues around genetic testing, and clear information about the topic should be provided to them.
5. Further research is required to understand the diversity of living with any genetic variation.
6. Women or couples considering NIPT and its results should be offered opportunities to understand more about Down syndrome and the value of the lives of individuals with Down syndrome (e.g. through meeting people).
7. Given the perceived strength of the negative discourse surrounding Down syndrome, further work is required to understand what is needed to support informed choice.
8. Policy makers should take steps to mitigate the potential for the implementation and presentation of NIPT to be perceived by people with Down syndrome as questioning the value of their lives and themselves as 'wanted'.
9. As the technology, reach, and implementation of NIPT changes over time ongoing consultation with people with Down syndrome is necessary to capture the possibility of changing perspectives.

Content

1. Introduction

2. Consultation Methods

2.1 Methodological Approach

2.2 Procedure

2.3 Ethical Review

2.4 Recruitment

2.5 Information Session

2.6 Individual Interview

2.7 Participants

2.8 Analysis

3. Consultation Findings

3.1 Primary Analysis – Summary of findings from consultation

3.2 Secondary Analysis – Triangulation findings

3.3 Convergence of themes across studies

3.4 Difference across studies

4. Discussion

4.1 Summary of Findings

4.2 Accessible Discussion

4.3 Are people with Down syndrome aware of prenatal testing?

4.4 What is informed choice?

4.5 What is balanced information?

4.6 A message to people with Down syndrome.

5. Limitations

6. Authors' Conclusions and Recommendations

Appendix 1 – Detailed summary of findings

References

1. Introduction

In the UK screening for trisomy is offered to all pregnant women from weeks 10-14 of pregnancy and depends on a combination of biochemical and ultrasound tests. If the combined test shows the chance a foetus has trisomy (Down syndrome, Edward syndrome or Patau syndrome) is higher than 1 in 150, this is deemed a 'higher-risk' result. Currently, women who have a 'higher risk' result have the option of an invasive diagnostic test: amniocentesis or chorionic villus sampling (CVS) (UK National Screening Committee, 2016).

Traditional screening approaches have a relatively low accuracy. For example, the combined test will identify around 84% of women who have a foetus with trisomy. The exact level of risk of miscarriage caused by amniocentesis and CVS is contentious. Different studies suggest it ranges from 0.1 to 1.4 per cent for amniocentesis and 0.2 to 1.9 per cent for CVS (Nuffield Council on Bioethics, 2017).

Cell-free DNA (cfDNA) testing, known as Non-Invasive Prenatal Testing (NIPT) measures the underlying genetic makeup of trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome) and works by counting the number of placental cfDNA fragments from the different chromosomes present in the mother's blood during pregnancy. NIPT is more accurate than the combined test (Taylor-Philips et al., 2015) and carries no known risk of miscarriage (UK National Screening Committee, 2016).

The proposed change is for Non-Invasive Prenatal Testing to be offered via NHS services to women who are deemed at 'higher risk' following the current primary screen. NIPT is not diagnostic and an invasive diagnostic test is still required to receive a definitive diagnosis (UK Screening Committee, 2016). The Department of Health issued a statement in November 2016 stating that the roll out of NIPT should be offered as a choice and it is likely to begin in 2018 or 2019.

In April 2016, the Nuffield Council on Bioethics set up a Working Group to consider the wider ethical implications of this proposed change in prenatal screening. In August 2016, The Nuffield Council on Bioethics invited the Royal Mencap Society to lead on gaining the views of individuals with genetic differences and associated learning disabilities. Mencap invited Dr Barbara Barter (Clinical Psychologist) to lead this project in light of previous research.

Down syndrome is the most common chromosomal difference and occurs in about 1 in 1000 of live births worldwide (WHO, 2016). Edward and Patau syndrome are less common and occur in approximately 1 in every 1500 pregnancies and approximately 1 in every 4000 pregnancies respectively (SOFT UK, 2016). The number of Down syndrome pregnancies increased between 1989 and 2003 due to an increase in maternal age. However, changes in prenatal screening methods towards 2003 and an increase in prenatal diagnosis followed by pregnancy termination resulted in an overall drop in Down syndrome births in that same timeframe (Crane and Morris, 2006). In England and Wales, the number of live birth born with Down's syndrome varied from approximately 580 to 780 between 1989 and 2012 (Morris and Springett, 2014). (Raz, 2003) highlighted that as the quality of life and life expectancy for individuals with Down syndrome has increased, the number of live births has fallen.

The increasing use of genetic technologies has resulted in social debate on the appropriateness of their application. Alderson (2001) points out that prenatal testing policies demonstrate tensions between clinical support for women's and families' reproductive choices and public health programs aimed to reduce disease and disability.

A public consultation exercise carried out by the UK's Human Genetic Commission (2005) found a high level of support for an increase in prenatal testing, provided that the focus conditions were 'serious' or 'severe'. Evidence suggests that there is more support for prenatal diagnosis than for termination of a pregnancy (Green, Snowdon & Statham, 1993; Singer, 2004). It would appear that attitudes to testing and termination vary according to the condition in question (Green et al., 2004), vary across culture (Hewison, Green, Ahmed et al., 2007) and predictors of general attitudes to prenatal testing and termination include religious affiliation, church attendance, and traditional family attitudes (Balakrishnan & Chen, 1990; Krishnan, 1991). Green, Snowdon and Statham (1993) highlighted that factors such as age, reproductive history and education were related to attitudes to prenatal diagnosis.

Disability rights writers argue that prenatal testing is questionable on three counts: 1. That societal attitudes toward disability are based on medical models, 2. That it is morally dubious, and 3. That it is often based on misinformation about specific conditions (Parens & Asch, 2000). Parens and Asch argue that general societal perceptions of disability are negative and discriminatory and are based on medical and functional models rather than on the perception of disability as a socially constructed phenomenon. The current use of prenatal genetic testing

is gaining considerable momentum within the area of healthcare. This expansion of new technology has led to considerable debate surrounding the future direction of prenatal testing and the impact on society as a whole. Overall, the arguments put forward by the Disability Rights Movement have been criticised on the grounds that they are based on ethical and philosophical considerations, rather than on surveys or research evidence (Raz, 2003).

Ravitsky (2016) highlights a number of ethical and social implications of NIPT which fall in line with previous concerns raised by prenatal screening as well as intensifying some of them. She outlines that the rapid development of technology and the potential routinisation of NIPT raises concerns with regard to the capacity of health care providers to counsel women and families appropriately and the potential social and medical pressure which women may experience. Ravitsky also outlines that on one hand NIPT may exacerbate eugenic social attitudes and in turn increase the social stigma which individuals with disabilities may face, or on the other hand, may be used by anti-abortion lobbyists to justify strategies to limit women's reproductive autonomy.

All too often, individuals with learning disabilities are omitted from influencing policy which may directly affect their lives. Shakespeare (2006) suggests that more research of the experience of individuals with learning disabilities is imperative to understand the diversity of living with disability.

To date, there are very few published studies exploring the topic of prenatal testing with individuals with learning disabilities. Ward et al. (2002) reported on two workshops explaining the use of prenatal testing to individuals with learning disabilities. They concluded that the discussions were accessible but made little comment on the views of participants. Alderson (2001) interviewed five participants with Down syndrome to assess whether the condition involved costs, limitations or suffering and concluded that people with Down syndrome can lead fulfilling lives. Skotko, Levine and Goldstein (2011) surveyed 284 people with Down syndrome in the USA about their self-perception and recommendations to prospective parents, and concluded that an overwhelming majority indicated that they live happy and fulfilling lives.

Barter, Hastings, Williams and Huws (2016) carried out interviews with eight individuals with Down syndrome about their experience of the topic of prenatal testing. In this exploratory study, we concluded that with a thoughtful approach to the consent process the topic was accessible to individuals with Down syndrome. We outlined the experiences of individuals in

the study, and the wider social discourses that underpinned those experiences. The method and findings used in this original research study were used to support the current consultation.

2. Consultation Methods

2.1. Methodological Approach

In the present consultation, we used direct interviews with individuals with Down syndrome and a thematic analysis approach to the analysis of the interview data. Braun and Clarke (2006) outline that thematic analysis is a method for identifying, analysing and reporting patterns or themes within data. Thematic analysis is used to organise and describe data in detail and to interpret various aspects of the topic under investigation.

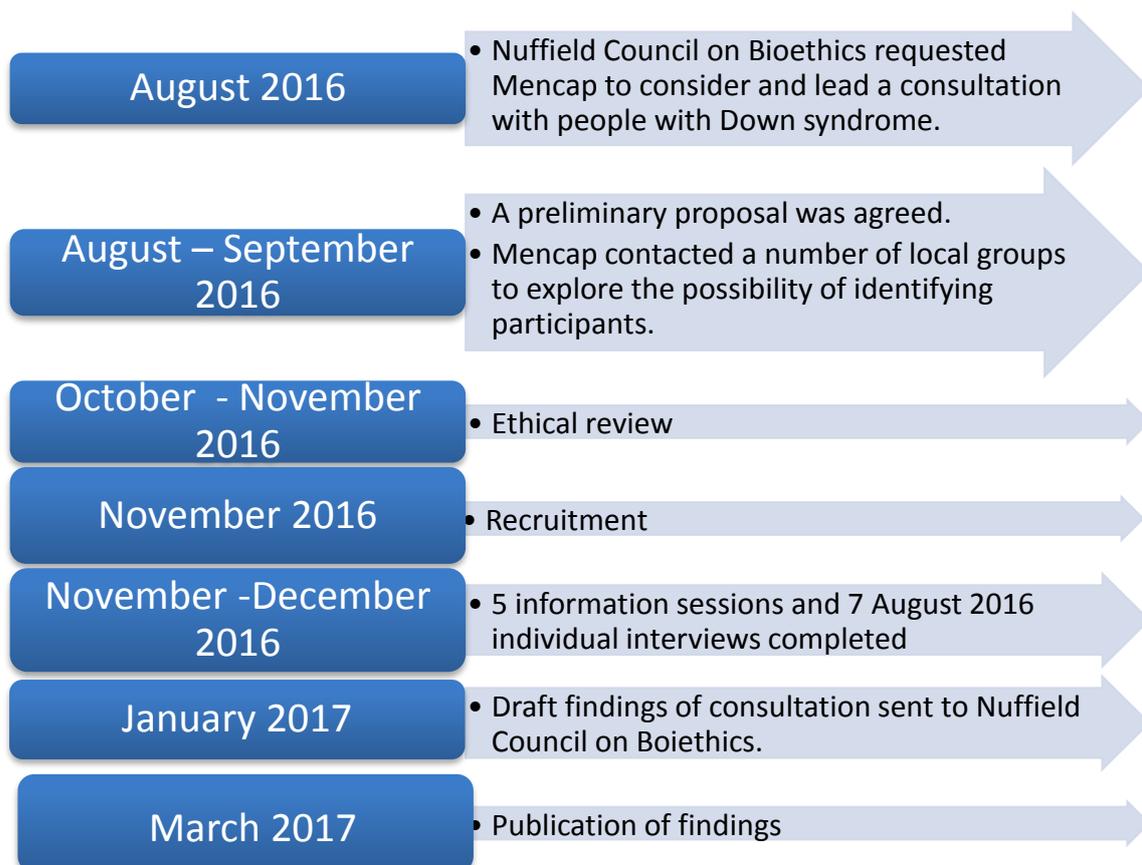
Braun and Clarke argue that one of the benefits of thematic analysis is its flexibility. Qualitative analytic methods can roughly be divided into two camps; those stemming from a particular theoretical or epistemological position such as conversational analysis, interpretative phenomenological analysis (IPA), grounded theory or narrative analysis, and secondly there are methods that are independent of theory and can be applied across a range of theoretical and epistemological approaches. Arguably, thematic analysis falls within the second category and as such its theoretical freedom provides a useful research or consultation tool which is compatible with essentialist and constructionist paradigms and can provide a rich and detailed, yet complex account of data. Braun and Clarke (2006) highlight that thematic analysis can be an essentialist or realist method, which reports experiences, meanings and reality of participants or it can be a constructionist method which examines the ways in which events, realities, meanings, experiences and so on are the effects of a range of different discourses operating within society. Thematic analysis can also represent a contextualist model sitting between realism and constructionism and characterised by theories of critical realism which place emphasis on the way individuals make meaning of their experience and also the way which broader social context influences those meanings, while retaining the focus on the limits of 'reality'. As such, thematic analysis can be a method used to both reflect and describe 'reality' while also deconstructing that reality.

2.2. Procedure

Summary of timeframes

The deadline of the Nuffield Council on Bioethics project meant that it was not possible to carry out a large scale consultation with individuals with learning disabilities. All too often,

individuals with learning disabilities are omitted from influencing policy which may directly affect their lives. As such, it was considered that a small scale consultation could be completed to ensure that the voice of individuals with Down syndrome was not entirely excluded. It was agreed that the consultation would inform the larger project as completed by the Nuffield Council on Bioethics. It was agreed that a separate report would be written to outline the consultation with individuals with Down syndrome, placing any findings reported by the Nuffield Council on Bioethics in context.



2.3. Ethical Review

The consultation proposal including the outline, design and resources were reviewed by the Nuffield Council on Bioethics Management and Risk Subgroup. Mencap also invited independent reviewers including academics from the field of learning disabilities and an individual with Down syndrome to review the proposal. Following some minor adjustments to the consultation proposal, both review panels concluded that the proposed consultation should go ahead and that the consultation team had appropriate plans to inform and protect the interests of the participants.

2.4. Recruitment

Members of the Royal Mencap Society led in recruiting participants to this consultation. Given the potential sensitivity of the topic, participants were recruited according to the following inclusion criteria: over the age of 18 years, self-identified as having Down syndrome, had the receptive and expressive communication ability to take part in an interview, had previous knowledge of sex and relationships, and had ongoing contact with the referring organisation who could ensure that follow up contact and ongoing support could be offered if necessary.

An initial scope for expressions of interest was outlined to local Mencap groups in August 2016. Following ethical review and approval in October 2016, formal recruitment ran from the end of October. Contact was made with a number of national groups which support individuals with Down syndrome including: Down Syndrome Association, Downs Syndrome Research Foundation, Don't Screen Us Out, Razed Roof, and a wider pool of local Mencap groups.

An invitation was posted on Mencap's social media channel. It is estimated that the online recruitment efforts reached a potential 65,000 people on Facebook and a potential 186,000 on Twitter. Potential participants were provided with an accessible information sheet outlining the aim and process of the consultation. When appropriate, a member of Mencap spoke with supporters, including family members or other supporters, to ensure that the invitation to participate was explained in a sensitive manner.

2.5. Information Session

Participants were invited to attend a voluntary information session about prenatal testing and all participants had the option to have a supporter such as a family member or carer attend. The aim of the information session was for the participants to have the opportunity to meet with the lead facilitator and members of Mencap, to have a discussion about the aims of the consultation, and for the project team to provide information about prenatal testing including NIPT. Despite the very tight timescales available, nine individuals with Down syndrome attended either the group or individual information sessions (4 individuals attended a group session and 5 individuals attended individual information sessions).

One individual who attended a group information session declined to take part in an interview. One individual who attended a group information session was not considered to have capacity to consent to the interview, and was not invited to take part. The materials for the information

session were based on the original materials previously used in the study by Barter et al. (2016). These materials had been co-designed with an individual with Down syndrome. The materials were updated for the purpose of this consultation. Table 1 outlines the topics covered in the information sessions including: sex and relationships, diversity, difference and disability, and testing for Down syndrome.

Table 1. Information topics for Information session

Sex and Relationships	Difference and Diversity	Prenatal Screening and termination
<ul style="list-style-type: none"> - A short review of sex and relationships 	<ul style="list-style-type: none"> - Discussion of difference. - Introduction to ‘genes’ and genetic difference (eye colour, height, right and left handed, etc). - Down syndrome as a genetic difference. 	<ul style="list-style-type: none"> - Discussion about prenatal screening - Introduction to the ‘new test’. - Discussion about termination.

The material was presented in verbal, written and pictorial formats and interactive activities were used to promote interaction and assimilation of information. The information sessions were led by Dr Barter and were supported by members of Mencap. The individuals were provided with a summary booklet at the end of the session in written and pictorial form. Each individual had access to a member of Mencap if they wished to discuss any aspect of the information session. A member of Mencap made contact with the individual in the week following the information session to ask if there were any follow up questions and to discuss anything that arose from the information session.

2.6 Individual Interview

During the follow up telephone conversation, individuals were invited to take part in an interview with the Dr Barter. Four interviews were carried out at a Mencap site and four others were carried out in a private space at a community centre. Prior to the interview, each participant met with Dr Barter who re-explained the purpose of the interview and the consultation along with the rights to withdraw and confidentiality. Accessible resources, along with the resources from the information session, were used to facilitate this conversation. The participant was considered to have capacity to consent to an interview if she or he could demonstrate that he/she understood: the aim of the consultation, confidentiality, the right to withdraw, that the session would be recorded, and that a report would be written and shared with the public. Participants were considered to have capacity to consent if they demonstrated

the ability to weigh up and communicate the decision to take part in the context of the information provided. The interviews addressed a number of areas and the interview topic guide is outlined in Table 2.

Table 2 – Topic Guide for Interview

Review information session	<ul style="list-style-type: none"> - Is there anything that we talked about when we last met that you would like to ask about? - When we met the last time we talked about screening for genetic difference (like Down syndrome). Do you remember what we said?
Familiarity with Down syndrome	<ul style="list-style-type: none"> - Do you know anyone with Down syndrome (or other genetic differences)?
Testing	<ul style="list-style-type: none"> - What would you say if someone wanted to have a test to find out if the foetus (the unborn baby) was going to have a genetic condition? Supplementary questions if appropriate – Do you think this is a good or bad idea? Can you think of any reasons why? How does it make you feel?
Decision following screening	<ul style="list-style-type: none"> - What would you say if a woman/couple/family wanted to continue with a pregnancy after a test? - What would you say if a woman/couple/family wanted to end a pregnancy if they knew that the foetus had Down syndrome or another genetic condition? Supplementary questions if appropriate - What are the good/bad things about this? What advice would you give a couple? What should they know about Down syndrome before making this decision? How should they be given this information?
Other	<ul style="list-style-type: none"> - Do you think screening will make a difference to your life? - Do you have any other things that you would like to say?
Supplementary question if appropriate	<ul style="list-style-type: none"> - Do you think a woman/couple/family should find out other things about the foetus (unborn baby)? e.g. – life limiting conditions, later life conditions or other characteristics.

The interview schedule was flexible and tailored to the individuals’ communication needs. At times, questions were re-worded or broken down, and at all times the lead facilitator asked open-ended questions. Participants had all of the resources presented at the information session in written and pictorial formats to support the interview. Participants had pens and paper which they could use throughout if they wished.

Interviews ranged from 30-60 mins and each participant completed one interview. Five of the six participants included in the consultation chose to be accompanied to the interview by a supporter or a member of Mencap. In three of these interviews, the supporter sat on the other side of a large room so were available if needed but were not part of the discussion. Following each interview, the lead facilitators ensured that the participant still consented to involvement in the consultation. Mencap followed up with each participant in the week following the interview to answer any follow up questions or concerns.

2.7. Participants

Seven individuals took part in an individual interview following the information sessions and the information from six interviews were used in this consultation exercise. Participants are described in Table 3 and pseudonyms have been used throughout.

Table 3. Participants

Pseudo name	Identified as	Age	Area of UK
Sarah	Female	32	Greater London Area
Mary	Female	27	Greater London Area
Dylan	Male	36	Greater London Area
Sophie	Female	21	West Midlands
Helen	Female	38	Greater Manchester Area
Sam	Male	28	Berkshire

One individual demonstrated capacity to consent to an interview on the basis of the pre-defined criteria to assess capacity to consent, but throughout the interview it became clear that he did not identify with the label “Down syndrome”. Given the public nature of the consultation, it was deemed unethical to include his data in this particular consultation.

Participants were given an accessible summary of the information gathered and an accessible version of the report. The participant whose information was not included in the consultation was provided with an accessible summary document which outlined the findings from the consultation, but this summary did not make direct reference to that person having Down syndrome (given that this person did not identify with the label).

2.8. Analysis

Dr Barter completed the process of thematic analysis following Braun and Clarke (2006) who highlight the importance of transparency with regard to the approach and method of analysis.

Analysis was driven by an analytic position which aimed to understand what the participants said and also how participants described their views. Themes were considered salient if they answered the main aims of the consultation. Semantic and latent themes were identified and combined throughout the analysis. That is, descriptions of what participants said and interpretations of these themes were considered. In this way, the analysis took a critical realist

stance by primarily describing what participants said and also adding interpretation to that content.

The analysis was completed in line with Braun and Clarke’s (2006) six-phase process. This included familiarisation with the data, and each transcript was transcribed verbatim. Each transcript was read a number of times, and initial notes were made about each transcript. These codes were then collated and organised into particular sub-themes, which were in turn reviewed, defined, and named. In turn, sub-themes were organised into themes. Following typical practice, the analysis was continually refined and edited throughout the process of writing the findings. Validity checks were completed on 50% of the data by the second author, Professor Richard Hastings, to ensure that the participants’ account was clearly represented in the themes and findings.

3. Consultation Findings

3.1 Primary Analysis – Summary of findings from consultation

Four themes and subthemes from this consultation were identified and are summarised in Table 4. The themes and subthemes are described in more detail in Appendix 1 to allow for transparency of the findings. The themes and subthemes are described and quotes are used as illustrations of those themes, but these are not an exhaustive list of quotes which made up the themes.

Table 4. Summary of themes and subthemes

Current Consultation	<p>Thematic Analysis (TA)</p> <p>What does Down syndrome mean?</p> <ul style="list-style-type: none"> - <i>Definition and understanding</i> - <i>Disability and difference</i> - <i>Self-Identity</i> - <i>Others’ perception of Down syndrome.</i> 	<p>TA</p> <p>A test for Down syndrome</p> <ul style="list-style-type: none"> - <i>Previous knowledge</i> - <i>Old and new test</i> - <i>View of testing</i> - <i>Decision following testing.</i> 	<p>TA</p> <p>What does test mean to me?</p> <ul style="list-style-type: none"> - <i>Personal impact</i> - <i>Impact on Down syndrome community.</i> 	<p>TA</p> <p>Balanced information</p> <ul style="list-style-type: none"> - <i>Balanced information and informed choice.</i>
----------------------	--	--	---	--

In summary, participants spoke of negative discourses about disability and a lack of understanding and fear of Down syndrome. This stood in stark contrast to their more positive life experience and the life possibilities for anyone born with Down syndrome or disability.

In general, participants agreed with testing as a means of preparation and as an opportunity for prospective parents to gain information. Participants' views of termination varied from strongly disagreeing to being unsure. Generally, participants were saddened and expressed disappointment with termination following testing. This sadness and disappointment could be understood in the context of their view of disability and their experience of living with Down syndrome. Although the majority of participants were saddened and disappointed by decisions to terminate following testing, they also respected women's right to choose.

The negative societal discourses surrounding disability, decisions to test, and decisions taken following testing may have a direct or indirect impact on individuals (such as those with Down syndrome) on an emotional and practical level.

There was a consistent and strong call from participants for balanced information, including the possibility to meet people with Down syndrome, as a means of supporting a more educated or informed choice for women and families.

2.3 Secondary Analysis – Triangulation of consultation findings with previous research findings

The core strength of data triangulation is the use of existing data for review and analysis. Rather than drawing conclusions from a single study data triangulation, by definition, uses multiple data sources to examine a situation. A larger pool of relevant data is likely to mean that areas of convergence and divergence will be discovered; areas of convergence and divergence that may not have been identified or noticed in the data from a single study. A parallel strength is the nature of the findings when they are drawn from multiple data sources and datasets (UNAIDS.org).

Tonkin Crine et al. (2016) outline that a triangulation protocol involves the integration of data when all datasets have already been analysed individually. Given the previous research by Barter et al. (2016) and the current consultation broadly addressed a similar question, it was considered that further depth could be offered by triangulating the data from both studies. Four main types of triangulation are identified in the literature including methodological, data, theoretical, and investigator. Barter et al. (2016) and the current consultation differed in time points, methodological, and theoretical approaches (see Table 5). As such, the current analysis drew on three types of triangulation: theoretical, data, and methodological.

Table 5. Summary of studies (Barter et al. 2016, & current consultation).

Current Consultation	Dataset	Theoretical Approach	Methodology
Barter et al (2016)	Data collected in 2010 from 8 individuals via individual interviews.	Interpretative phenomenological (IPA) analysis focussing on experience, and discourse analysis (DA) focussing on broader social discourse and adopting a social constructionist approach.	Exploratory interviews.
Current Consultation	Data collected in 2016 from 6 individuals via individual interviews.	Thematic analysis adopting a critical realist stance to the data.	Interviews with a particular focus on gathering views to inform a larger consultation.

Using these three types of triangulation incorporated a variety of approaches to the datasets, which strengthened the analysis by taking a more holistic approach to analysing data and evaluating the overall topic in question.

Given the differences in theoretical and, therefore, methodological approaches to each study, the resulting analysis yielded differing approaches to the data. Barter et al. approached the exploratory data from a phenomenological and social constructionist approach, which placed a particular emphasis on understanding the lived experience of participants and also the broad social discourses that shaped those experiences.

The current consultation was part of a larger consultation which aimed to gather the views of individuals about prenatal testing with a particular emphasis on a context consisting of changes being made to testing policy. As the consultation was interested in reporting participants' views, the theoretical approach of critical realism was adopted and themes with subthemes were identified.

Table 6. Summary of themes across studies (Barter et al. 2016, & current consultation).

Study	Key findings			
Barter et al (2016) (Study1)	Interpretative Phenomenological Analysis Devalued condition and a valued life <ul style="list-style-type: none"> - A valued life - Self beyond Down syndrome Discourse Analysis A discourse of social deviance		Interpretative Phenomenological Analysis A question of want <ul style="list-style-type: none"> - Down syndrome as wanted or unwanted for self and others. - Down syndrome as unwanted because of lack of awareness. - Self as wanted. Discourse Analysis A discourse of tragedy and catastrophe	
Current Consultation (Study 2)	Thematic Analysis What does Down syndrome mean? <ul style="list-style-type: none"> - Definition and understanding - Disability and difference - Self-Identity - Others' perception of Down syndrome. 	Thematic Analysis A test for Down syndrome <ul style="list-style-type: none"> - Previous knowledge - Old and new test - View of testing - Decision following testing. 	Thematic Analysis What does test mean to me? <ul style="list-style-type: none"> - Personal impact - Impact on Down syndrome community. 	Thematic Analysis Balanced information <ul style="list-style-type: none"> - Balanced information and informed choice.

Triangulation of both studies includes highlighting the converging and diverging findings as well as highlighting findings that are unique to either study. As the main differences in the studies are the theoretical and the associated methodological approaches used, the advantage of triangulation is to highlight the similar and different key findings from differing epistemological positions.

Barter et al. (2016) highlighted two key findings which included interpretative phenomenological analysis (IPA)-derived themes with subthemes and associated discourses, and the following highlights the convergence of themes with the current study (see Table 7).

Table 7 Triangulation of themes across studies (Barter et al. 2016, and current consultation).

Barter et al (2016) across and current consultation down	Discourse of social deviance		Discourse of tragedy and catastrophe		
	A valued life	Self beyond Down syndrome	Down syndrome as wanted or unwanted for self and others.	Down syndrome as unwanted because of lack of awareness.	Self as wanted.
Definition and understanding	X				
Disability and difference	X				
Self-Identity	X	X			
Others' perception of Down syndrome.				X	
Previous knowledge					
Old and new test					
Decision following testing.			X	X	
Personal impact			X		X
Impact on Down syndrome community					
Balanced information and informed choice.				X	

X- Areas of convergence

3.3 Convergence of themes across studies

The first of the Barter et al (2016) IPA-derived themes ‘a devalued condition and a valued life’ bears a number of similarities to the subthemes generated in the current consultation ‘what does Down syndrome mean?’ In particular, the first three subthemes map onto the current consultation findings: definition of Down syndrome, difference and disability, and self-identity. Although there was a difference in methodological approach, there is clear convergence in terms of both the views and experiences of the participants across both studies. In both studies, participants made reference to their understanding and experience of Down syndrome, their descriptions of disability and difference, and their own sense of self, achievement, and personal value beyond disability. Participants also extended these perspectives to possibilities for any person with Down syndrome, not just themselves.

Barter et al.

Becky - ' keep it, I mean and you know, you know, he or she might gain experience and, um and emmm gain everything in life in general, you know, yes'.

Current Consultation

Helen – 'We're still good people. Everyone's got good and bad in them. We just live life to the fullest as much as we can. And we learn like everyone else but we take longer to get to the achievements.'

Barter et al. identified a discourse of social deviance which underpinned the theme of a 'devalued condition and a valued life'. The social deviance theory of disability mirrors the medical view of normality and pathology as a means of understanding impairment. As deviance is a phenomenon situated in power, the 'deviant' group are labelled as different, less valuable, and powerless. In both studies participants used language of chronicity and difference to describe their experience and view of Down syndrome and disability.

Current Consultation

Ben – ' It means you have to live with it... Yeah, in your life, really... it means, like, you have to die with your own disability too... You can't change it.'

In both studies, there was also an alternative discourse that emerged which challenged this dominant discourse of disability.

Barter et al

Becky - 'Down syndrome, so what?'

Current Consultation

Sophie – 'Am I a burden? Am I a risk? No, I'm a person with feelings... Because I have a friend... she may need more support but she is still precious. That's what I am trying to get across... people with Down syndrome should be loved for who they are, not what they achieve.'

Barter et al identified the theme 'A question of want' which constituted three subthemes: 'Down syndrome as something that is wanted by the self or others', 'Down syndrome as unwanted because of lack of knowledge', and the 'self as wanted'. As the Barter et al study focussed on participant's experiences, these themes place an emphasis on the personal experiences that participants discussed. However, there is convergence between these themes and a number of the themes outlined in the current consultation. There is clear convergence for the themes of Down syndrome as 'not wanted' by others across both studies.

Barter et al.

Becky – 'as soon as the doctor or someone says there's complications around your baby... some people kind of take the oh my god I don't want that baby'.

Current Consultation

Mary – 'It's like if a woman wants a baby with Down syndrome or not'.

There is also clear convergence across both studies that participants viewed peoples' lack of knowledge or misunderstanding as central to 'not wanting'.

Barter et al.

Sam – 'I think it's to do with the family cos they don't know the Down syndom'.

Current consultation

Sarah – I don't think people really understand what Down syndrome is, really... and it's quite upsetting about that.

Across both studies, it is clear that prenatal testing has a personal impact on participants. In Barter et al. this was themed as the experience of the 'self as wanted' and in the current consultation was themed as the impact of testing on the self and outlined the emotional impact on the individuals.

Barter et al

Becky – '...my Mum had me and still wants to have me and still loves me.'

Current Consultation

*Sophie – Because it makes me feel like I'm not wanted in society. And no one loves us...
(Speaks about how her family loves her)*

But the government doesn't love me, because if they did love me they wouldn't do this.

In both studies, participants referred to a test as a means of preparation for a baby with Down syndrome. Barter et al. highlight two participants who refer to the 'shock' of having a baby with Down syndrome and, in spite of their personal views, both used language of understanding to identify with a woman choosing to terminate.

Barter et al.

Becky – '... kind of positive stories about, um, how mothers are having these tests and accepting the babies themselves'.

Current Consultation

Dylan – 'I think it's a good thing that they know. ... know what they are expecting. They know exactly what to do. How to treat the baby... They might not know, parents might now know what it'll be like.'

Across both studies, there was a general agreement and convergence that, in spite of personal preferences, views or experiences, decisions following testing are a woman's choice.

Barter et al.

Luke - That's not up to me to say really... I don't know really, I just don't know, it all depends on the lady actually.

Jen – 'it's her body and her life, yes... it's her choice... it's good that a Mum can decide'.

Current Consultation

Helen – 'They are getting rid of it if they're finding out it's Down syndrome because they don't want that life for their baby or something. I mean it's a personal choice. I'm a pro-choice person. I believe what the woman wants she should get. If she doesn't want a baby with Downs syndrome she doesn't have to... It's a woman's rights.'

3.4 Differences between Barter et al (2016) and the current consultation findings

There were a number of differences across the two studies either in terms of the presence or absences of certain themes, or the emphasis on certain themes.

Barter et al (2016) identified others' view of Down syndrome and disability as underpinned by a negative discourse of social deviance and also identified that others had little understanding of Down syndrome. In the current consultation there was a stronger view from participants that other people were scared of disability which caused them to avoid or 'run away' from it. The emphasis on fear of disability was not identified to such an extent by Barter et al (2016).

Current consultation

Helen – 'The first thing people think of Down syndrome is it's sad and that it's scary. They don't know much about it.'

Sophie – 'And that's what I am trying to get across to the world. What's so threatening about Down syndrome?'

Previous knowledge of testing, and understanding of the old and new test were unique themes to this consultation. New testing was of primary interest in this consultation, and as such there was some emphasis on it during the information session which may have influenced the discussions at interview.

Current consultation

Sophie – 'I understand that it's 90% accurate and it can detect Down syndrome in the womb. It's different because it's more accurate. It can detect at an early stage. It (the old test)... didn't do good to the foetus.'

Helen – 'There's this new test called NIPT test that's non-invasive. That means that it doesn't harm the baby at all. It's a blood test through the arm, like, if you can get a normal blood test. But they can find out if your baby is Down syndrome through that.'

Although participants called for balanced information to be provided across both studies, this was much more emphatic in the current consultation. As the current consultation was interested

in understanding the views of participants toward prenatal testing, it is likely that the interviews placed more emphasis on eliciting views about testing, and the implications of this.

Current consultation

Helen – ‘Educate them. Mostly online at the moment... you can find out about many different disabilities... Educate themselves. They can research it... So they can have a more informed choice of whether or not to keep the baby. I think it’s good to get more educated choice... but I think they should have more information for them’.

Although there was a convergence of themes surrounding the importance of women’s right to decisions following a test regardless of personal view, there was again more emphasis in the current consultation on participants’ view of testing. This can also be understood in terms of the purpose of the consultation, and the emphasis on eliciting views and perspectives.

Current consultation

Dylan – ‘That I guess I would be shocked... Because I think you would love the child...’

Sophie – ‘I think it’s a very sad choice that people are terminating.’

Barter et al. highlighted a theme of ‘the self as wanted’ which was not as apparent in the current consultation. Again, this could be interpreted in terms of the purpose and methodology of the studies. Barter et al (2016) were interested in exploring personal experience and the meaning of testing for participants, there was a more personal tone to the themes, and more emphasis placed on understanding the personal experience throughout the analysis.

Barter et al.

Jen – ‘Mum had a test for me and her, they both decided to keep me... (Puts her thumbs up)...’

Barter et al. also highlighted a discourse of tragedy and catastrophe drawing attention to the narrative that the birth of a child with a disability is a tragedy in personal terms and a catastrophe in public terms. It was interpreted that the decision to test and terminate can serve to re-establish social order if this discourse is accepted. A discourse analytic approach was not adopted in the current consultation and from the findings it is not clear that such a discourse

was present. However, there was one reference to a recent documentary film which directly challenged this discourse in study 2

Sophie – ‘You could look at the good bits. How much love and humour they can bring to the family. Because Sally Philips Said it’s a comedy, not a tragedy.’

4. Discussion

4.1. Summary of findings

Down syndrome was identified as a devalued condition by others and society at large across Barter et al (2016) and the current consultation, regardless of methodology and theoretical approach. It was clear that across both studies, participants experienced a negative discourse about disability. The current consultation also highlighted that participants viewed Down syndrome as something that caused sadness and fear for others. Participants across both studies described their own life experience and view of what is valued and valuable in terms of love, achievement, and their own sense of self which stood in stark contrast to the wider more negative discourse. Participants in both studies linked others’ lack of understanding of Down syndrome and, in the current consultation, the fear of disability to the decisions to test and decisions made following testing.

The majority of participants in both studies supported women’s right to choose, regardless of their personal view about testing and termination following testing. Across both studies, but more emphatically in the current consultation there was a consistent call for balanced information to be offered to women or families making decisions. Barter et al. placed a greater emphasis on understanding the personal experiences and wider societal discourses surrounding the topic of prenatal testing. Although it was clear from both studies that testing has an impact on the participants, Barter et al. placed a clearer emphasis on considering the ‘self as wanted’.

4.2 Accessible Discussion

Overall, both studies have explored prenatal testing for Down syndrome with people with Down syndrome and have demonstrated that the conversations were accessible and that the participants in both studies could offer a valuable contribution to the current consultation. Given the sensitive nature of the topic, we recruited individuals very carefully, had clear inclusion criteria, approached the topics sensitively, and had support in place throughout. A

number of the organisations contacted felt unable to support this piece of work as there were concerns about the impact on individuals. This is an understandable concern from organisations whose primary concern is to support and advocate for individuals with disabilities. However, it is worth noting that, in a number of cases, participants also expressed a sense of satisfaction and value for having the opportunity to express their views.

It is imperative that we continue to include individuals with disabilities in research and policymaking, and every effort should be made to support this in a meaningful way. Given that NIPT is proposed to be rolled out from 2018/2019, it is crucial that individuals Down syndrome and other disabilities have a meaningful role in considering the social and ethical concerns.

4.3 Are people with Down syndrome aware of prenatal testing?

There may be an assumption that prenatal testing and subsequent termination on the basis of disability goes unnoticed by people with learning disabilities. As described the participants in our studies met a number of inclusion criteria and as such are not representative of all people with Down syndrome. However, it was clear during both studies that some participants were previously aware of the topics. Many individuals made reference to TV programmes and to the recent BBC documentary by Sally Phillips, which in some cases led to discussions within their family or support network. Some participants had some knowledge prior to taking part in the consultation and valued the information session as a way to understand the issues more clearly. Therefore, we must not assume that people with Down syndrome and learning disabilities are unaware of prenatal testing. It is also important to be aware that individuals with disabilities may not have access to accessible clear information which clearly explains genetic testing.

4.4 What is informed Choice?

There are on-going debates about the level of informed choice that women have about the uptake of a prenatal test. Although there is a consensus about the importance of informed choice, there are few definitions and measures of this construct (Marteau and Dormandy, 2001). There are two core characteristics of an informed choice: for it to be based on high quality relevant information and that it reflects the decision maker's values. Marteau and Dormandy (2001) highlight that to make a decision about prenatal testing; women and families should have information about the condition, and understand the characteristics and implications of the test. Van den Berg, Timmermans, Kleinveld, et al. (2005) studied 1,159

pregnant women's extent of informed choice about prenatal testing and the psychological effects of informed decision-making. They demonstrated that 83% of women were deemed to have enough knowledge about prenatal testing, 82% made a value-consistent decision, and in total 68% made an informed choice. Van den Berg et al. argued that although the rate of informed choice was high, there were a large number of women who made uninformed choices due to insufficient information, value inconsistency, or both. They also pointed out that informed choices appeared to be psychologically beneficial, as informed choice was associated with higher satisfaction with the decision made.

4.5 What is balanced information?

Disability Rights authors suggest that prenatal genetic counselling is driven by misinformation because it is based on a failure to understand what life is like for disabled individuals and their families (Raz, 2003). Hastings (2016) concluded, through a critical review of the literature, that despite potential stresses, parents of children with disabilities report significant positive outcomes and experiences. Mateau and Johnston (1986) demonstrated that those with a disability perceived the conditions as less serious than those without a disability.

Parens and Asch (2003) point out that despite evidence, positive information about childhood disability and family satisfaction are rarely discussed with prospective parents. Information provided by professionals about genetic conditions tends to be brief (Murray, Chuckle, Sehmi, et al., 2001) and negative (Asch, 1999). Sooben (2010) carried out a phenomenological study to understand the experience of parents during antenatal testing and the subsequent birth of their baby. They outlined that after birth children's 'differentness' rather than health needs were the main focus of care interventions and concluded that professionals in maternity care must be equipped with appropriate knowledge of Down syndrome, and must include a 'balanced view' of the future of the child consistent with the principle of reproductive autonomy.

Participants across both of our studies called for information to be provided to women and families to ensure that they understand what Down syndrome means and what these individuals' experience of, and perspective on, life. Participants had some initial ideas about the type and format of information which they saw as useful for prospective parents, such as: meeting people with Down syndrome, meeting family members, and understanding the value of the lives of people with Down syndrome.

Skotko et al. (2011) surveyed 284 people with Down syndrome in the US about the self-perceptions, including advice to parents of children with Down syndrome. They suggested that this information collected could be used in prenatal counselling sessions. There is currently no such large scale UK based study assessing the self-perceptions of individuals with Down syndrome and a study of that nature may go some way to include the voice of individuals with Down syndrome.

Previous research suggested that presenting positive information about a condition may have little effect on the choices that women and families make. For example, Figueiras, Price and Marteau (1999) conducted an analogue study comparing the effects of various forms of information about children with Down syndrome to 814 undergraduate students. There was no reported difference between the presentations of positive and negative information, either in text or pictorial format, and all groups, including a control group, reported a high level of concern about having a child with Down syndrome.

As such further research is required to understand 1) what type of information should be presented to prospective parents and 2) is information at the time of testing likely to challenge negative discourses so strongly identified by participants?

4.6 A message to people with Down syndrome?

Parsons and Asch (2003) argue that general societal perceptions of disability are negative and discriminatory and are based on medical and functional models rather than on the perception of disability as a socially constructed phenomenon. Lippman (1991) coined the term 'geneticization' to emphasise the dominant discourse in contemporary society surrounding the stories that are told about disease and disability. Lippman argues that the discourse around genetic testing is based on 'story telling' of themes such as choice, reassurance, and control, arguing that dominant discourse suggests that a child born with a disability represents some type of failure.

The argument that prenatal testing followed by selective abortion expresses discriminatory attitudes toward the disabling trait, and also those who carry, it has been called the expressivist argument (Buchanan, 1996). Asch (2000) argues that prenatal testing sends out a hurtful message to those with specific traits, and he argues that this message is: 'As with discrimination in general, with prenatal diagnosis, a single trait stands in for the whole, a single trait obliterates the whole' (Parsons & Asch, 2003 p.42). Clarke (1997) points out that prenatal testing, which

aim to reduce suffering, may inadvertently increase suffering through promoting stigmatisation and intolerance which is a major source of difficulty to affected individuals and families.

There is disagreement with the expressivist argument on several counts. Kittay (2000) argues that women and families choose to take up prenatal testing for a number of reasons, and it is impossible to conclude exactly what ‘message’ is being sent. Arguably, prospective parents do not choose to undergo prenatal testing with a wish to hurt disabled people, but rather to consider their own family life. In this way, Kittay argues, there is no broader message at all. A second criticism of the expressivist argument is that it calls into question the morality of nearly all abortion as it assumes that we can morally distinguish between aborting ‘any’ foetus and a ‘particular’ foetus.

Regardless of the intent or rationale behind decisions to test and terminate, both of our studies have outlined that the policy and practice sends a hurtful message to those with Down syndrome which further reinforces the lack of value that individuals experience. Study 1 and to a lesser extent, Study 2 also highlighted experiences of participants feeling unwanted and unloved. This message needs to be considered very carefully in current policy making both in terms of discriminatory messages, and the impact on individuals with any disability.

5. Limitations

The primary limitation of this consultation was the very short timescale available to meaningfully involve individuals with Down syndrome. Given that it was not possible to extend the deadline for the completion of the work, it was considered that some participation, conducted in an ethical and sensitive manner would be more beneficial than excluding the voice of individuals with Down syndrome entirely.

A tight timeline left little time to recruit participants and response rates were very low. A number of organisations, although interested in the issues, were not able to recommend people to take part in the consultation work. Reasons included a fear about the potential negative impact on the participants and a lack of their availability during the consultation period to commit to the study. A number of local groups we followed up also expressed concerns that there would not be enough time for them to work with (i.e., prepare) people before they could participate in the consultation. Almost all organisations suggested that the time frames within which we were working were, to a greater or lesser extent, prohibitive of their involvement.

The participants recruited to this consultation, and to the Barter et al. (2016) study, all met the strict inclusion criteria used to ensure that the study was conducted ethically. The small number of participants made a valuable contribution to the larger consultation about NIPT, but naturally these views and themes are not presentative of many people with Down syndrome who could not, or were not able to, take part. There may also have been a selection bias in terms of the individuals recruited. Participants who took part may have had more interest in the topic and may have clearer or well-formed views than other individuals with Down syndrome who were not reached during the consultation.

Author's Conclusions and Recommendations

1. Two small scale studies, Barter et al. and the current consultation have been carried out with individuals with Down syndrome who met a strict set of inclusion criteria. Due to the small number of participants, and potential selection bias, it is clear that the findings are unlikely to be representative of many people with Down syndrome. This initial work has demonstrated that it is both feasible and valuable to gain the perspectives of individuals with Down syndrome. Further larger scale consultation is required to continue to build on this initial work to include perspectives from larger numbers of people, and those with potentially differing views and experiences.
2. In spite of these clear limitations, a number of key themes have been identified which have been shared with the Nuffield Council on Bioethics. This has ensured that, at the very least, the voices of a small number of individuals with Down syndrome have been included in the current consultation.
3. Based on the results in the current report, it was evident that a number of the participants with Down syndrome were aware of prenatal screening prior to their involvement in either study. It should be assumed that individuals with Down syndrome may be aware of the issues around genetic testing, and may wish to have clear information about the topic.
4. Directly seeking the views and experiences of people with disabilities about prenatal screening is valuable and necessary. The present findings of the current consultation and Barter et al. (2016) also emphasise the importance of understanding the narratives that people with disabilities have about their own lives – as the background to their perspectives about NIPT. Further research is required to understand the diversity of living with any genetic variation. For example, a UK replication of a study such as

Skotko et al.'s (2011) would go some way to provide clear information about how individuals with Down syndrome or other disabilities view and experience life.

5. Across both studies, participants highlighted a negative societal view of disability which they described in terms of lack of understanding and fear; standing in stark contrast to their own more positive life experiences. Participants called for people to understand more about Down syndrome and the value of the lives of individuals with Down syndrome. Participants suggested that potential parents in particular should have opportunities to learn about Down syndrome, meet people with Down syndrome and their families, and hear positive stories about disability.
6. Given the perceived strength of the negative discourse surrounding Down syndrome, it is important to consider how and in what way information is provided to ensure informed choice in relation to NIPT.
7. Individuals in both studies perceived a personal message to them from testing policy. There is a clear need on the part of policy makers to take steps to mitigate the potential for the implementation and presentation of NIPT to be perceived by people with Down syndrome as questioning the value of their lives and themselves as 'wanted'.
8. Overall, we found that it is possible to include individuals with learning disabilities in discussions about sensitive and complex topics. However, this must be carried out in a sensitive manner, with preparation and support available, and adequate time must be allowed for meaningful participation.
9. A number of participants gave feedback to the supporting organisations that helped to recruit them to say that they felt valued through inclusion in this consultation. This is a further reason to ensure that people with Down syndrome and other disabilities are not excluded from further discussion about NIPT and similar issues.
10. It is evident that the differing focus of Barter et al. and the current consultation yielded some different findings as well as several similarities. One learning point from these observed differences is that the perspectives and experiences of people with Down syndrome are sensitive to context. As the technology, reach, and implementation of NIPT changes over time, it will be crucial to continue to monitor the experiences and perspectives of people with Down syndrome. It is recommended that ongoing consultation with people with Down syndrome is carried out because it will be both valuable and necessary in any potential roll-out of NIPT.

References

- Alderson, P. (2001). Down's syndrome; cost quality and value of life. *Social Science and Medicine*, 53, 627-638.
- Asch, A. (1999). Prenatal diagnosis and selective abortion. *American Journal of Public Health*, 89 (11), 1649 – 1657.
- Asch, A. (2000). Why I haven't changed my mind about prenatal diagnosis In Parens, E., Asch, A. editors. *Prenatal Testing and Disability Rights*. Washington DC: Georgetown University Press, 234-258.
- Barter, B., Hastings, Richard P., Williams, R. and Huws, J. (2016) Perceptions and discourses relating to genetic testing: interviews with people with Down syndrome. *Journal of Applied Research in Intellectual Disabilities*. ISSN 1468-3148 (In Press).
- Balakrishnan, T.R. and Chen, J. (1990), Religiosity, nuptiality and reproduction in Canada. *Canadian Review of Sociology*, 27: 316–340.
- Bell, M. and Stoneman, Z. (2000). Reactions to Prenatal Testing: Reflection of Religiosity and Attitudes Toward Abortion and People With Disabilities, *American Journal on Mental Retardation* 2000 105:1, 1-13
- Braun, V and Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology*, 3 (2). PP 77-101.
- Buchanan (1996) In Parens, E. & Asch, A (2003) Disability Rights Critique of prenatal genetic testing: Reflections and Recommendations. *Mental Retardation and Developmental Disabilities*, 9, 40-47.
- Clarke, A. (1997) Prenatal genetic screening: Paradigms and perspectives. In P. Harper & A. Clarke (Eds.), *Genetics, society and clinical practice* (pp. 119-140). Oxford: Bios.
- Crane E, & Morris J., K. (2006). Changes in maternal age in England and Wales -implications for Down syndrome. *Down syndrome Research and Practice*. 10, 41-43.
- Department of Health, 2016. (<https://www.gov.uk/government/news/safer-screening-test-for-pregnant-women>) retrieved 20 January 2017.
- Figuerias, M., Price, H., Marteau, & T. M. (1999) Effects of textual and pictorial information upon perceptions of Down syndrome: An analogue study. *Psychology and Health*, 14, 761-771.

- Green J.M., Snowdon C. & Statham, H. (1993). Pregnant women's attitudes to abortion and prenatal screening, *Journal of Reproductive and Infant Psychology*, 11, pp. 3-39.
- Hastings, R. P. (2016). Do children with intellectual and developmental disabilities have a negative impact on other family members? The case for rejecting a negative narrative. *International Review of Research in Developmental Disabilities*, 50, 165-194.
- Hewison, J., Green, J. M., Ahmed, S., Cuckle, H. S., Hirst, J., Hucknall, C., et al. (2007). Attitudes to prenatal testing and termination of pregnancy for fetal abnormality: a comparison of white and Pakistani women in the UK. *Prenatal Diagnosis*, 27, 419–430.
- Knoppers, B. M. and Chadwick, R. (2005). Human genetic research: emerging trends in ethics. *Nature Review Genetics*, 6, 75-79.
- Kittay, E. F. & Kittay, L. (2000). On the expressive and ethics of selective abortion for disability: conversations with my son. In Parens, E., Asch, A. editors. *Prenatal Testing and Disability Rights*. Washington DC: Georgetown University Press, 165-195.
- Krishnan, V. (1991). Abortion in Canada: Religious and ideological dimensions of women's attitudes. *Social Biology*, 38, pp. 249-257.
- Lippman, A. (1991). Prenatal Genetic Testing and Screening: Constructing Needs and Reinforcing Inequalities. *American Journal of Law and Medicine*, 15, 1&2, pp. 15-50.
- Marteau, T. M., & Dormandy, E. (2001) Facilitating informed choice in prenatal testing. How well are we doing? *American Journal of Medical Genetics*, 106, 3, 185-190.
- Marteau, T., & Johnston, M. (1986) Determinants of beliefs about illness: a study of parents of children with diabetes, asthma, epilepsy and no chronic illness. *Journal of Psychosomatic Research*, 30, 673-683.
- Morris, J. and Springett, A. (2014). The National Down syndrome Cytogenic Register for England and Wales. 2012 Annual Report. 2014.
- Murray, J., Chuckle, H., Sehmi, I., Wilson, C., & Ellis, A. (2001) Quality of written information used in Down syndrome screening. *Prenatal Diagnosis*, 2, 138-142.
- Nuffield Council on Bioethics (2017) *Non-invasive prenatal testing: ethical issues* (London: Nuffield Council on Bioethics).
- Parens, E. & Asch, A. (2000). *Prenatal testing and disability rights*. Georgetown University Press, Washington.
- Parens, E. & Asch, A (2003) Disability Rights Critique of prenatal genetic testing: Reflections and Recommendations. *Mental Retardation and Developmental Disabilities*, 9, 40-47.

- Ravitsky, R. (2015). Non-invasive prenatal testing (NIPT). Identifying key clinical, ethical, social, legal and policy issues. Nuffield Council on Bioethics. Retrieved from (<http://nuffieldbioethics.org/wp-content/uploads/NIPT-background-paper-8-Nov-2015-FINAL.pdf>)
- Raz, A. (2003). "Important to test, important to support": attitudes toward disability rights and prenatal diagnosis among leaders of support groups for genetic disorders in Israel. *Social Science and Medicine*. 59, 1857-1866.
- Shakespeare, T. (2006). *Disability Rights and Wrongs*. Routledge, UK.
- Skotko, B. G., Levine, S. P. and Goldstein, R. (2011). Self-perceptions from People with Down syndrome. *American Journal of Medical Genetics*, 0 (10); 2360- 2369.
- SOFT UK (2016) *Help for families*. Retrieved from: <http://www.soft.org.uk/Help-for-Families>
- Sooben, R. D. 2010 Antenatal testing and the subsequent birth of a child with Down syndrome: a phenomenological study of parents' experiences. *Journal of Intellectual Disabilities*. 14, 2, p. 79-94 16 p.
- Taylor-Phillips, S. Freeman, K., Geppert, J., Agbebiyi, A., Uthman, O.A., Madan, J., Clarke, A., Quenby, S., Clarke, A. (2016). Accuracy of non-invasive prenatal testing using cell-free DNA for detection of Down, Edwards and Patau syndromes: a systematic review and meta-analysis. *Obstetrics and Gynecology Research*, Vol 6, I.
- Tonkin Crine, S., Anthierens, S., Hood, K., Yardley, L., Cals, J. W. L., Francis, N. A., Coenen, S., van der Velden, A. W., Godycki-Cwirko, M., Llor, C., Butler, C.C., Verheij, T.,J., M., Goosens, H., Little, P. (2016). Discrepancies between qualitative and quantitative evaluation of randomised controlled trial results: achieving clarity through mixed methods triangulation. *Implementation Science*; 11, 66.
- UK Human Genetics Commission. (2005). *Choosing the Future: Genetics and Reproductive Decision-Making. Analysis of Responses to the Consultation*. Retrieved from www.poeplescienceandpolicy.com.
- UK National Screening Committee (2016). Retrieved 14 January from <https://legacyscreening.phe.org.uk/fetalanomalies>.

UNAIDS, An introduction to triangulation. Retrieved from
(http://www.unaids.org/sites/default/files/sub_landing/files/10_4-Intro-to-triangulation-MEF.pdf)

van den Berg M, Timmermans D., R., M., Kleinveld, L.P, *et al.* Are pregnant women making informed choices about prenatal screening? *Genet Med* 2005;7:332–8.

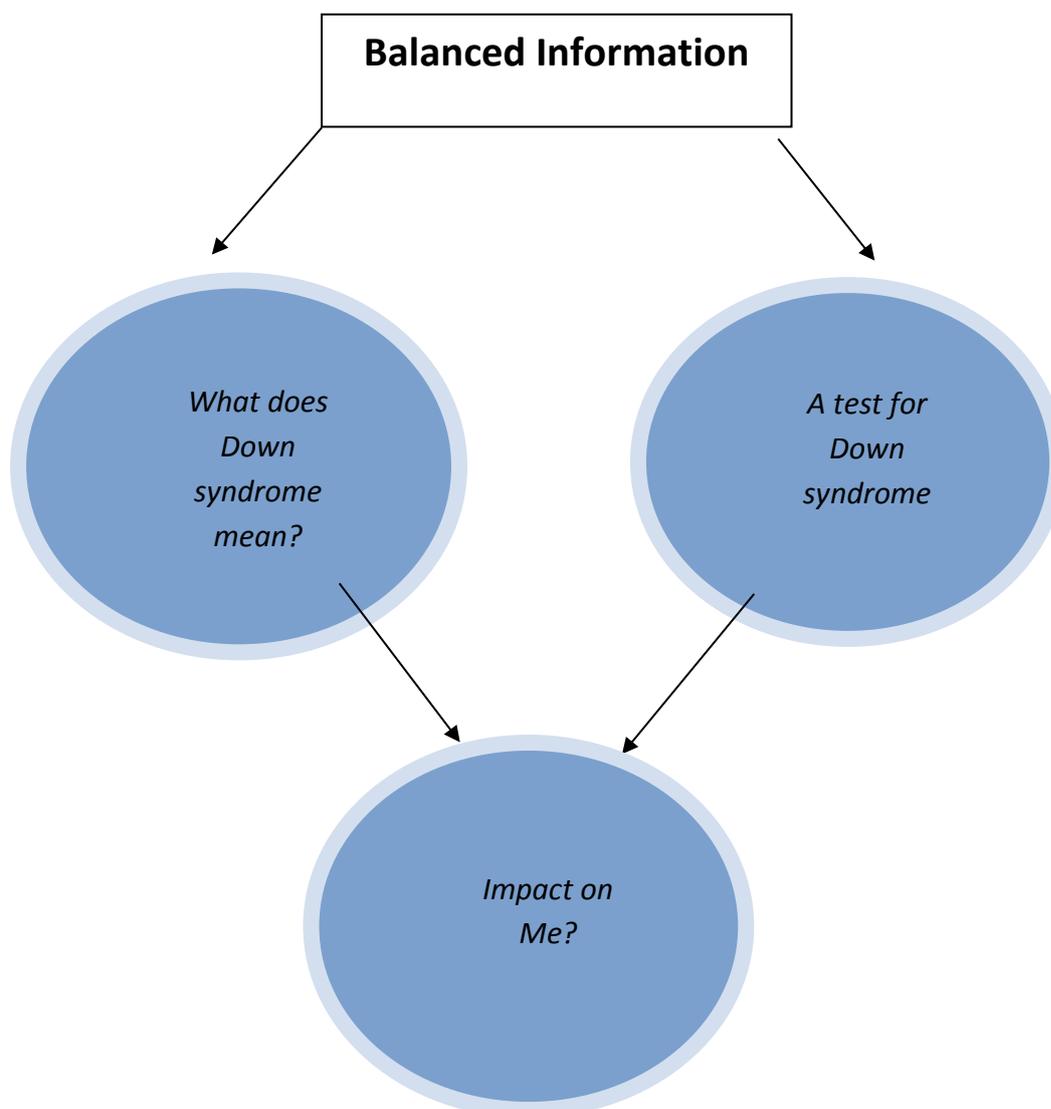
Ward, L., Howarth, J, & Rodgers, J. (2002). Difference and choice: exploring prenatal testing and the use of genetic information with people with learning difficulties. *British Journal of Learning Disabilities*, 30, 50-55.

World Health Organization (2016) *Genes and human disease*. Retrieved from:
<http://www.who.int/genomics/public/geneticdiseases/en/index1.htm>.

Appendix 1 - Detailed Findings from Consultation

Following thematic analysis four themes including subthemes were identified; i) What does Down syndrome mean, ii) A test for Down syndrome, iii) Impact on me? and iv) Balanced information. The themes and subthemes along with the relationships between themes are outlined below including a summary discussion. It is important to note that the quotes are merely used as examples and should be read as supporting information. A number of quotes for each theme and subtheme are provided to offer a good level of transparency to the reader. Pseudonyms have been used throughout and all identifying information has been removed. When providing extracts from interviews the following transcript conventions are used:

... . Short pause. (...) Words omitted to shorten a quote. (Text) provides contextual informatio



Theme 1 – What Does Down syndrome mean?

Participants used language of difference or deviance to discuss disability. There was an emphasis on medical terminology to describe Down syndrome and reference to various types, levels of severity, associated health difficulties, physical characteristics and need for support. Down syndrome or learning disability was described as something fixed, unchanging and lifelong.

Participants outlined a positive view of the self alongside, in spite of or beyond disability. Participants spoke proudly of achievements, independence, happiness and love. Participants challenged a negative view of disability by referring to people as more than achievement and raised a question of what is valued.

There is a stark contrast between the participants' experience of Down syndrome and their view of others' perception of Down syndrome or disability. They outlined that others do not understand Down syndrome, make assumptions about disability including lack of ability, causing a burden or posing a risk.

i) Definition and Understanding

A number of participants described their understanding of Down syndrome in literal terms. They described a chromosomal definition of Down syndrome.

Helen - It's not a disease. It's a chromosomal disorder... It affects the chromosomes, the cells in the body.

Dylan – It's something inside.

Sarah - It's to do with chromosome.... 21 chromosomes.

Participants also described the types of Down syndrome.

Helen – ... Down syndrome, Mosaic Down syndrome, Translocation Down syndrome, Mosaic Translocation Down syndrome, there's many sorts of it, like it's under an umbrella.

Mary paid made reference to the perceived physical characteristics of Down syndrome.

Mary – Like people are short with Downs...

Participants spoke about associated physical health difficulties.

Helen – Some people could have heart defects with it... and their speech. And some people may not be able to walk as many as others and stuff like that.

ii) Disability and Difference

Participants described Down syndrome and associated learning disability in terms of ability, or disability. There was an emphasis on the use of language of difference.

Ben – Down syndrome. It's a kind of disability.

It was described as lifelong, fixed or unchanging.

Mary – Babies are born with Down syndrome.

Ben - It means you have to live with it... Yeah, in your life, really... it means, like, you have to die with your own disability too... You can't change it.

They also described levels of ability or disability and the role of support as essential.

Helen – I think it's just a developmental thing where you're slower in learning.... And sometimes it's more or less severe in different forms.

Dylan – (in response to 'what does Down syndrome mean?') – They might act differently. We're all clever... But in different ways... Some people need more help.

Participants used language of difference to describe Down syndrome.

Ben – It means, like a normal baby has got different chromosomes.

iii) Self Identity

All participants self-identified with Down syndrome.

Mary – I know I've got Down syndrome.

Dylan – I know what it's all about, having a disability.

There was an emphasis on individual differences between people with Down syndrome.

Ben – Some people are not the same Downies, really.

Dylan – (with reference to a community group) I think I learnt a lot about each different person with a disability.

Helen - ... It varies a lot on what people can do.

A number of participants described a sense of belonging to a community or group of other people with Down syndrome.

Ben – I know a lot of Downies... There's different kinds of Downs. It's actually around the world.

Participants described their own sense of self,

Mary - ...like people have got their own lives, really.

Helen – It's them. It's what makes them.

achievements and talents

Sophie – But it's not debilitating. If it was debilitating, I wouldn't be living on my own.

*Helen – But we have different talents that someone could bring to the world...
They could be really good Olympians. Someone could be into media and work...
(Omitted identifier)*

Sophie - Because I've got... friends who have Down syndrome and they are still achieving... in the documentary (Sally Philips), there's a boy with Down syndrome who has a six week contract on Coronation Street.

and self beyond disability

*Helen – We're still good people. Everyone's got good and bad in them.
We just live life to the fullest as much as we can. And we learn like everyone else but we take longer to get to the achievements.*

Sarah – (Answering, 'What does Down syndrome mean?') – They're happy.

*Sophie – Am I a burden? Am I a risk? No, I'm a person with feelings...
Because I have a friend... she may need more support but she is still precious.
That's what I am trying to get across... people with Down syndrome should be loved for who they are, not what they achieve.*

iv) Others' perception and understanding of Down syndrome

Participants highlighted their views about others' understanding of Down syndrome. Participants outlined that people have a general lack of knowledge and awareness about Down syndrome and that there is an assumption of inability.

Sarah – I don't think people really understand what Down syndrome is, really... and it's quite upsetting about that.

*Helen – They are ignorant to the fact that we can still do things.
I understand that Down syndrome is upsetting for some people but it doesn't mean that we can't do anything.*

There was an emphasis on peoples' fear and sadness.

*Helen – The first thing people think of Down syndrome is it's sad and that it's scary.
They don't know much about it.*

Sophie – And that's what I am trying to get across to the world. What's so threatening about Down syndrome?

Helen outlined that fear causes people to run away.

Helen – It's like, oh wow, that's scary. Run Away. I don't think so. Face it.

Helen and Sophie made reference for people wishing to have a perfect baby and that Down syndrome is not seen as perfect.

Sophie – They want a perfect baby. They don't think Down syndrome is perfect. They want a perfect baby.

Mary made reference to the lack of visibility of Down syndrome in the media

Mary – I've never seen young people with Down syndrome on TV.

Ben outlined that people should inform themselves about Down syndrome.

Ben – (In reference to a question about what he would say if someone didn't know much about Down syndrome) – So, I'd say, that's your mistake... Talk to the doctors again.

Helen expressed anger that people don't understand Down syndrome and terminate without fully understanding.

Helen – I think most people, being 90% are getting rid of it because it's Down syndrome. They don't fully understand what that means for the child.

Theme 2 – A test for Down syndrome

There are two subthemes within this theme. The first is participants' view of testing. Some participants expressed that they were aware of testing prior to this consultation. Some participants were aware of the difference between the old and new test.

Overall, participants' views about testing ranged from a strong belief that testing for anything was wrong viewing to testing as a means of preparation. There was a general view that prospective parents may want to know in advance and may wish to prepare.

Participant's views of termination following testing also ranged from strongly disagreeing with termination to disagreeing, feeling saddened or shocked by termination and at the same time respecting a woman's choice. They also referred to choice as helpful as a means of ensuring that child is wanted. Participants grappled with their own disagreement and a wish to respect women's' choices.

Considering the context set by the previous theme it is can be interpreted that participants were in favour of testing as a means of preparation as people are seen to lack knowledge and understanding of Down syndrome. It is possible to interpret that participants disagree with termination following a test on the basis that they see value and worth in their own lives and extend this to anyone with Down syndrome.

i) View of testing

Previous Knowledge of testing

Some of the participants had heard about testing for Down syndrome prior to taking part in the consultation and made reference to gaining awareness from watching TV.

Sophie – Me and my Mum, we've been campaigning.

Mary – Yeah, I've been watching the soaps.

Sarah – Well, actually, I talked to my Mum about the test. So we watched it (Sally Philips documentary).

Dylan – My Mum told me.

Old test and new test

A number of the participants were aware and understood the difference between the 'old' and 'new' test.

Sophie – I understand that it's 90% accurate and it can detect Down syndrome in the womb. It's different because it's more accurate. It can detect at an early stage. It (the old test)... didn't do good to the foetus.

Helen – There's this new test called NIPT test that's non invasive. That means that it doesn't harm the baby at all. It's a blood test through the arm, like, if you can get a normal blood test. But they can find out if your baby is Down syndrome through that.

View of testing

There were a number of different views with regard testing for Down syndrome. A couple of participants viewed the possibility of testing as a helpful way to gain knowledge.

Dylan – I think it's a good idea. It'll be much easier to find out in advance.

Dylan linked this to helping parents to prepare.

Dylan – I think it's a good thing that they know. ... know what they are expecting. They know exactly what to do. How to treat the baby... They might not know, parents might now know what it'll be like.

Similarly, Ben and Helen outlined that it would give prospective parent(s) knowledge.

Helen – It's a good thing. I think because it will help people find out if the baby has Down syndrome.

Sarah was unsure of her view of testing.

Sarah- I'm not sure.

Sophie held a strong view that she did not agree with testing under any circumstance.

Sophie -I do not agree with testing for anything.

ii) Decision following a test

Sophie held a very strong view that she was not in favour of termination of a pregnancy following testing.

Sophie – I don't agree with anything that takes life away... because life is precious, life is valuable.

Sophie linked her view to her religious views and expressed sadness at the decision to terminate based on a Down syndrome.

Sophie – It doesn't matter... you are still made in God's image.

Sophie – I think it's a very sad choice that people are terminating.

She went on to say that she would respect someone's decision and links to her religious beliefs.

Sophie – Well I would respect their decision. Because if God was, because the bible says love your enemy.

Sarah found the topic difficult to speak about and asked to move on. She did discuss in relation to watching the Sally Philips documentary and expressed sadness.

Sarah – (Speaking about the Sally Philips documentary) Yes, it was okay, it got a little upsetting but it's fine... It was to do with when they... the end of the pregnancy'.

Sarah – 'I would say it's a good idea' (with reference to continuing with a pregnancy).

Ben, Dylan and Mary outlined that they were upset, shocked, saddened by the decision to terminate. They also saw the decision to terminate as a woman or couple's choice.

Ben – It's up to them, really.

Mary – It's like if a woman wants a baby with Down syndrome or not.

Ben went onto discuss how babies with Down syndrome were previously abandoned and outlined that parents can't change their minds once a baby is born.

Ben – It's all happening a long long time because years ago, in my parents' days, there are actually some people like me, like Downies, ... normal people they will leave you somewhere out and your own parents left you... for good...

... They can't change their mind at all. They have to live with it . They have to live with it forever.

Dylan outlined that he would feel shocked if a woman/couple chose to terminate based on Down syndrome.

Dylan – That I guess I would be shocked... Because I think you would love the child...

And went on to say that he thought it was the parents' choice.

Dylan - It's up to the parents.

Helen outlined that she did not agree with termination based on Down syndrome and also thought that a woman has the right to choose.

Helen – They are getting rid of it if they're finding out it's Down syndrome because they don't want that life for their baby or something. I mean it's a personal choice. I'm a pro-choice person. I believe what the woman wants she should get. If she doesn't want a baby with Down syndrome she doesn't have to... It's a woman's rights.

Theme 3 – What does testing mean for me?

Participants referred to their own sense of feeling shocked and saddened by people's decision to terminate. Even though participants generally referred to informed choice, they still expressed disagreement and sadness on a personal level. One participant expressed a tone of resignation that people wish to know and may wish to terminate highlighting the power of the negative view of disability and the negative discourse that surrounds it in society. One participant considered that testing sends a negative message to her and others with Down syndrome of feeling unloved and unwanted.

Participants also discussed the impact on the Down syndrome community, especially the possibility that less people with Down syndrome would be born.

i) Personal impact

A number of participants referred to feeling shocked and sad when speaking about testing or terminating due to Down syndrome.

*Sophie – I think it's a very sad choice that people are terminating.
It makes me want to cry.*

Helen – (Speaking about 90% termination rates and watching Sally Philips Documentary) - ...It's shocking, really.

Sarah – It's upsetting, that bit.

Helen was clear that she believed that it was a woman's right to choose and also highlighted

Helen – I also think it's sad.

Sophie highlighted the message she feels is sent to her and people with Down syndrome.

*Sophie – Because it makes me feel like I'm not wanted in society. And no one loves us...
(Speaks about how her family loves her)*

But the government doesn't love me, because if they did love me they wouldn't do this.

Helen expressed a tone of resignation, explaining that she saw it as inevitable that people would like to know in advance.

Helen – It's sad, but it's part of life. It's society in a way because people want to know if they've got a Down syndrome child. If they've got it or not.

ii) Impact on the Down syndrome community

Sophie also spoke about the impact on the Down syndrome community as a whole.

*Sophie – I was thinking we could all get together and have a ballot... we should vote:
Do we want people with Down syndrome in our community? I think that's what we should do.*

A number of participants also discussed the possibility of less people with Down syndrome being born.

Helen - ...there will be less families for me to support.

Sarah – I think, for me, I'd like to have more Down syndrome people...

Theme 4 – Balanced Information

As highlighted in the first theme, all participants spoke about their view that people have a lack of understanding about Down syndrome. They also referred to the assumptions and prejudice that people may have about Down syndrome. They refer to people as feeling scared and consider that this will cause people to 'run away'. They refer to people searching for perfection. Participants' consider that a negative discourse about disability and a lack of information about Down syndrome leads to decisions to test and influences decisions following testing which have a possible impact on the self and potentially all people with Down syndrome.

For this reason all participants called for people to have more information about Down syndrome, to hear some positive aspects, educate themselves and meet people with Down syndrome and other parents.

It is possible to interpret that participants consider that people may make different decisions if they understood Down syndrome better.

i) A need to provide information for informed choice

All participants highlighted the need to provide information about Down syndrome to women or couples.

Mary, Sarah, Helen outlined that in order to make a decision about testing or termination a woman or couple should have access to information.

Mary - To read it from leaflets... and got pictures with kids with Down syndrome inside a book... you can get stuff on the internet about it.

Sarah – They should know about, they need to read a special book about Down syndrome. ... On the internet... Newspapers.

Helen – Educate them. Mostly online at the moment... you can find out about many different disabilities... Educate themselves. They can research it... So they can have a more informed choice of whether or not to keep the baby. I think it's good to get more educated choice... but I think they should have more information for them.

Dylan, who placed a key emphasis on testing as a means of preparation outlined the importance of giving a woman or couple information about Down syndrome.

Dylan – They might get information about it... I think doctors do give them leaflets... about the baby. How they would cope with the baby... (referring to the content of the leaflets) Questions like how do you feel having a child with Down syndrome?... It might help them a lot. (Later) I think they should be a lot clearer before they make that decision.

Dylan, Helen, Sarah and Sophie also placed an emphasis on meeting people with Down syndrome.

Dylan – (ask) other people in that situation... I would say - a lot of friends with Down syndrome. I would say – Would you like to come see (community theatre group)? ... See what we do. So, to see the group with Down syndrome. To see how we work.

Sarah – Because they need to talk to a person about their Down syndrome... If they come to (group) they will know what Down syndrome is, because they learnt or somebody will tell them.

Helen – Meeting people is important because it's meeting them personally and finding out about them... what their journey and their story. Like, you wouldn't think it of many Down syndrome people but we're actually quite sharp and strong people....

Sophie – They also want to meet someone who has that condition to talk to them about it.

They also outlined that it would be helpful to meet with other parents.

Helen – ... And they can get together in different support groups and talk to the different parents... They ask each other questions about how they got through different thing and milestones in the child's life and how they doubted it. So their support can go to another family...

Dylan also outlined what he would say to a woman or couple making a decision to test or terminate.

Dylan – I would tell them about who I am... I'll tell them that my Mum and Dad love me... Yeah, who I am. Because I've got Down syndrome.

Helen outlined her view of the type of information that people should receive.

Helen – We can educate them with all the information that we have about Down syndrome. The developmental delays and the speech delays and the heart defects, if it happens to be full Down syndrome... we just need to give them the information to make a well and informed decision for themselves.

Sophie views the information given to people as biased and highlighting negatives.

Sophie – (referring to doctors) because he'll give you lots of leaflets about things that go wrong... They also say that people are a burden to society and a risk...

Sophie - Because at the moment they only give them leaflets and everything that goes wrong, because Down syndrome is classed as a severe disability. But it's not.

Sophie highlighted the need to look at a more balanced view and also made reference to Sally Philips documentary and the message that her son had brought love to the family.

Sophie – You could look at the good bits.

Sophie – How much love and humour they can bring to the family. Because Sally Philips Said it's a comedy, not a tragedy.

