



WHAT CAN
WE DO
FOR YOU?

WHAT CAN A GENETIC COUNSELLOR DO FOR YOU?

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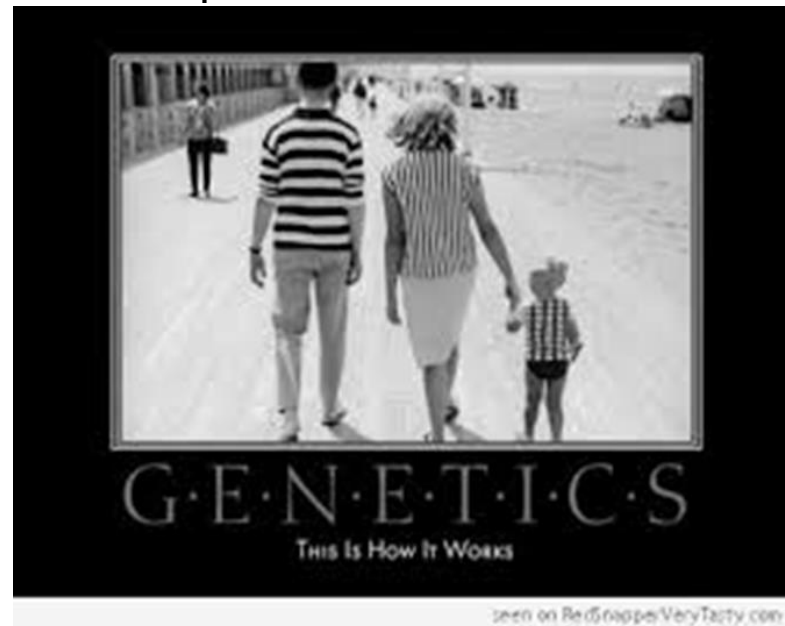
OVERVIEW

1. What is genetic counselling?
counsellor bring to a patient interaction?
2. Process of genetic counselling
3. History of profession
4. Training of genetic counsellors
5. How does a genetic counsellor differ from a geneticist?
6. What does a genetic
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WHAT IS GENETIC COUNSELLING

National definition:

- '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'



PROCESS OF GENETIC COUNSELLING

Process integrates:

- **Interpretation** of family and medical histories to assess the chance of disease occurrence or recurrence
- **Education** about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources and research
- **Counselling** skills used to promote informed choices in view of risk assessment, family goals, ethical and religious values
- **Support** to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder

HISTORY OF GC PROFESSION

Profession established in Australia since 1987

Genetic counselling has been undertaken for 50+ years

First course Sarah Lawrence College, New York State, 1969

Now an international allied health specialty with genetic counsellors in countries such as (not limited to):

- USA
- UK
- Australia
- Philippines
- China
- Japan
- India
- Lebanon



WHAT TRAINING DOES A GC HAVE

Historically GC's entered the profession from different backgrounds

- Social work
- Nursing
- Cytogenetics/research scientists (laboratory)
- Pure counselling

Since 2011, people entering profession require two year Clinical Masters

- Includes a research thesis

Trained in biology, genetics, education, counselling, research

Full Genetic Counsellor certification is obtained via case reports and literature reviews whilst working in the profession

HOW DOES A GC DIFFER FROM A GENETICIST

The act/process of genetic counselling is done by both geneticist and GC

GC's specifically trained to provide support, education and counselling

Geneticists are medically trained professionals specialising in genetics

Often practice as a team (co-counselling)

GC's can practice alone once medical issues have been addressed

WHAT GC'S BRING TO A PATIENT INTERACTION

Genetic counsellors are uniquely placed to provide psychosocial support to patients

Patients are often experiencing strong emotions after genetic diagnosis (or lack thereof) in themselves or their child

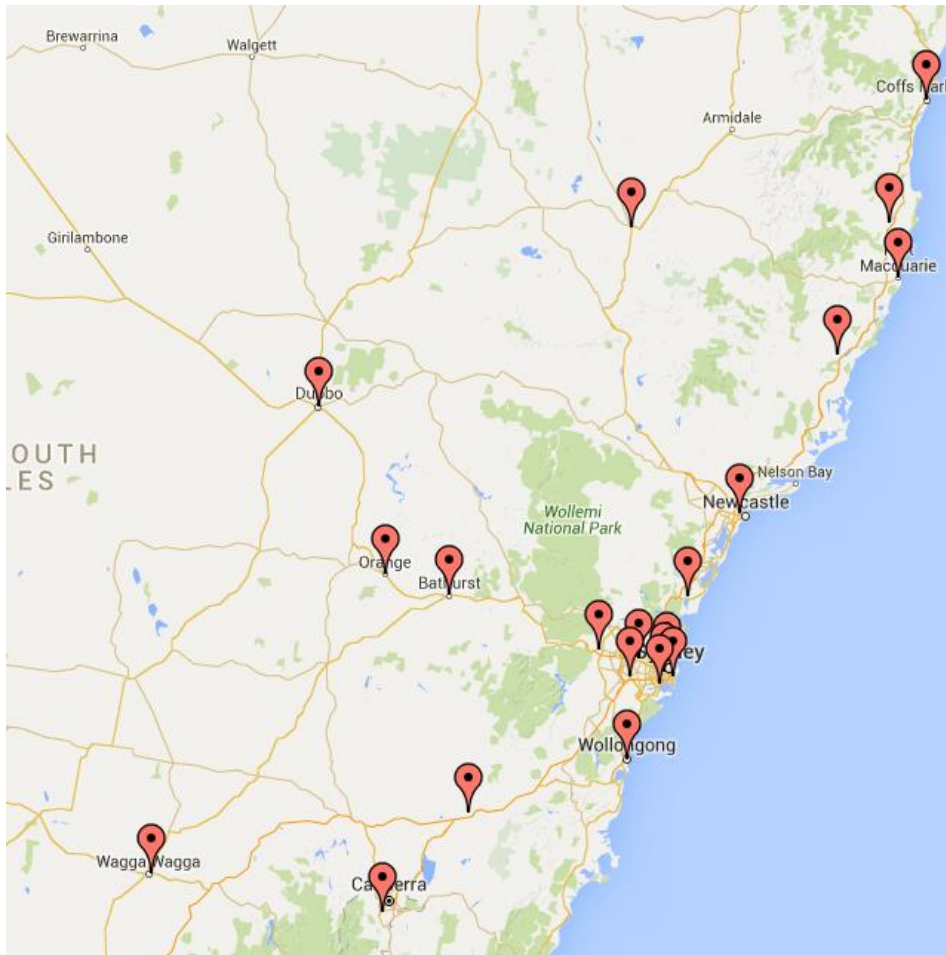
Can provide supportive counselling to cope with feelings such as grief, anger, guilt, shame

Also able to assess if require long term support and refer to on psychologist if required

GENETICS SERVICES IN AUSTRALIA



GENETICS SERVICES IN NSW



- Where feasible, people will be referred to their closest genetics service
- This helps facilitate both billing for testing, and also hopefully also more convenience for patients
- Some rural patients are referred to metropolitan paediatric hospitals (Sydney Children's Hospitals or Children's Hospital at Westmead) and referrals to these two locations are considered if attending for admission

WHAT'S SO SPECIAL ABOUT A GENETIC COUNSELLOR?

Consequences of initiating genetic testing

- Interactions with wider family
- Time
- Unique set of skills to deal with family dynamics

Impact of a genetic result

Giving bad news

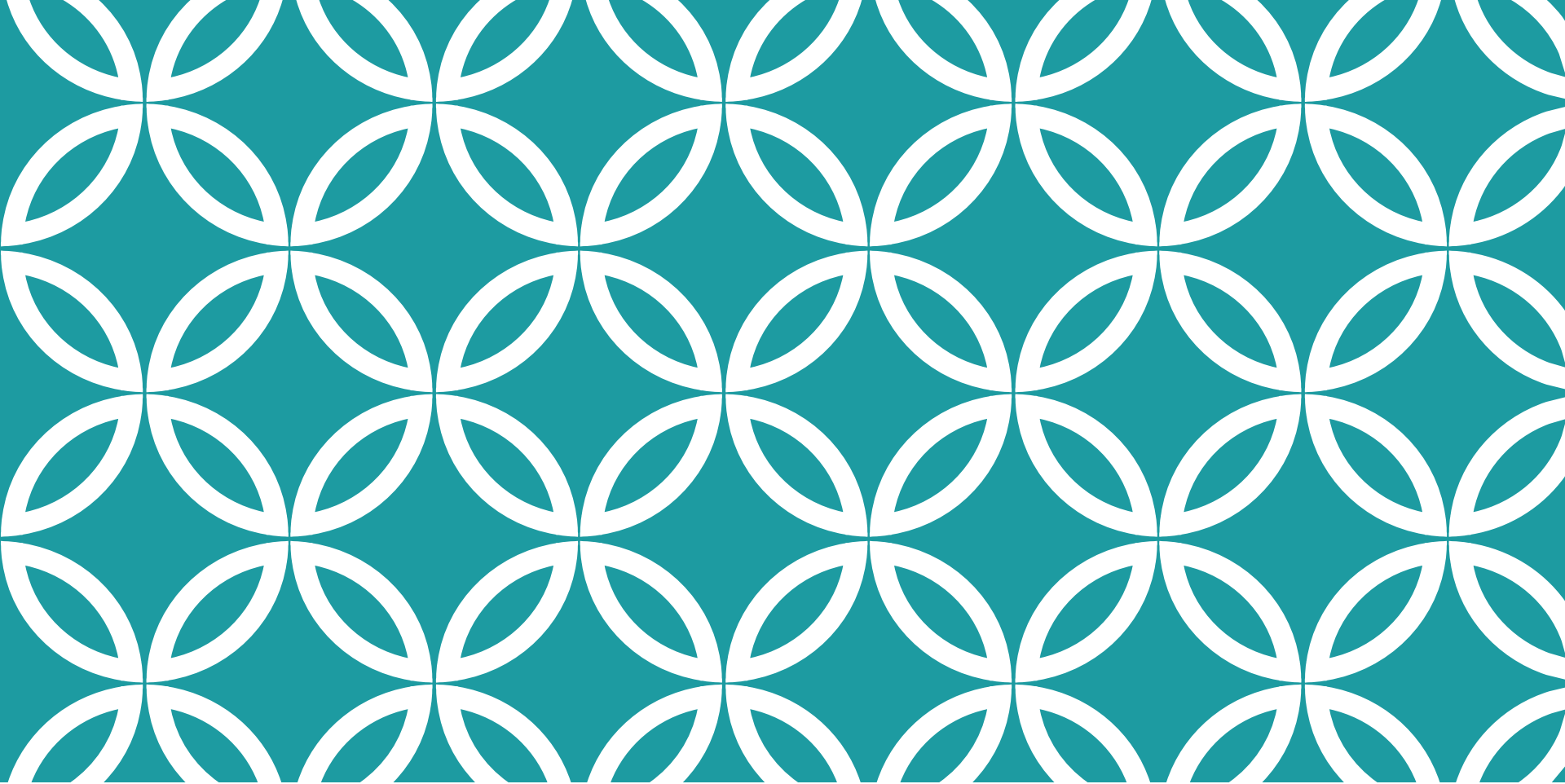
DNA MAPPING
IN PROGRESS

Chris Madden

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THE G-NOME PROJECT



CASE EXAMPLES



“GENETIC INTERPRETATION”

Genetic counsellors help with providing easy to understand language for complex genetic concepts

- Often use analogies to simplify complex medical jargon
- Real world examples (Celebrities with genetic conditions!)
- Can provide explanations or clarifications for information provided by geneticist / doctors / internet / social media
- Will explain genetic test results in simple language with education and information regarding:
 - Test process
 - Limitations
 - Possible outcomes
 - Uncertainty vs certainty
 - Timeframes

“GENETIC INTERPRETATION”

Example 1:

- Parent of child with severe intellectual disability is a member of a Facebook group (all members are parents of children with changes in the same gene *ZEB2* causing condition called Mowat–Wilson Syndrome)
- Parent rings the genetic counsellor and advises that parents have been discussing that there is a reduced life expectancy reported in an online article, and that affected patients only live until 40
- Advised parent that this information is incorrect, there are documented cases of patients in their 60's
- No reason to expect that there would be a life limiting effect with this condition, other than if individual has a severe health complication
- Explained that much information that is available on the internet regarding this issue is biased or incorrect
- This particular article gets circulated on this Facebook group every couple of months to year and creates concern

“GENETIC INTERPRETATION”

Example 2:

- Patient diagnosed with a change in a gene in a gene called *SCN1A*
- Associated with multiple phenotypes (clinical presentations) ranging from mild to severe epilepsy
- When parents “google” this gene, information can be overwhelming and scary, and not necessarily relevant to their child
- Provide guidance on relevant sites to visit and accurate and relevant information for their family



FAMILY RELATIONS

Can help you talk to your family about the diagnosis

- Genetic conditions may have implications for the wider family
- Can write letters, or help facilitate testing of the wider family
- Can help with discussions of other family members about results
 - Means you do not have to be the genetics expert

Example

- Patient with suspected mitochondrial disease (inherited through maternal (mother)'s line)
- Other family members concerned, and mother felt not able to explain herself – with mother's permission, spoke to several family members on the phone including mother's mother (grandmother) and her sister (aunt)
- Facilitated testing in all family members and provided accurate information

FAMILY RELATIONS

Also able to provide advice about future risks and options for family planning

Able to facilitate testing in future pregnancies if molecular (DNA) cause known in affected person

- CVS
- Amniocentesis
- Pre-implantation genetic diagnosis

GC PROFESSION INTO THE FUTURE

Diversifying into specialty areas

- Laboratory GCs
- Cardiac/Prenatal/Cancer/Specialty clinics
- Private practice
- GP clinics

Workload increasing with the introduction of whole exome and genome sequencing

Will be involved with result interpretation

Constantly changing profession

Precision Medicine

INFORMED CONSENT IN GENETIC TESTING

More than just co-signing consent form

Genetic testing, particularly panel testing/exome sequencing require informed consent – testing increasingly complex

Genetic counsellors specifically trained to explain process of testing, possible outcomes, limitations of testing, explore specific issues and elicit informed consent

HOW TO GET US INVOLVED

Just ask!

We are happy to help

Referrals will be triaged by GC/geneticist

Waitlists are lengthening due to increasing demand

We are happy to help, but are also a limited resource, so triage occurs based on clinical urgency and resources available

Questions?



HETEROZYGOATS

Just allele uneven.