



Genetic counselling and prenatal testing

By Dr Jan Hodgson and
Dr Melody Menezes

Photo of Isabella kindly provided by Renee Bell Studios in Port Macquarie, www.reneebell.com.au

Genetic counsellors play an important role in informing and supporting parents throughout the process of prenatal testing. We suggest readers also read the previous article, 'New technology for prenatal testing' by Marleen Susman, to understand the science behind prenatal testing.

Genetic counselling is a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions. (Resta, 2006)

If diagnostic testing reveals Down syndrome or another chromosome anomaly, parents are generally offered a choice about continuing or terminating their pregnancy. For many women and their partners this is a very difficult and emotional time, especially if they were unprepared for this outcome.

Role of the genetic counsellor

Genetic counsellors provide information and education about the full range of prenatal tests that can be offered, the conditions that are being tested for and the options that will be available following a diagnosis. Genetic counsellors practise in a non-directive manner to ensure that their clients receive relevant, non-discriminatory information about Down syndrome that avoids negative stereotyping. In addition, counsellors are able to provide emotional support for women and their partners throughout this often challenging time and beyond. Most importantly, they aim to facilitate informed decision-making at all stages of this process by talking with couples to ensure that they understand any risks associated with testing and encouraging them to consider whether the available tests will provide them with information that they wish to know.

Prenatal genetic counsellors can provide information and support for women and their partners who:

- have a family history of an inherited condition

- may be at increased risk for fetal anomaly, either because of advanced maternal age or as a result of a screening test
- receive a diagnosis of/are concerned about their risk for a fetal anomaly
- have questions about any of the above.

Access to clinical genetics services and genetic counselling varies throughout Australia according to location. To locate a genetic counsellor call your nearest public hospital or visit www.asgc.org.au

To learn more about prenatal tests that are available during pregnancy you may wish to read *Your choice, Screening and diagnostic tests in pregnancy*, www.mcrc.edu.au/Downloads/PrenatalTestingDecisionAid.pdf

Research

We are currently conducting a research study on prenatal testing in Victoria. This study will provide much needed data on women's experiences following the diagnosis of a fetal abnormality including the psychosocial impact and the supports available. We are strong advocates of promoting informed choices that would include:

- provision of accurate and non-discriminatory information about the conditions that are being tested for (including appropriate referral to support groups)
- allowing couples an opportunity to deliberate about the implications of a diagnosis for their pregnancy
- non-judgmental care and support for couples regardless of the pregnancy outcome.

continues on page 19

independent lives; not living with family or others of their choosing; no valued lives.

In 1994 I began working at a community based, family focused agency. Children were now mostly living with their families at home. School is compulsory for every child. Special schools are commonplace and integration is available for many. Local council provide some in home support and recreation services are becoming available for weekend and school holiday activities. A year later individual funding packages are becoming available, with case management to help people to identify what it is they want or need. \$2,000 a year! What'll that get someone with full time care needs? How will it help a young adult with Down syndrome to develop the skills they need to move into their own home one day; to work at a fulfilling job; to learn how to get about on public transport?

So now, in 2013, we are on the verge of a new era. The National Disability Insurance Scheme promises individual funding to provide services and supports that meet the needs of individuals. People with a disability and their families have had to fight for change and slowly change is coming. Now working for Down Syndrome Victoria I see that people with Down syndrome are certainly more visible in the community: many have jobs and there are people succeeding in the media and sporting arenas. People are getting married, participating in decision making in many fields, including politics, policy making and advocacy. The average life expectancy for someone with Down syndrome is now mid 60s. Medical intervention and greater societal expectations of what is a meaningful life have enabled people with Down syndrome to be valued, contributing members of our community.

Is that it? Should we sit back and congratulate ourselves on what we have achieved in 30 years and expect nothing more? Oh no! The fight goes on. Society is still not fully inclusive. Expectant parents are still told that people

with Down syndrome have a raft of medical issues, won't amount to anything and that there are no supports for families. Hope is still being destroyed.

But we know there is another side to the Down syndrome story and when we get calls from women who could be expecting a baby with Down syndrome, we try to restore hope. Parenting a child with Down syndrome isn't easy. There are challenges of course, it would be wrong to deny that, but I'm sure many of you will agree, there are many, many rewards too. Almost every day I hear of an achievement of someone with Down syndrome, somewhere in the world. It can be simple stuff or it can be huge.

Pablo Pineda is a 37-year-old Spanish man who was lead actor in the 2009 film *Yo, tambien* (Me Too), which explores some of the issues for people with Down syndrome, particularly in relationships. Pablo is also an academic with a bachelor of arts in educational psychology! Ashleigh recently performed with a local dance school, included in a team of dancers, where they all performed a hip-hop routine at the end of year concert. Dan has finally learnt to ride his bicycle without training wheels. In July 2012 Stuart got his driver's license. Ed, Chris, Robyn and Keith all live in their own units without live-in support staff. No, people with Down syndrome don't amount to much! Many people I know with Down syndrome have busier, more fulfilling lives than I do!

So I get excited, most days, about the opportunities for people with Down syndrome and celebrate how far we have come in 30 years and dream of the possibilities for the future.

Kerry Hands is the adult support manager at Down Syndrome Victoria.

from page 15

References

Hodgson, J., Gillam, L., Sahhar, M., & Metcalfe, S. (2010). Testing times, challenging choices; an Australian study of prenatal genetic counseling. *Journal of Genetic Counseling* 19(1), 22-37.

Resta, R., Biesecker, B. B., Bennett, R. L., Blum, S., Estabrooks Hahn, S., Strecker, M. N., et al. (2006). A new definition of genetic counseling: National Society of Genetic Counselors' task force report. *Journal of Genetic Counseling*, 15(2), 77-83.

Dr Jan Hodgson is a senior lecturer and the research coordinator for the master of genetic counselling course at the University of Melbourne and the Murdoch Childrens Research Institute.

Jan's PhD research, Testing times, challenging choices (2010), explored the process of prenatal genetic counselling and women's experiences of being at increased risk for fetal anomaly. Jan is on the board of management of DSAV. She can be contacted by email, jan.hodgson@mcri.edu.au

Dr Melody Menezes is a prenatal genetic counsellor at Monash Ultrasound for Women and an academic researcher at the Murdoch Childrens Research Institute. Her PhD research, Life on the front line, explored the personal and professional impacts on healthcare professionals working with couples making decisions during pregnancy. She can be contacted by email, melody.menezes@mcri.edu.au