Experiences of prenatal diagnosis and decision-making about termination of pregnancy: A qualitative study

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Background: Advances in genetic technologies and ultrasound screening techniques have increased the ability to predict and diagnose congenital anomalies during pregnancy. As a result more prospective parents than ever before will receive a prenatal diagnosis of a fetal abnormality. Little is known about how Australian women and men experience receiving a prenatal diagnosis and how they make their decision about whether or not to continue the pregnancy.

Aims: This qualitative study aims to describe parental experiences and examine how best to provide support after a prenatal diagnosis.

Results: Individual in-depth interviews were conducted with 102 women and men approximately six weeks post-diagnosis of fetal abnormality. Data were elicited using a narrative, chronological approach and women (n = 75) and a sample of male partners (n = 27) were separately interviewed. Thematic analysis, involving a rigorous process of qualitative coding, enabled iterative development and validation of emergent themes. Participants identified that the shock of the diagnosis can be lessened when good care is delivered, by provision of: clear, accurate and respectful communication; empathic, non-judgemental, professional support; timely access to further testing and appointments; seamless interactions with services and administration; appropriate choices about invasive testing; acknowledgment of the enormity and unexpected nature of the diagnosis, and of the subsequent decision-making challenges; and discussion of the myriad feelings likely to emerge throughout the process.

Conclusions: This study has demonstrated the importance of providing timely access to accurate information and supportive, non-judgemental care for women and their partners following prenatal diagnosis of a fetal abnormality.

Key words: decision-making, fetal abnormality, prenatal diagnosis, qualitative, termination of pregnancy.

Introduction

Little is known of Australian women’s and couples’ experiences of prenatal diagnosis and subsequent decision-making; international data reveal shock, anxiety and acute grief reactions and report intense distress, regardless of the severity of the condition diagnosed. Decision-making after prenatal diagnosis is known to be challenging for women and their partners and can arouse decisional conflict and decision regret.

Recent data from Victoria, Australia indicate that, when a fetal abnormality such as Down syndrome is diagnosed prenatally, the majority of pregnancies are terminated. Termination of pregnancy (TOP) performed in these circumstances is associated with greater psychological morbidity than TOP for non-fetal indications. It is therefore imperative to determine how best to support parents after a prenatal diagnosis and facilitate informed decision-making about TOP.

The PeTALS project (Prenatal Testing: A Longitudinal Study) is exploring the experiences of women and their male partners who receive a prenatal diagnosis for a variety of conditions with variable severity. The study aims to identify social and professional supports needed and used at the time of diagnosis and in the months and years that follow. Here we report a large sample of individual interviews conducted approximately six weeks after a definitive prenatal diagnosis, describing women’s and men’s experiences, their support needs and the factors
they considered when making a decision about whether or not to terminate their pregnancy.

**Materials and Methods**

**Setting**

Prenatal screening is utilised by over 80% of pregnant women in Victoria. While second trimester serum screening is fully funded by the Victorian Department of Health, first trimester combined screening (first trimester nuchal, nasal bone and serum screening) incurs a private fee. Non-invasive testing of cell-free DNA (maternal plasma) is commercially available in Australia and is a user-pays test. Genetic counselling is usually available to Victorian couples who receive a prenatal diagnosis.

**Ethics approval**

Human Research Ethics Committee approvals were obtained to recruit patients from three antenatal settings in Victoria, Australia: one private ultrasound practice and two public hospitals (University of Melbourne: 1237351; Monash Surgical Private Hospital 12100; Mercy Hospital for Women: R12/60; Royal Women’s Hospital: R12/41).

**Participants**

Purposive, convenience sampling identified eligible participants who: had recently received a prenatal diagnosis of a fetal chromosomal, cardiac or other structural abnormality; were ≥ 18 years of age; had attended genetic counselling; and were English speaking.

**Recruitment**

Genetic counsellors approached eligible women and couples at, or shortly after, the time of diagnosis and provided verbal and written study information. Those who agreed to their contact details being passed on to researchers were telephoned after two weeks to enrol in the study.

**Participation**

Telephone interviews involved participants relating their story of prenatal diagnosis and the events that followed. Eligible couples agreed to be interviewed separately, at the same time or within a few days of each other.

The semi-structured interview guide followed a narrative and chronological style, exploring experiences of: early pregnancy, undertaking screening in pregnancy, first indication of abnormality, considering and undertaking invasive testing, making a decision about termination of pregnancy, support sources utilised during this time (formal and informal) and perceptions of impact on the couple’s relationship. Experiences of termination procedure and after-care, or expectations of remaining months of pregnancy will be reported elsewhere.

**Data management and analysis**

Interviews were audio recorded and transcribed verbatim. Transcripts were validated, de-identified and participants assigned pseudonyms. Analysis was managed in NVivo 10 (QSR International Pty Ltd, Melbourne, Australia). Thematic analysis, involving a rigorous process of qualitative coding and discussion by several team members to identify similarities and differences, enabled iterative development and validation of emergent themes.

**Results**

**Participation**

Between July 2012 and October 2013, 59 women were invited to participate; of these, 39 women agreed. Additionally between May 2014 and April 2015, 61 couples were approached with 36 consenting, comprising 27 male/female couples (interviewed as individuals) and nine women who participated without their partner. Overall, 75 pregnancies are represented by the 102 participants in this sample, from 120 eligible pregnancies, giving a participation rate of 62.5%. Characteristics of the participants and the pregnancies are described in Table 1.

Findings are presented using quotes as exemplars and attributed to participants by pseudonym, diagnostic category and pregnancy outcome, either a TOP or intention to continue the pregnancy (CP). Some quotes have been truncated for ease of reading without changing meaning as indicated by ‘…’.

‘…we didn’t see it coming’

First indications that something was amiss often occurred during a ‘routine’ ultrasound; participants anticipated seeing their baby and felt unprepared for an extreme shift in emotional state after disclosure of a high-risk result:

I just thought .....it would be a routine ultrasound.....we didn’t see it coming. Kate, other chromosomal, TOP

Being given a private space to absorb the unexpected information was appreciated:

They said, ‘OK we’ll leave you in the room for a minute just to get yourselves together’. And then (my partner) and I cried together. They came back in, and that’s when I felt like we could hear more information. Rochelle, structural anomalies, TOP
resulted in fetal death

Intention to continue pregnancy †

Pregnancy outcome, †

Single gene mutation:

Cardiac anomalies:

Other chromosomal:

Other trisomies:

Structural anomalies:

Trisomy 21 24 (32)

Prenatal diagnosis, †

Parity, n = 75

Gravidity, n = 75

Men 27 (26)

Women 75 (74)

Age range of participants

18–29 years 18 (18)

30–35 years 40 (39)

36–39 years 28 (27)

40–49 years 16 (16)

Gravidity, n = 75

1 19 (25)

2+ 56 (75)

Parity, n = 75

0 32 (43)

1+ 43 (57)

Prenatal diagnosis, n = 75

Trisomy 21 24 (32)

Structural anomalies:

(arthrogryposis, renal agenesis, unilateral cleft lip and absent corpus callosum, acrania, omphalocoele, vertriculomegaly, intrauterine growth restriction, posterior urethral valves, multiple brain abnormalities, hydrocephalus, limb and bone abnormalities)

Other trisomies: 12 (16)

(T18, T13)

Other chromosomal: 11 (15)

(monosomy X, uniparental disomy 14, derivative chromosome, mosaic T4, triploid, Smith-magenis, 22q11 triplication)

Cardiac anomalies: 10 (13)

(hypoplastic right heart, hypoplastic left heart, tetralogy of Fallot, right atrial isomerism, ventricular septal defect, transposition of great arteries, right aortic arch)

Single gene mutation: 3 (4)

(spinal muscular atrophy, Marfan syndrome)

Pregnancy outcome, n = 75

Termination of pregnancy 59 (79)

Intention to continue pregnancy† 16 (21)

Gestation at diagnosis Range: 10–21 weeks

Interview length, mean 84 min

(range: 35–179 min)

†At the time of the first interview, two of these pregnancies had resulted in fetal death in utero.

‡A mean gestation cannot be calculated as these data were often approximated or self-reported gestation.

### Considering and experiencing diagnostic testing

When considering further invasive diagnostic testing, participants commonly assessed the risk of a procedure-related miscarriage and sought advice from health professionals to allay fears:

The risk of miscarriage from chorionic villus sampling (CVS) was a lot less likely than the chances of it being (condition) so there was really no issue in my decision. Melinda, other trisomy, TOP

I actually spoke to (health professional) quite a bit during that week because she was a really good support, just to have someone to talk to. Grace, structural anomalies, TOP

Confidence in the practitioner was cited as reassuring and contributed to the decision to have invasive testing:

You hear that the amnio risks depend on who does them and I was confident that (health professional) had done many of them. Zoe, other trisomy, TOP

Prompt referral for the diagnostic procedure was highly valued (row 1, Table 2). Despite diagnostic testing being incredibly stressful, participants reported aspects of care that contributed to a better experience. In particular: being given choices about viewing ultrasound images (row 2, Table 2) and feeling well treated by caring staff (row 3, Table 2).

### An agonising wait

Following prenatal diagnosis, participants who paid a surcharge could access fluorescent *in situ* hybridisation (FISH) testing with preliminary results available the following day but others waited ≥ 2 weeks for the full karyotype. Results were usually disclosed over the telephone and aspects of the disclosure process that were important included: being informed about when results would be available and, for those having FISH testing, when results would be finalised and having choice in how and to whom results would be disclosed (row 4, Table 2). When expectations of receiving results were not met, stress and anxiety were exacerbated:

(Health professional) was supposed to ring with the results on Friday but she wasn’t working that day....I was planning to tell family and friends on the weekend. ....I thought ‘oh my god, how am I going to go the whole weekend?’....I was constantly ringing the hospital to see if they had my results. Kylie, other chromosomal, TOP

### A ‘shocking’ disclosure

Even when participants knew when and how they would receive results, hearing the result was described as ‘shocking’. In some cases this was made worse by the perceived insensitivity of the person involved:

I don’t like the way they delivered the news at all....‘oh well that’s what you expected wasn’t it?’
was trying to hold on to some hope. *Madeline, trisomy 21, TOP*

Many participants recalled feeling unsupported immediately after receiving the diagnosis and expressed a need for better access to comprehensive information:

We weren’t given any particular booklets or brochures or directed to any particular website . . . . we had to go seek the information ourselves. *Poppy, trisomy 21, TOP*

Empathic acknowledgement of the enormity of the news by health professionals and access to timely follow up were greatly valued (rows 5 and 6, Table 2).

### The ‘hardest part’ of the process

Participants identified the ‘hardest part’ of their experience as having to make a decision about continuing or terminating the pregnancy:
Discussion

This study describes experiences of a large sample of women and their male partners at the time of prenatal diagnosis and during decision-making about TOP for a range of conditions with variable severity. The findings provide further evidence of the significant demands imposed on parents’ psychological resources after prenatal diagnosis of fetal anomaly.

Despite limited empirical evidence, practitioners in this area are likely aware that effective preparation for disclosure of a prenatal diagnosis can reduce shock and distress and thereby increase ability to process information and deliberate about further options. The present study provides critical evidence to support this and demonstrates how the emotional impact of the diagnosis can be lessened when good care is delivered, namely by ensuring: timely communication of clear, unbiased information; seamless access to appointments; acknowledgment of the enormity of the news of the diagnosis; and discussion and validation of the decision to terminate the pregnancy. A recent Swedish study also articulated the positive impact of well co-ordinated care when women receive clear and consistent information and help with navigating health systems and are treated with dignity by health professionals.13

In the present study, and others, the detection of a fetal abnormality continues to be unexpected and shocking4,14,15. Ultrasound is increasingly seen as a ‘routine’ part of pregnancy,16 and non-invasive prenatal tests and chromosomal microarray analyses now frequently provide more detailed information earlier in pregnancy than has hitherto been possible.14,15. It is therefore vital to consider how extreme emotional responses to prenatal diagnosis may impact upon an individual’s capacity to process information and make informed choices.

Supportive care from health professionals appeared to ameliorate participants’ anxiety about diagnostic procedures and test results. Ideal care included being given a number of acknowledgments: of the enormity and unexpected nature of the diagnosis; of the subsequent decision-making challenges; and of the myriad feelings likely to emerge throughout the process. Sensitive communication in prenatal testing is essential.2,17. Many participants clearly recalled words spoken, words that either caused further distress or provided great solace (Table 2). Words and statements that conveyed information or results, as well as acknowledging the emotional impact were the most helpful: ‘I’m afraid, it’s not good news’. In contrast, when information was given without overt empathy and acknowledgment of the enormity of the diagnosis, ‘oh well that’s what you expected wasn’t it’, distress was exacerbated.

Decision-making was a complex process; sourcing accurate and balanced information about the condition that had been diagnosed was a particular challenge. Many were wary about accessing information from condition-specific support groups, anticipating biased perspectives.

Valuing confirmation of the decision about the pregnancy

Those who chose to terminate their pregnancy appreciated having their decision validated by others and viewed this as a subtle, but very helpful, aspect of the care they received:

When we had that conversation, the specialist then said ‘what you’ve decided is what the majority of people do in your situation, I couldn’t tell you that before because I can’t influence your decision, but you have really made the right decision for the baby’. Patricia, cardiac anomaly, TOP

For most participants the decision was not immediately clear:

Being confronted with it you have no choice but to make a decision…..at the time I just thought I don’t want to make a decision because I don’t want either option, I wish that neither of them were there. Janet, trisomy 21, TOP

Several described changing their prior views about termination of pregnancy:

At first we thought we will terminate if (baby) turns out to be sick but once we got the diagnosis and we were in the counselling session, we were sort of ambiguous but we were leaning towards keeping (baby) Evelyn, other trisomy, CP

Many described having to choose between two horrible options:

Making that decision…..I felt like I’d been given two bags, or two buckets of shit and I was being asked to choose which bucket I’d like. Dianne, trisomy 21, TOP

 Provision of relevant, unbiased information, in a caring way, about likely prognosis and termination procedure options contributed to more positive experiences:

(Health professional) went through all of the results and was really open and honest, went through the process of being induced and again it was still my choice and I didn’t feel like I was being rushed into making the decision. Georgia, structural anomalies, TOP

Participants articulated important elements of their decision-making about whether or not to continue the pregnancy (Table 3). In addition to specific information about the condition, these included consideration of: prior views on abortion and disability, perceived impact on family life, what/how others might think about their decision and what others would do/have done in similar circumstances.

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Table 3 Considerations in the decision about whether to terminate the pregnancy

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| Uncertainty about diagnosis/unclear results | I didn’t feel like I wanted to have or needed to have that conversation (about termination of pregnancy) until we actually had some solid results  
Eloise, cardiac anomalies, CP  
We had the FISH results....‘Everything is clear’ and I burst into tears I was so happy....It was the best news in the world. She said ‘the full results haven’t come back, they will come back in two weeks’....she phoned us back and said ‘I’m really sorry we found that there’s a (chromosomal) abnormality’....it just felt like the world had ended and I don’t think we really functioned that night....we just didn’t know what to do at that point you know. It couldn’t get any worse  
Ava, other chromosomal, TOP                                                                 |
| Uncertainty about prognosis or severity     | At first I didn’t know what I wanted to do...I thought that with surgery it might be alright....but then each scan, once it was like three or four or five things going wrong with the baby’s heart....That’s when I realised that it’s probably not the best thing for me or for the baby (to continue the pregnancy)  
Claire, cardiac anomaly, TOP                                                                                                                        |
| Expected survival: to term or shortly after | Every now and then I still think ‘did I do the right thing?’ The thing which keeps me going is, I don’t know if I’m just grasping on to it to make myself feel better or not, that it’s more than likely that the baby wouldn’t make it past 5 months, and I couldn’t go through that  
Heather, trisomy 21, TOP                                                                                                                            |
| Worry about baby experiencing pain          | We both felt we didn’t want to bring (baby) to term for (baby) only to survive a little while and it to be very painful and lots of tubes  
Fiona, structural anomalies, TOP                                                                                                                     |
| Drawing on prior views about what they might do |                                                                                                                                                                                                                     |
| Personal views on abortion and disability   | It has never been an option for my husband and I, we would never consider it....just our beliefs, and our religious beliefs.....we’ve always known  
Beth, cardiac anomaly, CP  
While it was really hard to deal with, unfortunately that’s life, everything happens for a reason. We made the decision before we even found out that I was pregnant that if the baby had Down syndrome that we couldn’t go ahead because that wasn’t a life that we wanted for our child, we knew that we were never going to go ahead and our families supported that 100%  
Deborah, trisomy 21, TOP                                                                                                                            |
| Self-perceived ability to parent a child with a disability | I guess quality of life for the actual child as well as everybody around them is something that I really took into consideration. ....if you had a child who had severe problems it would have an impact in so many ways on so many people......it just got to a point where I felt that it was just beyond our capabilities I suppose  
Janet, trisomy 21, TOP  
It doesn’t scare me to have that sort of a challenge....I feel I have the ability to deal with these things if they came along  
Natalie, other trisomy, CP                                                                                                                             |
| Considering impact on family life           |                                                                                                                                                                                                                     |
| Potential for long-term care needs          | I think I have safeguarded him (baby) as well....it comes to a point when I can’t, I can’t you know, can’t take care of him  
Caitlin, trisomy 21, TOP  
For me the real thing that I couldn’t get past was the fact that they always need so much care and that they never leave too so I felt like I could’ve had that child, if at 20 (years of age) it was going to leave home but knowing that it would live with us forever....that’s what I couldn’t resolve  
Suzanne, trisomy 21, TOP                                                                                                                             |
| Societal treatment of people with disabilities | We decided because of this society we couldn’t bring a Down syndrome child into the world because I know how cruel people can be and I saw it at school....it’s already hard enough for a child these days let alone having you know disabilities and so we couldn’t do that, we just thought it was too unfair and it would’ve been selfish.....for us to have the child because we need a child, we thought of the child first before us  
Madeline, trisomy 21, TOP                                                                                                                             |
| Impact on other children                    | If something was to happen to us....it would put a huge burden on our other children too, for the responsibility  
Diane, trisomy 21, TOP                                                                                                                             |
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| Impact on career and finances | My ability to work, my family’s quality of life and my son is going to need a decade or so of follow up. Bringing a child into this world that could have serious complications with their health was going to just make everything even so much more hard. …we wouldn’t have the home that we have and it’d be very difficult but it’s just heart breaking to have to make a choice to end the life of a child that you so dearly wanted  
  *Poppy, trisomy 21, TOP* |
| Potential maternal risks in pregnancy | I had to consider my own health obviously as well. If the baby didn’t do well with the right side of the heart that could put stress on my organs.  
  *Claire, cardiac anomaly, TOP* |
| Not the life they imagined for their child | We were both very much on the same page straight away that if there was any sense of disability where they wouldn’t get to live the kind of life, a full life that (baby) should get to live then we didn’t feel it was fair to bring (baby) into the world  
  *Fiona, structural anomalies, TOP* |
| Considering what others might think about their decision to have a TOP | Because as much as it doesn’t really matter what other people think, unfortunately I do care, and if people thought I was an evil person that would hurt  
  *Patricia, cardiac anomaly, TOP* |
| Fearing judgment | (Partner) she was you know having real difficulty. …what would people, judge her you know, she did have a problem with the perception of judgement that other people might judge her  
  *Trevor, other trisomy, TOP* |
| Societal views about abortion | I didn’t want to make that decision but I was just in a way hoping it would take care of itself or something. …I was watching a TV show I think the day after (the amniocentesis)….about how abortions are wrong no matter what reason…was watching that and I was thinking ‘oh my god how bad’  
  *Kylie, other chromosomal, TOP* |
| Wondering what others would do/have done in the same situation | I think it’s still a bit of a sensitive issue with a lot of people as well in terms of abortion  
  *Ben, single gene change, TOP* |
| Seeking out shared experiences, eg blogs and forums | So I was feeling very much like I was only getting one side of the story (from an online support group). …I started feeling a little bit, no one was forceful at all, like they were all really supportive but obviously encouraging of one side (to continue the pregnancy)  
  *Patricia, cardiac anomaly, TOP* |
| Asking health professionals about their experience | (I was part of) a forum type website and I put it out there asking if anyone else had a baby who had this and a lot of people came back. …they terminated or they lost their baby anyway because they pass away all the time before they are born  
  *Evelyn, other trisomy, CP* |
Instead pregnancy or pregnancy loss-related internet sites, largely unmoderated and lacking evidence-based information, were sought. Popularity of web-based resources means health professionals are becoming less likely to be the primary information source. As such they have a new and important role in assisting patients to navigate and discriminate between these resources.18

In our sample it was evident that understandings of Down syndrome were highly variable. Negative perceptions about what it might mean to care for a child with Down syndrome (Table 3) included outdated stereotypes about disability that portrayed ‘exclusion’ and ‘burden’ rather than a more contemporary reality of ‘inclusion’ and ‘potential’. A challenge in prenatal testing, particularly with conditions of variable phenotypes, is to provide realistic condition-specific information that does not overplay or underplay lived experiences.19

Participants who chose to terminate their pregnancy were often fearful of being negatively ‘judged’ by others (Table 3). Many appreciated having their ultimate decision validated and sought reassurance from health professionals. If parental decisions about whether or not to terminate a pregnancy are indeed considered to be a choice between two equal options, are informed by accurate information and, have been deliberated upon, then our findings suggest that health professionals offering such validation may promote adaptation and possibly reduce later regret.

Participants in the present study were recruited from genetic counselling services and therefore all were exposed to some degree of counselling and aftercare. As such the findings may under-represent parental distress at this time. All participants were English speaking; future research is needed to explore the experiences and support needs of women and couples of varied cultural and language backgrounds. A further limitation of the findings is that a third of the sample received a diagnosis of Down syndrome and all had a termination; experiences of those who continued a pregnancy after a diagnosis of Down syndrome are not captured by this sample. This study has interviewed women and their male partners; ensuring the male perspective is heard is essential. Male participants described (Table 2 and Table 3) being present at the time of diagnosis and invasive procedures, making decisions about results disclosures and supporting their partner as she grappled with making a decision about abortion. It is clear that male partner experiences should be further researched to allow support and care to meet their needs after prenatal diagnosis. Greater detail of our male participant experiences will be reported in a subsequent manuscript.

The rate of detection of congenital abnormalities during pregnancy is likely to rise with the introduction of newer genetic technologies having greater specificity and sensitivity.20 There is an ethical imperative to ensure that those who receive a prenatal diagnosis of fetal abnormality are supported appropriately and can make informed decisions about available options.

In summary, evidence from this study shows that best care includes provision of: clear, accurate and respectful communication of results and testing procedures; empathic, non-judgemental, professional support; timely access to further testing and appointments as needed; seamless interactions with services and administration processes; information to assist with making appropriate choices about invasive testing; acknowledgment by health professionals of the enormity and unexpected nature of the diagnosis, and of the subsequent decision-making challenges; and discussion of the myriad feelings likely to emerge throughout the process for the woman, her male partner and both of them as a couple. We recommend prenatal diagnosis care services incorporate these components into their practice, and it would seem unethical to provide prenatal diagnosis unless this is available.

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Conflict of Interest

The authors have no conflicts of interest to disclose.

References


