CONFRONTING THE DISTORTIONS: MOTHERS OF CHILDREN WITH DOWN SYNDROME AND PRENATAL TESTING

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Prenatal diagnosis represents the most immediate and widespread application of the current explosion of genetic knowledge and technology. Presently hundreds of genetic conditions can be identified through prenatal diagnosis, although testing commonly focusses on chromosomal analysis and testing for neural tube defects (Wertz 1992:162). The major diagnostic tests are diagnostic ultrasound, amniocentesis and chorionic villus sampling (CVS). In recent years, there has been a drive to confirm test results earlier in pregnancy both by performing these tests earlier and by developing faster methods of chromosome analysis. The development of screening tests, including first trimester ultrasound and maternal serum screening tests, has already reshaped the landscape of prenatal diagnosis. Prenatal testing, once limited to women considered “high risk”, is increasingly a routine part of all pregnant women’s antenatal care.

In the United Kingdom do-it-yourself urine tests to screen for Down syndrome are being developed. Other promised developments include blood tests which could isolate foetal cells in maternal blood. Such testing matches what Western women already expect during standard antenatal care. Testing is presented as not only benign and unproblematic, but also as fulfilling responsible pregnant behaviour. Alongside this construction of prenatal testing as “beneficial medical advance”, disability continues to be constructed in wholly negative and prejudicial ways.

My attention to the ethics of prenatal testing arrived five years ago with the birth of my son Sean who has Down syndrome. As part of my doctoral research, I have interviewed seventeen other mothers of children with Down syndrome about how they experience the ethical and personal choices about prenatal testing, particularly in subsequent pregnancies.
Over the last four years I have also acted as a new parent support contact for the Down Syndrome Association of Queensland (DSAQ). The DSAQ has found that increasingly couples are receiving the news that their child has Down syndrome prenatally. Also many pregnant women are facing the possibility that their child has Down syndrome when so-called “soft signs” are revealed on ultrasound or when serum screening tests place them in a high-risk category. The DSAQ held long discussions about how to respond to these new circumstances. Most of our literature presumed that parents were receiving the news of their child’s diagnosis shortly after birth and was inappropriate for parents receiving this news prenatally. Those of us involved in the new parent support work were also concerned about stories of pressure to terminate pregnancies and the lack of information about Down syndrome which was being provided to those receiving positive testing results. We decided to develop new information kits for expectant couples and material for professionals which would outline our expectations of the information and support which should be available.

Following a long process of debate the DSAQ also determined to have a position statement on prenatal testing. There were many issues to work through in coming to this decision. There were fears about entering such a controversial area and pessimism about producing an agreed statement given our diverse beliefs and feelings. We persevered because it was recognized that prenatal testing is an issue of utmost importance for people with Down syndrome. It seemed important not to be silent on this issue and to assertively place the lived experience of people with disability and their family members in the public arena.

This article explores some of the issues presented by prenatal testing which the DSAQ position statement (presented in Appendix) seeks to address. It also draws upon my own research to illustrate how the perspective of mothers of children with Down syndrome disrupts many of the assumptions which underpin prenatal testing.

**Messages about disability implicit in testing**

There is an implicit message within the goals of prenatal testing that society believes that raising a child with disability, is such a grave burden that it is both morally correct and medically appropriate to take expensive measures to ensure that such children are not born. (Murray 1996:132).
Testing is frequently presented as a compassionate enterprise, assisting families to avoid the birth of a child with disability. Early testing is sought to avoid the difficulties of late terminations. However, earlier testing and multiple, periodic forms of testing also emphasize the importance of detecting all affected pregnancies and further entrench a consensus that the rational response to a positive prenatal diagnosis is termination. The language of “choice”, “reassurance” and “family well-being” obscures a reality that universal population screening is introduced because it is supported by cost-benefit analyses which assess the prevention of the birth of babies with disability as a core social benefit.

One mother in my study had revealing discussions with a geneticist shortly after her son’s birth:

One thing the genetic counsellor said was, you know, when you have a child with Down syndrome you do need to consider the impact socially, the child has on society, that they will never be a wage earner and that they draw from society and I thought, I remember thinking at the time that’s one very odd sort of statement!

The genetic counsellor then suggested she consider the impact having one child with disability was already having on her daughter and her marriage. Jenny reflected on her unspoken reaction:

Have I let the whole of society down and my family down and my daughter down? Is this what you’re saying to me?

I’m unable to contribute to society because I’ve got a child who is non-productive, excuse me?...He’s like three months old....I mean John may grow up to be a terrific artist, or he may just be himself and give a lot of people a lot of joy. Then, the relationship with Emma, I felt crucified over that. And then my marriage...and I felt well there’s probably not a lot left when you take that away.

The idea that people with disability contribute to their families and to the community and that the experience of disability adds to our understanding of what it is to be a human being, is readily dismissed as irrational or sentimental. Even where it is acknowledged that parents can find the experience rewarding, they are considered “lesser” rewards. It might be conceded that once a child is there you have no choice but to make the most of it, but it is an experience which it is sensible to avoid.
Because there are few treatment options, testing is fundamentally linked to the option of “therapeutic” abortion. This reality is often obscured. Prenatal diagnosis is marketed as a form of “reassurance” rather than a form of prenatal selection (Lippman 1992:144). The reassurance rationale is undermined by evidence that testing may merely relieve the anxiety which has been artificially raised by the testing itself (Gates 1994). Testing also offers a very limited form of reassurance; a normal set of chromosomes does not rule out the possibility of disability.

Within the marketing of testing the “fear” of disability is both encouraged and overstated. In March this year there was a rush of publicity over the development of laboratory processes which give chromosomal results more rapidly. Dr Ian Findlay, one of those responsible for developing the so-called “one day test for Down syndrome”, was quoted:

This means parents can decide on the next step sooner and do not have to experience the despair of waiting three weeks for an answer. (University of Queensland News, March 14, 2000).

This exemplifies key problems within the dominant biomedical explanation of prenatal testing. Prenatal testing rescues couples from “despair”. It is common for expectant mothers and fathers to feel anxious about the health of their baby and waiting for test results can be a difficult time. Nevertheless does the mere possibility of a diagnosis of Down syndrome warrant despair? Is prenatal testing actually reinforcing an unjustifiable fear and horror of disability?

There is also the existence of the implied “next step” and a momentum which makes almost instantaneous decision making preferable to any prolonged introspection about the implications of a positive test result. Statistics indicate that less than 2% of couples in Victoria continue pregnancies following a diagnosis of Down syndrome (Ford 1999:69). Proponents of prenatal testing often offer the high rates of termination as proof that they are meeting consumer demand. I suspect something different in these statistics. I fear decisions made in haste and without adequate information about what disability is likely to mean for their child and for themselves as parents.
How different the statistics might look if couples were given the knowledge that families with disability are able to provide—not only the demands but the resources, not just the stresses but the rewards, the news that siblings are not usually adversely affected but share warm relationships with their brother or sister with disability, that many couples find their marriage enriched not threatened and that overall the experience is positive and ordinary. Many, maybe most, would probably still terminate. However evidence from an American genetic counselling service where couples are actively encouraged to have contact with families with a member with Down syndrome, that 38% continue the pregnancies (Parens and Asch 1999, s9) compared to the 2-16% reported elsewhere (Rapp 1999:223, Ford 1999, Drugan, Greb et al 1990).

**Pressure of decision-making in a time of crisis**

In July this year reports surfaced on the termination at a Melbourne hospital of a 32-week old foetus with dwarfism. The pregnant woman was reported to be actively suicidal. Again, the medical story of prenatal testing as a compassionate enterprise was established.

There would be few clinicians who cannot empathize with the pain and suffering of both this patient and her practitioners, when faced with such a clinical situation. (Dr John De Campo quoted in *The Australian*, Wednesday July 12, 2000).

As a new parent support contact, countless professionals have told me that parents are not coping with the news or that they are severely depressed. When I speak to the parents and listen to what they have been told (or significantly not told), the reaction of family and friends and all the aspects of the diagnosis which they must come to terms with, I am not surprised that they are “depressed”. I know that my own reaction to Sean’s birth was made so much more difficult by the “empathy” (read: pity and scaremongering) of some professionals. There can be no doubt that for most individuals the news that your child or potential child has a disability will be painful, disorientating, even devastating. My ongoing contact with families I visited in those raw early days, and current research, makes it clear however, that parental “suffering” does end and that the great majority of families adjust well to their new circumstances, despite the often real demands which exist. If obstetricians and counsellors consulted families who had survived this painful early period, they may be more likely to offer more than an “empathetic” termination.
The women I interviewed described remarkably similar stories of adjustment to the news of their child’s disability; familiar stories of disappointment and compromised dreams, gradually replaced by hopefulness and a realization that life continues “normally” and “ordinarily”. The salience of the label of Down syndrome was greatly reduced or as one woman said “You don’t wake up every morning thinking “Oh my God, he’s got Down syndrome”.

I haven't got anything to compare it with so it doesn't bother me that much and I get a bit shocked actually when people start making Down Syndrome talk with me....I guess I see her as a baby and....it still comes as a jolt at times to realise that she's got Down Syndrome.

He must have only been hours old and I remember looking at him and thinking and it broke my heart and I thought to myself no-one’s ever going to fall in love with him ... What a ridiculous thought, ‘cause I had.

Many mothers had vivid recollections of their early fears. It is likely that these fears are similar to those experienced by women receiving a positive diagnosis prenatally.

I knew immediately I had to arrange a termination. Looking at my son, he’s my main reason...He needs my attention. He doesn’t need a little sister who’s going to be a vegetable for the rest of her life. (Woman explaining her decision to terminate a pregnancy following a diagnosis of Down syndrome. *The Australian Magazine*, January 20-21, 1996).

From their present standpoint the participants described their initial expectations as foolish, rooted in ignorance and based on a skewed expectation that their life would be irrevocably changed in negative ways.

I mean life is completely different having a Down’s kid...but it’s full of so much fun and joy that it’s...it’s just different.
Is women’s reproductive autonomy enhanced by prenatal testing?

Routine testing can be difficult to decline and so can diminish (rather than enhance) autonomous choice. Many feminists have critiqued prenatal testing on the grounds that prenatal testing regimes contribute to the medicalization of pregnancy and actually diminish women’s control over their own bodies. Additionally many have recognized that decision-making often occurs without adequate knowledge of the purpose of the testing or about the conditions for which testing is conducted. Two women who had amniocentesis following ultrasound findings were not told the miscarriage risk of the procedure. Sally related that the test was described as absolutely standard, presented as no more serious than having a urine test, “Why wouldn’t you want it?”.

Down syndrome is “well recognized” and particularly surrounded by unhelpful stereotypes. The diagnosis of Down syndrome is assumed to provide sufficient information on which to make a decision to continue or terminate a pregnancy. In contrast, the women’s stories indicated the way they now contested their initial understandings of Down syndrome. The mothers overwhelmingly saw their children as healthy (not ill or diseased) and saw Down syndrome as a “lesser disability” or scarcely a disability at all. They also saw the diagnosis as inherently ambiguous. While the chromosomal result for Down syndrome is definitive, the message from mothers of children with Down syndrome is that it does not tell you how your life or the life of your child will be.

It is the common wisdom of parents of children with Down syndrome that the first days are the hardest and the time when it is most difficult to think calmly and realistically. Some doctors push women to make decisions quickly while still reeling from the diagnosis. Julie was strongly encouraged to terminate her pregnancy the same afternoon she received the diagnosis.

They were ready to take us up there straight away and do it. There was no, oh here’s the number for the Down syndrome association or anything, give them a call...there was no, no information. They were just thinking, Down syndrome get rid of it.

A number of participants spontaneously suggested that they would have previously terminated a pregnancy for a diagnosis of Down syndrome. This awareness led many to suggest they were extremely grateful that they had not known because they would have “gone into panic mode”: 
In retrospect if someone had’ve told me that you’re going to have this, and this and this (extra medical problems) I probably would have considered my options then, although I’m very, very glad that we didn’t.. you know I’m really pleased with the way everything’s worked out. We think that we have the most beautiful baby in the world.

Four women in my study continued pregnancies following a positive diagnosis of Down syndrome. Three of the women stated that termination was not an option. One woman, intended to terminate her pregnancy but changed her mind in the forty-eight hours following the diagnosis:

I just walked out and I said I don’t want this kid, get rid of it. As soon as I was out the door of the office that's the first thing I said. We’ve still got him, something happened somewhere (laughing)... Once I sort of got over that, you know, it didn’t take long. I just sort of settled down and went well, you know, you’re being stupid...could be worse.

**Will we have a community without people with Down syndrome?**

Prenatal diagnosis raises profound questions about the value of children and the sort of human community we are creating.

Prenatal testing promotes an emphasis on control and choice. But these are largely illusory in the context of parenthood. The journey into parenting is a leap of faith, one which requires considerable courage for we cannot control either our children or the circumstances they are likely to meet. Barbara Rothman questions the helpfulness of “standards of acceptability” in preparing for the “reality of parenthood” (Rothman 1986:7). As Thomas Murray suggests “Good families are characterized more by acceptance than control” (Murray 1996:31). It may be that “good communities” are characterized by similar values of acceptance and flexibility.

Many mothers described a journey of surrendering control: that there were “no guarantees”, that “things happen for a reason” and that giving up one’s prior expectations of motherhood and of your offspring could be a gift.
This idea that having a child with disability had brought a richer life was very common in my interviews. The positive changes included having one’s prejudices about disability dismissed, discovering one’s own resilience and strength and the ability to speak up for yourself and your child, and feeling a strong sense of what really mattered.

Many participants believed that losing potential society members with Down syndrome was a “loss for society”.

   It would be like doing without the colour yellow in a tapestry. We’re not doing anybody a favour, because they really...they have their own input, their own colour, they make a difference, they light up things... like yellow does.

Jenny commented on her mixed emotions while undergoing testing. While endorsing women’s rights to testing and termination, and believing that having testing to be forewarned was the right decision for her personally, she felt some degree of complicity with a system which sees the prevention of Down syndrome as a social good.

   I told someone he was doing the test and they said “Oh, he’s very good, he rarely misses one”, and I just felt so ashamed! It’s terrible, that’s such a dreadful thing to say, and it’s such a loss, and it’s a loss for our society and the way we think and what we value...you hear stories about millionaires, or women who’ve, people like Poppy King and the lipsticks or whatever and they’re being held up as these you know, icons and everything...(But) because you are a really nice person like John is and has such potential for joy and just, you know, being true, you’re not considered worthy.
Conclusion

There is a disturbing gap between the technological momentum and discussion on the impact of these developments. Despite continuing media interest in the issues, I contend that the current practice of routine prenatal testing presumes that the ethical questions are “settled”. The position statement adopted by the DSAQ challenges the dominant narrative of disability as disaster. It claims that people with Down syndrome should not have their potential or humanity written off. The stories and experiences of people with disability and their families is proposed as a support for those who are making decisions about the composition of our next generation. In particular, stories of surviving the “crisis” of disability should help both expectant couples and professionals to understand that the shock and grief following a diagnosis is not predictive of the long-term future for families. The statement suggests a series of guidelines which could ensure that testing is truly voluntary and informed, and that decision-making does not rely on myths. Most strongly it challenges the idea that reducing the incidence of Down syndrome through abortion is an honourable goal. It identifies this goal as a threat to the richness and diversity of our community. It asserts that people with disability should be seen as valuable and necessary members of our community. It is presented in the sincere hope that a future where people with disability are welcomed is possible.
REFERENCES


DOWN SYNDROME ASSOCIATION OF QUEENSLAND (DSAQ)
POSITION STATEMENT ON PRENATAL TESTING

The Mission of the DSAQ Inc is to support, advocate for, and empower Queensland children and adults with Down syndrome so that they can be valued and contributing members of the community, as is their right.

In support of our Mission Statement, the DSAQ believes:

- People with Down syndrome are valuable human beings who deserve respect and equality. All people with Down syndrome have the potential to lead full and rewarding lives and make an important contribution to our community.

- Down syndrome is not, in itself, a reason for termination. We recognize a valid role for prenatal genetic testing, but the primary goal of prenatal testing should not be to reduce the birth prevalence of Down syndrome.

- The use of prenatal testing to identify and abort fetuses with Down syndrome threatens the richness of our society. We value diversity.

- Prenatal testing should not be offered or promoted by using outdated information, negative stereotypes, prejudicial images or offensive terminology, which have the potential to stigmatize people with Down syndrome and increase fear of disability.

- Prenatal testing must be voluntary. The trend towards routine testing does not remove the obligation to obtain informed consent. Pregnant women and their partners must be advised they can refuse testing. They must not be subjected to pressure to undertake particular tests, to terminate a pregnancy or to make decisions in undue haste.

- Comprehensive and balanced information about Down syndrome and about the implications of testing must be available before and during the screening and diagnosis process. The information should include current information about opportunities for children and families including community based services, early intervention programs and opportunities for inclusive lifestyles.
Written information from the DSAQ and the opportunity to speak to parents of children with Down syndrome should be offered, particularly when a positive diagnosis is given.

The decision to continue or terminate a pregnancy belongs to the individual woman or couple alone. Counselling and support should be available, regardless of the individual choice.

Couples who continue pregnancies, following a positive diagnosis, should not be denied access to services or benefits.