

Discussing Prenatal Screening: The genetic counsellor's perspective

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What is genetic counselling?

A communication process that deals with the issues and challenges associated with the occurrence or risk of occurrence of a genetic condition in the family. This process involves the attempt to help the individual or family to:

- Comprehend the medical facts
- Appreciate the hereditary nature of the condition
- Understand the alternatives for dealing with risk
- Choose a course of action that is consistent with family goals, ethical and religious standards
- Make the best possible adjustment to the condition

(From: Baker DL *et al.* 1998. *A Guide to Genetic Counselling*)

Applying these principles to prenatal screening informed discussions – points to consider:

Educate: What is the patient's understanding of Down syndrome? – Are they misinformed? Do they have a personal experience with disability?

Tip: Ask your patient what they know about Down syndrome. Correct any misconceptions. Describe both ends of the spectrum of Down syndrome – low and high functioning (give examples) most people with Down syndrome will likely be in the middle of the spectrum.

Some common misconceptions from the Down Syndrome Association (UK)

http://www.dsa-uk.com/DSA_detBackground.aspx?ba=2

Myth	Fact
Only older mothers have babies with Down syndrome.	Although older mothers have a higher individual chance of having a baby with Down syndrome, more are born to younger mothers, reflecting the higher birth rate in this group.
People with Down syndrome cannot achieve normal life goals.	With the right support, they can. The vast majority of people with Down syndrome learn to walk and talk, and many are now attending mainstream schools and living full, semi-independent adult lives.
People with Down syndrome don't live very long.	Today, people with Down syndrome can look forward to a life of 60 years plus.
People with Down syndrome are always happy and affectionate.	We are all individuals and people with Down syndrome are no different to anyone else in their character traits and varying moods.

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Analyze: Screening versus diagnostic testing – explaining the difference:

Tip: “A screening test can tell you if your baby has a higher than average chance of having Down syndrome. For example 1 in 100 chance to have a baby with Down syndrome. Screening tests cannot tell for certain whether your baby truly has Down syndrome. A diagnostic test can tell you for certain if your baby has Down syndrome.”

Analyze: Concept of risk, everyone has a unique way of interpreting numbers.

Tip: Describe the risk figure in different ways. Example: 1/200 risk of Down syndrome

- One person in 200 people will have a baby with Down syndrome
- 0.5% chance to have a baby with Down syndrome
- Less than 1% chance to have a baby with Down syndrome
- 199 chances in 200 to have a baby who does not have Down syndrome
- Greater than 99% chance to have a baby who does not have Down syndrome

Alternatives to risk: - available testing options

Tip: Know the options in your area. Not sure? Call your local obstetrics department, genetic centre or laboratory.

For example if first trimester screening (FTS) is available in your area but chorionic villus sampling (CVS) is not - What if your patient's result is screen positive for Down syndrome and they want diagnostic testing? They may have to travel to another city to have CVS or wait 2-3 weeks to have an amniocentesis – how might your patient feel in this situation?

Nuchal translucency not available in your area and your patient would like the highest detection rate possible – consider Serum Integrated Prenatal Screening.

Facilitate informed-decision making:

Tip: Discuss possible scenarios and potential outcomes.

Example: Does your want to know if their baby has Down syndrome before birth? If the answer is yes:

“If you have prenatal screening and the result is screen positive and you went on to diagnostic testing and found out for sure that your baby had Down syndrome – what would you do?” *Consider how your personal beliefs might influence how you discuss these options:*

- Continue the pregnancy
- Adoption
- Abortion

Would your partner and family support your decision?

How might your decision affect your relationship with your partner, children and family?

How do you think you might feel about this decision in 6 months, 1 year, 10 years?

Would you want to meet a family who has a child with Down syndrome – would that help your decision?

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Sensitivity: to ethnic and cultural diversity, potential ethical issues.

Tip: Patients from different ethnic/cultural backgrounds consider - What are your patient's cultural views on pregnancy, prenatal testing, Down syndrome, disability, etc. How do they differ from your own? Learn from your patients!

Sensitivity: language choices

Tips: Think before you speak – some suggestions from the Down Syndrome Association (UK) http://www.dsa-uk.com/DSA_detBackground.aspx?ba=2

Common language	Better choices
Mongol/retard	Person/baby/child with disability
Suffers from OR victim of Down syndrome	Has Down syndrome
A Down's baby/child/person	A baby/child/person with Down syndrome or who has Down syndrome
Retarded/mentally handicapped/backward	Learning disability
Disease/illness/handicap	Condition or genetic condition
The risk of a baby having Down syndrome (in relation to prenatal screening and probability assessments)	The chance of a baby having Down syndrome

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Resources:

Canadian Association of Genetic Counsellors: <http://www.cagc-accg.ca> or 905-849-8299
List of medical genetic centres located across Canada.

Canadian Down Syndrome Society: <http://www.cdss.ca> or 1-800-883-5608
More information about Down syndrome with links to local parent support groups.

The Genetics Education Project: <http://www.mtsinai.on.ca/FamMedGen/>
Resource for your practice.

The Genetics Home Reference - Your Guide to Understanding Genetic Conditions:
<http://www.ghr.nlm.nih.gov>
An excellent site to learn more about genetics including Down syndrome, trisomy 18 and more.

March of Dimes: <http://marchofdimes.com/>
Excellent source of information about pregnancy including common prenatal tests, complications during pregnancy, things to avoid during pregnancy, birth defects etc.

Motherisk: <http://www.motherisk.org/> or 416-813-6780 or 1-877-327-4636
Information service for women who have been exposed to environmental agents, drugs and alcohol during pregnancy.

Ontario Multiple Marker Screening (MMS) Program: <http://www.lhsc.on.ca/programs/rmgc/mss/>
Information on prenatal screening tests.

S.O.F.T. Support Organization For Trisomy 18, 13 and Related Disorders:
<http://www.trisomy.org> or 1-800-716-7638 or 585-594-4621
More information about trisomy 18.

Spina Bifida and Hydrocephalus Association of Canada:
<http://www.sbhac.ca> or 1-800-565-9488
More information about spina bifida.