

RIDING THE ROLLERCOASTER:

**The challenges we
universally face**

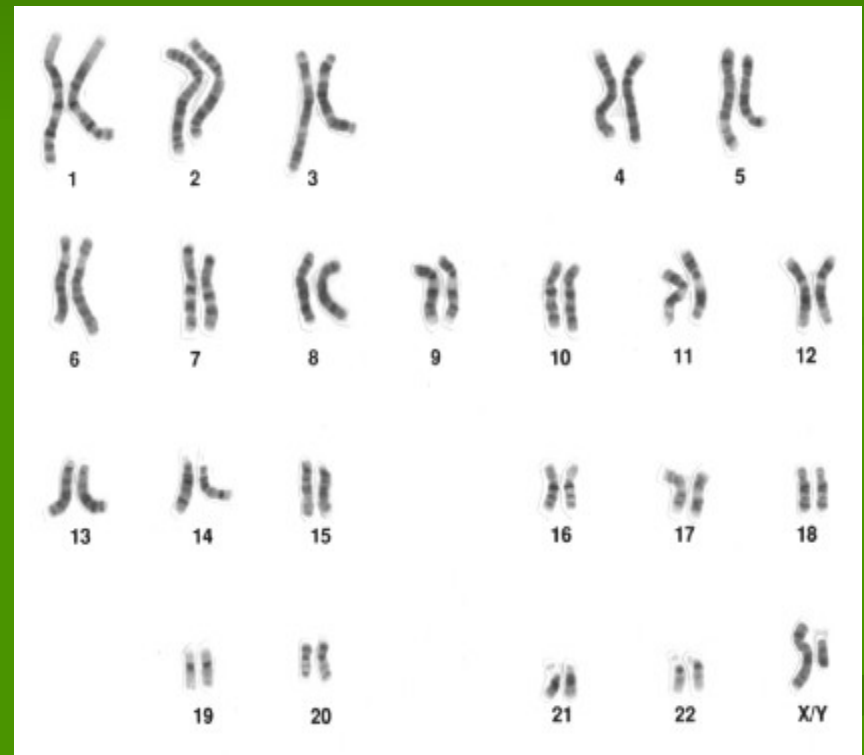
Presented by Kelly Woods

OUTLINE:

- * Define and discuss genetic testing
 - * Share personal family stories
 - * Discuss what the literature says
 - * Consider possibilities for improvement
- Feedback from NZ Bioethics Council
forum, 'Who gets born?'
- * Personal reflections

What is genetic testing?

- Genetic testing is the examination of a person's DNA (deoxyribonucleic acid) usually via a blood sample.
- Genetic testing can show changes to the numbers, and/or arrangements of chromosomes, or inactivations of particular genes.



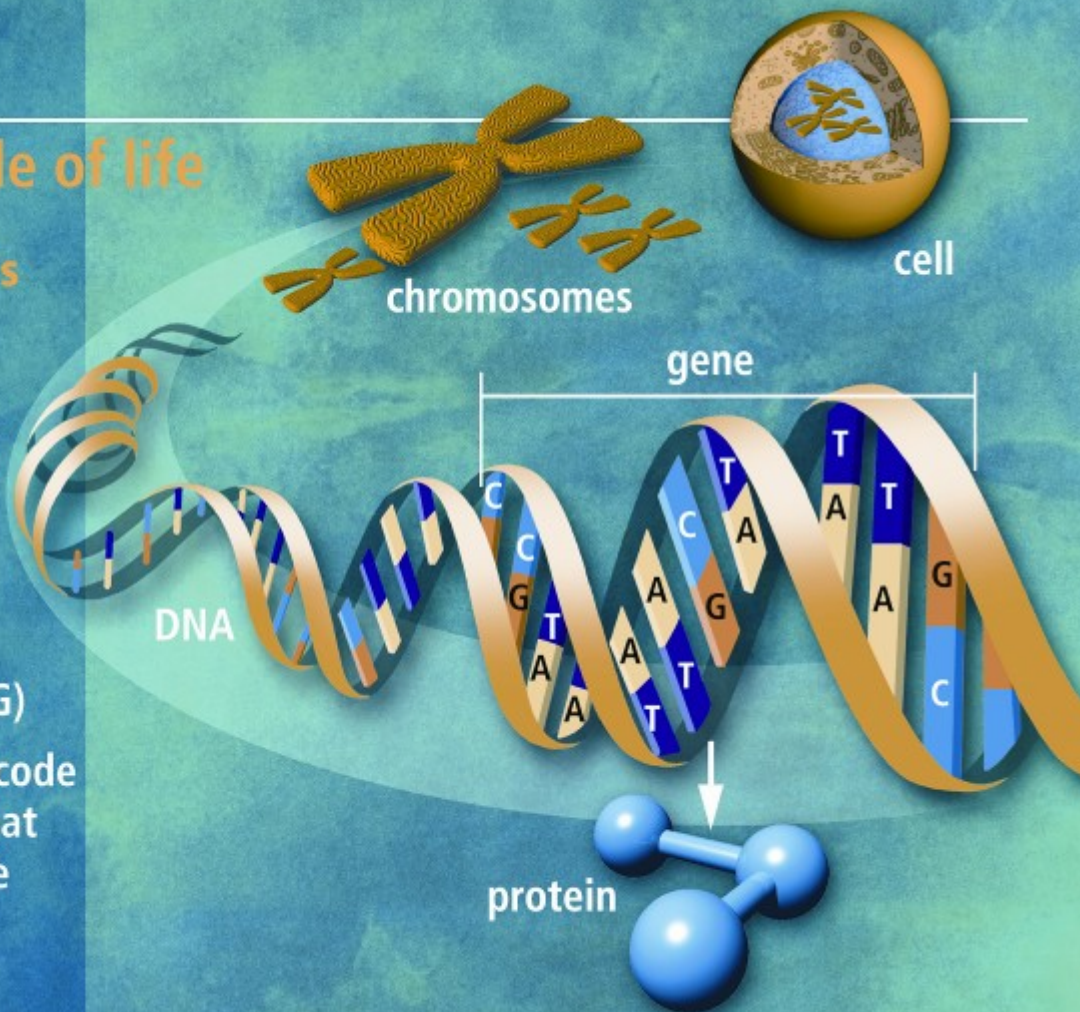
DNA

the molecule of life

Trillions of cells

Each cell:

- 46 human chromosomes
- 2 m of DNA
- 3 billion DNA subunits (the bases: A, T, C, G)
- 80,000 genes code for proteins that perform all life functions



Reasons for genetic testing:

- Family history of a medical condition
 - Pre birth test that indicates a genetic condition (e.g. ultrasound)
 - A child born with several abnormalities or indications of a genetic disorder
 - Women >35yrs who are pregnant (e.g. amnio.)
 - Women who experience recurrent miscarriages
 - Family history of cancer (particularly from a young age)
 - Anybody who is uncertain re: their genetic risks
- Northern Regional Genetic Service, ADHB, 2008

What to expect?

- Concerns are discussed
- Family medical history detailed
- Possibility of further investigations or information may be required
- Examination of child and/or adult
- Genetic testing may or may not be carried out on first visit
- Documentation of what has been diagnosed and/or discussed
- Provision of further information and support
- Northern Regional Genetic Service, ADHB, 2008

WHAT HAS BEEN THE REALITY OF GENETIC TESTING FOR FAMILIES?



**“Nothing in life is to be
feared, it is only to be
understood.”**

Madame Curie



What does the literature say?

- Around the time of diagnosis, key needs have been identified:
 - Information regarding the diagnosis
 - Educational and social service information
 - Emotional support from professionals
 - Contact with informal and formal social networks/support groups
- Rahi, J.S., Manaras, I., Tuomainen, H., & Hunt, G.L. (2004). Meeting the needs of parents around the time of diagnosis of disability among their children: evaluation of a novel program for information, support, and liaison by key workers. *Pediatrics*, 114(4), 477-482.

THE INDIVIDUAL EXPERIENCE:

- Grief: a personal and emotional reaction to a significant loss, a loss of dreams and ideas, hopes and expectations
 - Dowling, C., Nicoll, N., & Thomas, B. (2006). *A different kind of perfect: Writings by parents on raising a child with special needs*. Boston: Trumpeter.
- Initial stages of grief – shock, denial, anger, resignation
 - Whiteman, N.J., & Roan-Yager, L. (2007). *Building a joyful life with your child who has special needs*. Philadelphia: Jessica Kingsley Publishers.

IT TAKES A VILLAGE TO RAISE A CHILD

African Proverb



COMMUNITIES OF PRACTICE

- “A group of people who come together to learn from each other by sharing knowledge and experiences about the activities in which they are engaged...”
- Gradually shared stories and insights emerge that contribute to a common store of knowledge that accumulates over time...
- Trust, empathy and reciprocity are the building blocks for relationships that unite members.”
- Preece, J. (2004). Etiquette, empathy and trust in communities of practice: Stepping-stones to social capital. *Journal of Universal Computer Science*, 10(3), 294-302.

INFORMAL SUPPORT

- “...a life enriching togetherness, the core of which is natural human caring.”
 - Lindbland, B.M., Holritz-Rasmussen, B., & Sandman, P.O. (2007). A life enriching togetherness – meanings of informal support when being a parent of a child with a disability. *Scandinavian Journal of Caring Sciences*, 21(2), 238-246.
- “Parents of children with special needs are uniquely qualified to help each other. The challenge is to ensure that health professionals are aware of the potential benefits of parent-to-parent support and provide parents with information about appropriate local organisations.”
 - Kerr, S.M., & McIntosh, J.B. (2001). Coping when a child has a disability: Exploring the impact of parent-to-parent support. *Child: Care, Health and Development*, 26(4), 309-322.

**Like the body that is made up
of different limbs and organs,
all mortal creatures exist
depending on one another**

Hindu Proverb



Toi te taiao

THE BIOETHICS COUNCIL

- Is a ministerial advisory committee:
- Providing independent advice to Government re: biotechnological issues
- To promote and participate in public dialogue on the cultural, ethical and spiritual aspects of biotechnology
- To provide information on the cultural, ethical and spiritual aspects of biotechnology
- www.bioethics.org.nz

Who Gets Born?

- A report on the cultural, ethical and spiritual aspects of pre-birth testing (June 2008)
- Pre-birth testing – encompasses all tests that women and/or their unborn babies can have either before or during a pregnancy (e.g. ultrasound, amniocentesis, chorionic villus sampling)

Who Gets Born? Introduction

- DELIBERATIVE APPROACH
- 6 face-to-face ‘framing days’ (July & August 2007). Including a hui for Māori and a Pacific fono (56 participants)
- Four prominent themes emerged
- 18 face-to-face deliberative events . Including 4 Māori hui and a Pacific fono (256 participants).
- Online deliberations (November 2007 – February 2008). Use of moderator.
- Evaluation
- www.bioethics.org.nz

WHY DELIBERATION?

- Ordinary citizens have the right to express views
- Ordinary citizens can offer insights and knowledge that complement and enlarge the knowledge of experts
- People need to explore and compare a range of viewpoints or approaches
- People talk *through* a problem, rather than talk *about* it
- Can increase trust between public and decision makers/government agencies
- Can produce unique forms of non-expert or ‘socially constructed’ knowledge
- Can turn personal opinion into sound public judgement
- Is dynamic and interactive

THE FOUR THEMES:

- My choice, my right
- Life is a gift
- Tāngata Whenua
- Knowledge is power
- www.bioethics.org.nz



WHAT WAS HEARD

- Valuing human life & diversity
- The difficulty of decision-making – role of parents, others and the scope of decisions
- Information about pre-birth testing
- Access to pre-birth testing
- Concern for families of children with disabling conditions
- Concern for the future
- Personal experiences of pre-birth testing
- What people wanted

RECOMMENDATIONS

- 11 recommendations
- Recommendations addressing:
 - Decision making processes
 - Appropriate and holistic information
 - Further research to ascertain why some women choose not to pre-birth test
 - Provision for an adequate national genetic service
 - Identifying barriers to antenatal care
 - Improving coordination of disability services
 - Further research re: PGD for creating embryos that tissue-match to a sick sibling
 - Insufficient reasons to prohibit PGD for sex selection
- www.bioethics.org.nz

FOOD FOR THOUGHT

- “It is crucial that prospective parents are offered both information about disability and the opportunity to explore the values, desires, fears and dreams that enter into deciding what to do with prenatal genetic information.”
- “The first, crucial step in helping parents achieve truly informed consent...is to give providers access to good information about what disability is really like for children and their families. Education about life with disability is still too rarely offered to those who deliver genetic information.”
- Parens, E., & Asch, A. (1999). The Disability Rights Critique of Prenatal Genetic Testing: The Hastings Center Report

MORE FOOD...

“Perhaps our best hope is that good information about disability will permeate our culture more thoroughly - more television and radio shows, more plays, more newspaper articles that accurately portray the lives of people with disabilities.”

- Parens, E., & Asch, A. (1999). The Disability Rights Critique of Prenatal Genetic Testing: The Hastings Center Report



FINAL THOUGHTS...

- Genetic testing is a complex process
- Whether a genetic test identifies an abnormality or not, it is clearly a distressing time for families
- The type of information and the way in which it is presented to families can often be ambiguous
- The amount of information a family can process during and after genetic testing is challenging, and can have long lasting effects

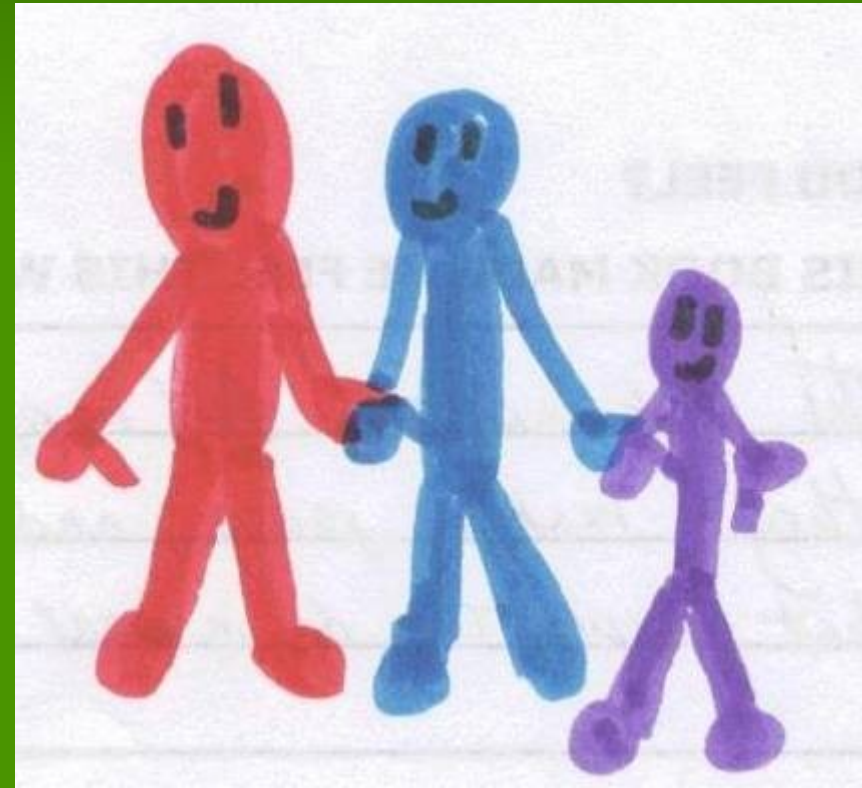
How can we strive for excellence?

- Respect
- Empathy
- Partnership
- Trust



So – what have I done about it?

- Become proactive
- Share knowledge through the ‘lived experience’ (with specialists and families)
- My readiness to connect with prospective parents via genetic services (if/when my daughter’s chromosomal anomaly is diagnosed for another child)



TIME FOR REFLECTION

“Inspiration comes to us slowly and quietly...prime it with a little solitude.”

— Brenda Ueland



Thank you

- Thank you to the families who found the courage to tell their stories
- Thank you for listening