



# PATHS TO A GENETIC DIAGNOSIS

*The Role of Genetic Counsellors*



Everybody has their journey.

Each person has their story.

Genetic testing is not always straightforward and finding a diagnosis is not always easy.

# *Different paths to a diagnosis in 2019.*

*Pathways to a diagnosis can be different, some have a linear path, some have an odyssey and some are still undiagnosed.*

Prenatal diagnosis

Newborn screening

Diagnosis in childhood: Complex condition, Intellectual disability

Clinical signs/symptoms → unknown diagnosis → Genomic sequencing  
→ genetic condition identified or still unknown or uncertain

Diagnosis in adulthood

- Cancer
- Cardiac Condition
- Neurological condition

Incidental finding

Clinical Diagnosis → Genetic Test

Direct to consumer genetic testing

- Ancestry testing
- Proactive screening

Sharing family information

Cascade screening of relatives

# A **genetic diagnosis** is important because it helps to...

1

End the diagnostic  
journey and minimize  
additional tests



2

Understand  
recurrence risk  
for families



3

Provide a roadmap  
for the future



4

Identify opportunities  
to network with  
similar participants

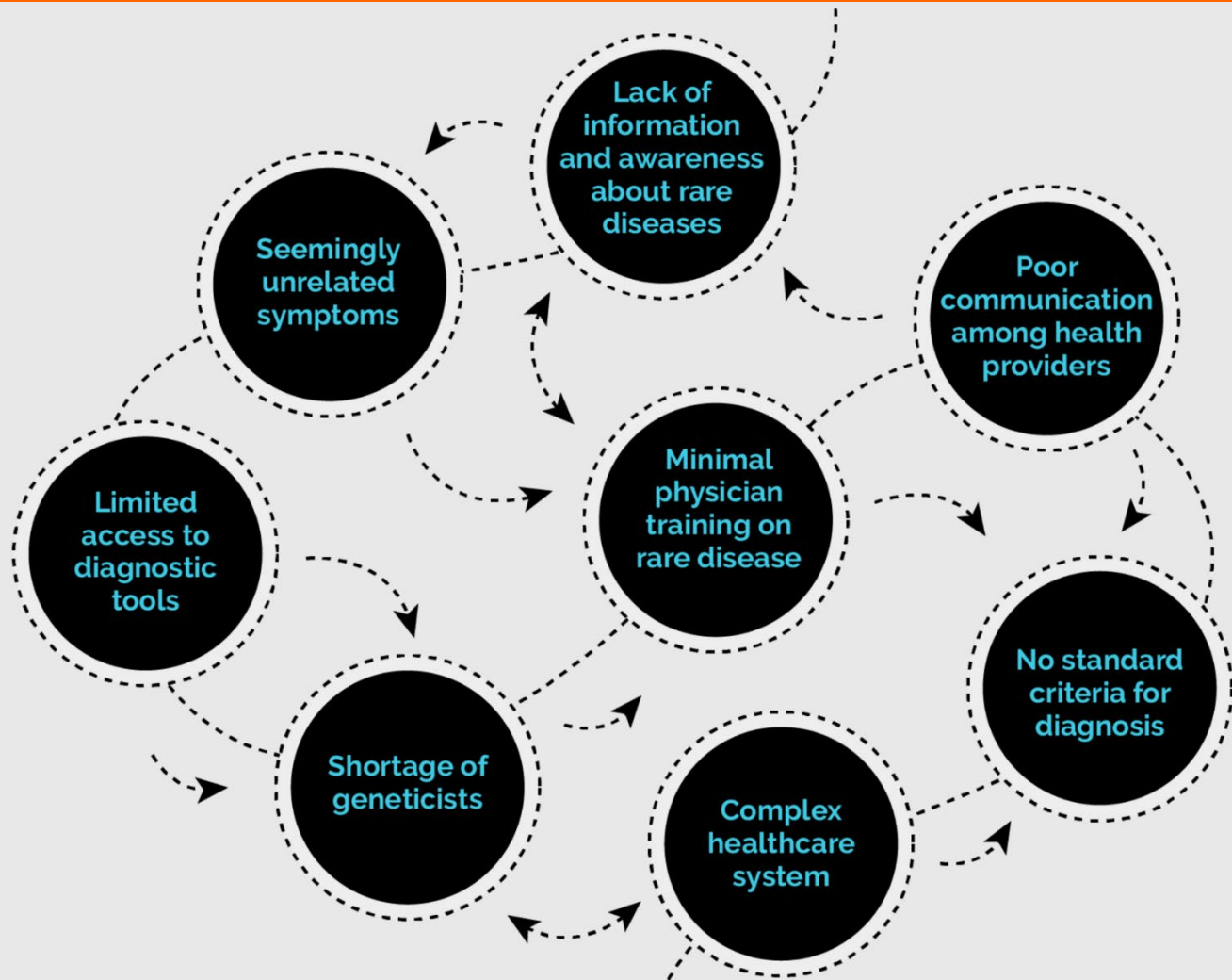


5

Learn about clinical  
trials and/or  
treatments specific  
to your diagnosis.



*For many families, the search for a diagnosis can become an odyssey.*





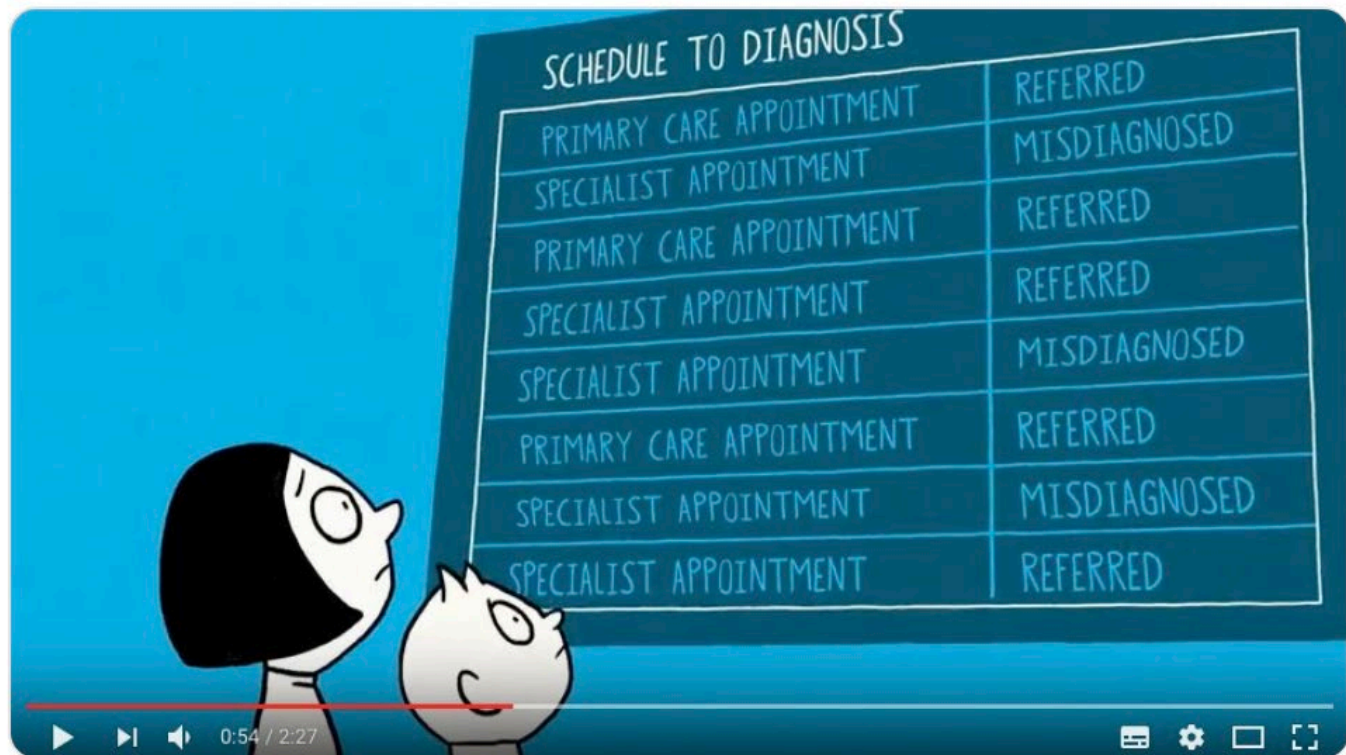
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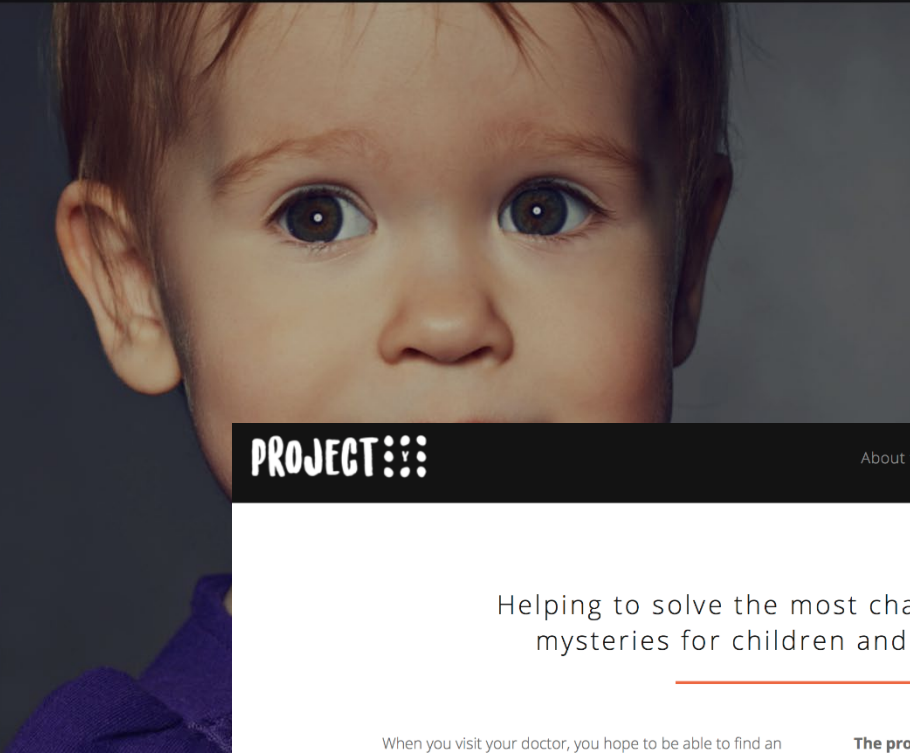
The #diagnostic odyssey takes a huge toll on families. Raising awareness remains crucial for #RareDisease [bit.ly/2flproO](https://bit.ly/2flproO)





# 500,000 children in Australia are living with a rare disease.

Project Y, lead by Dr Gareth Baynam, is a groundbreaking initiative that brings together specialist doctors to provide an answer and a diagnosis to the thousands of families who desperately need them.



## Helping to solve the most challenging medical mysteries for children and young adults.

When you visit your doctor, you hope to be able to find an answer, a diagnosis. An answer that helps doctors guide your treatment and reduces uncertainty for you and your family.

But for too many of our children and young adults, a diagnosis comes late or not at all. This is often because the disease is rarely seen, or perhaps it has not even been named. Many of these 'rare diseases' occur in the population at very low rates, but cumulatively rare diseases are common.

There are many expert doctors doing a fantastic job in diagnosing and managing the children and young adults they see. Increasingly, there are new approaches that improve the chance of reaching a diagnosis.

Project Y supports these new approaches, one of which is the Undiagnosed Diseases Program (UDP), WA. Established in 2016, the UDP incorporates a team of doctors from a broad range of specialties working together in partnership with researchers.

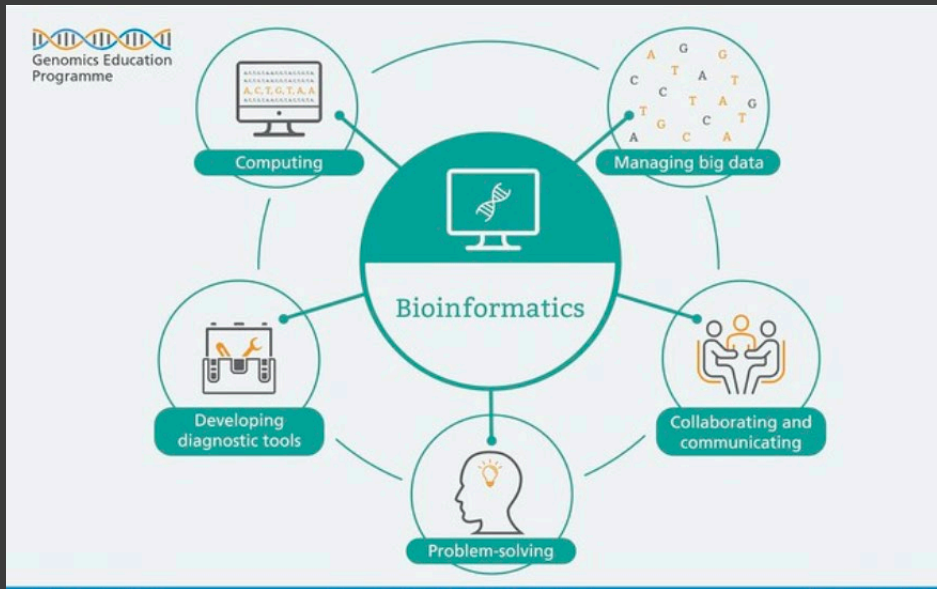
**The program sees one child (under the age of 16)  
and one young adult (aged 16-25 years) per month.**

### Referral criteria

- 6 months of age to 25 years old
- Chronic, complex and multisystem diseases
- Multiple specialist assessments and frequent hospital admissions
- Referred by specialist doctors working with Princess Margaret Hospital/ Perth Children's Hospital, King Edward Memorial Hospital and Sir Charles Gairdner Hospitals (i.e. referrals from other hospitals and/or specialists are not currently accepted).

**Further information on referral criteria can be found [here](#).**

*It takes an average of 5 years for a patient with a rare disease to get a diagnosis  
... but things are changing.*



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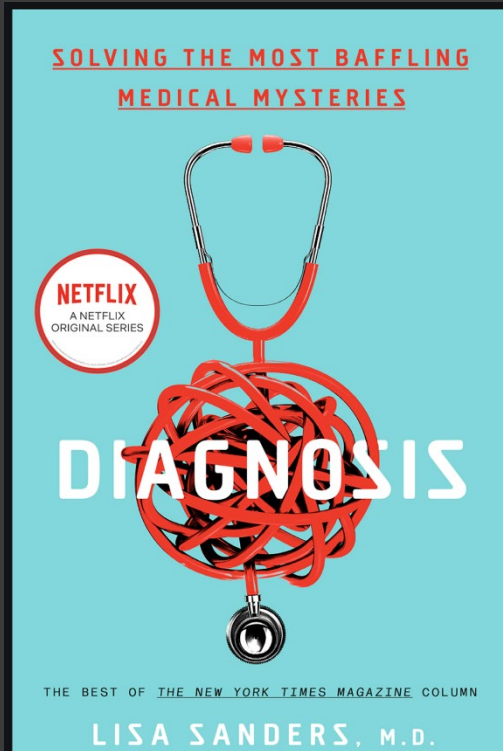
NEWS

### Faster genetic diagnosis for critically ill babies and children in Australia

Rapid genomic testing will soon be available to critically unwell Australian babies and children with suspected genetic conditions, as part of four new national genomic research studies launched by Australian Genomics.



*It takes an average of 5 years for a patient with a rare disease to get a diagnosis  
... but things are changing.*



# Useful contacts and help on your diagnostic pathway

## Local Genetic Services (Publicly funded)

- Waiting list may exist
- GP Referral needed
- Clinically indicated genetic testing covered by local health district

[www.genetics.com.au](http://www.genetics.com.au)

## Private Genetic services now available

- Clinical Geneticists
- Genetic Counsellors
- Often have shorter waiting time
- Can facilitate other testing not covered in public hospitals

Keep connecting:

[www.hgsa.org.au](http://www.hgsa.org.au)

[www.australianclinicaltrials.gov.au](http://www.australianclinicaltrials.gov.au)

Facebook groups

[www.geneticalliance.org.au](http://www.geneticalliance.org.au)

# *What is genetic counselling?*

The goal of genetic counselling is to help you learn more about the causes of genetic conditions and how they affect you.

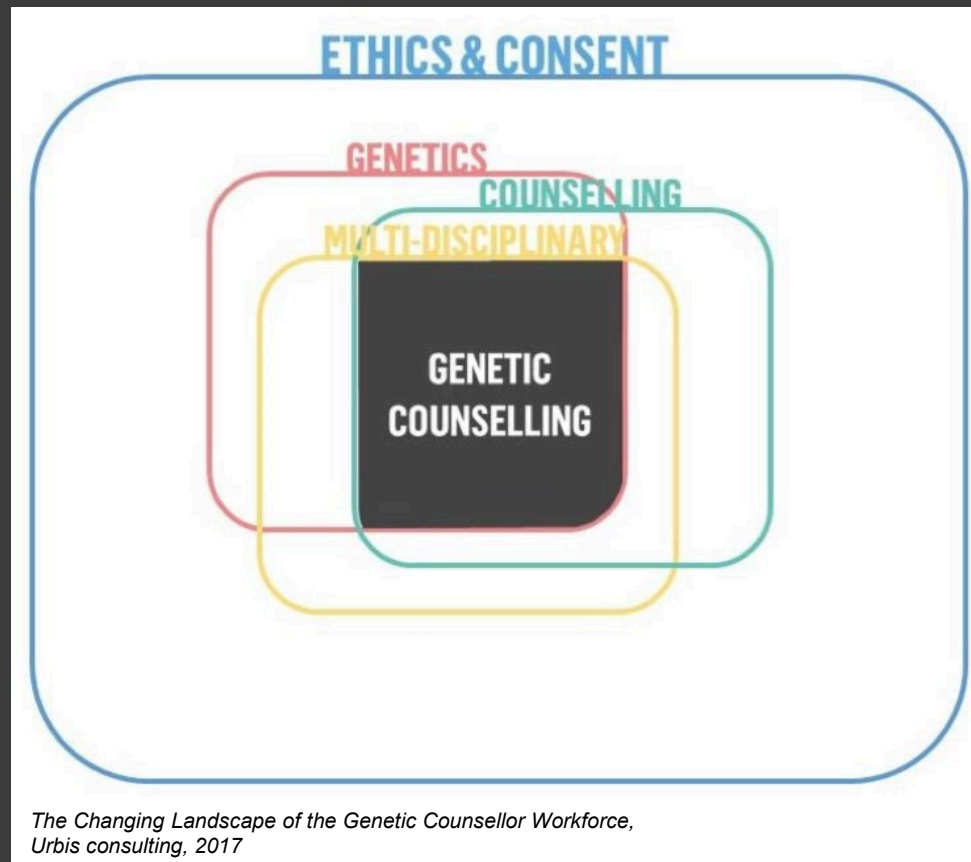
*A genetic counselling session is a conversation.  
Your input is very important to the session*

Helping

Guiding

Facilitating

Supporting



## *Genetic counsellors can:*

- Review your family and medical histories.
- Explain how genetic conditions are passed down in families.
- Figure out if you or your family members are at risk for disease.





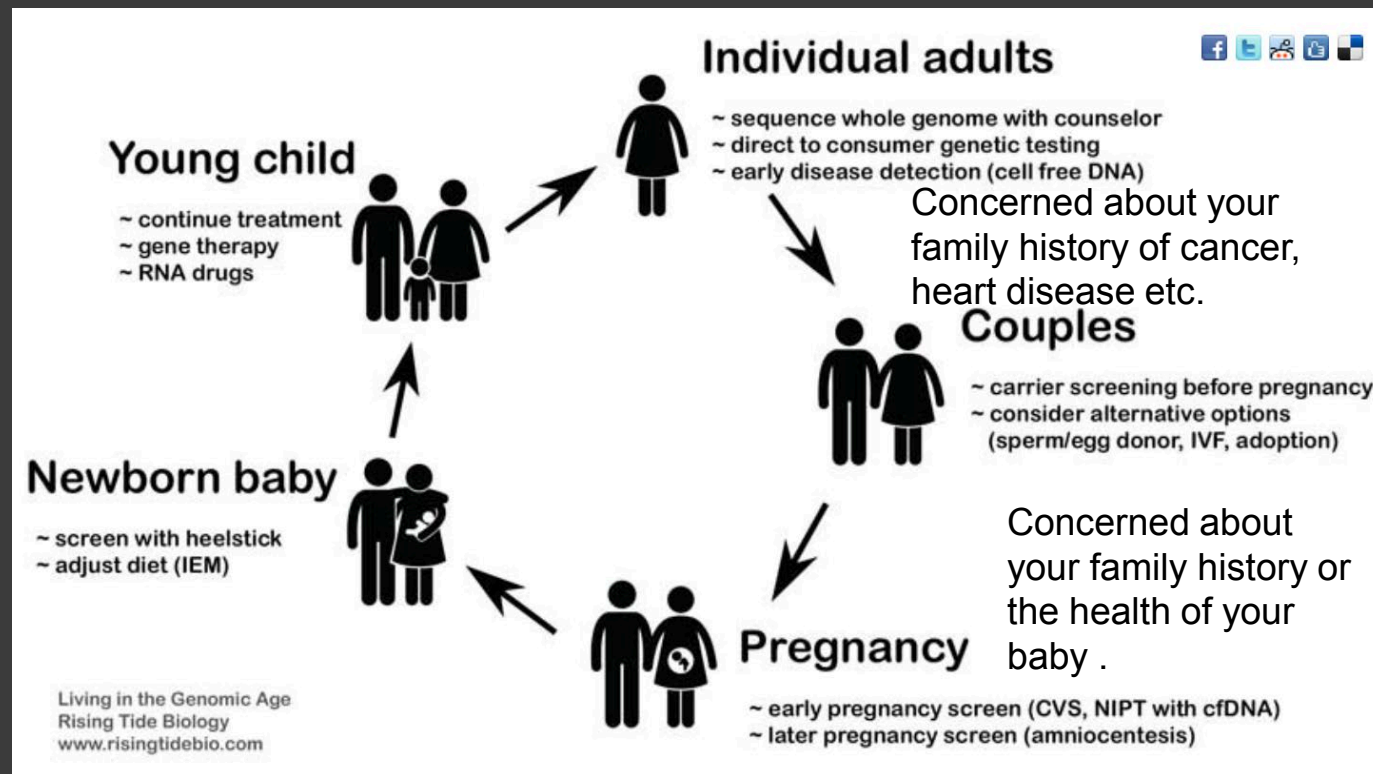
## *Genetic counselors can:*

- Find and give you information about genetic conditions.
- Offer guidance to help you make informed choices or life plans.
- Provide information about testing options and help you decide what is best for you and your family.
- Help you find referrals to medical specialists, advocacy and support networks, and other resources.

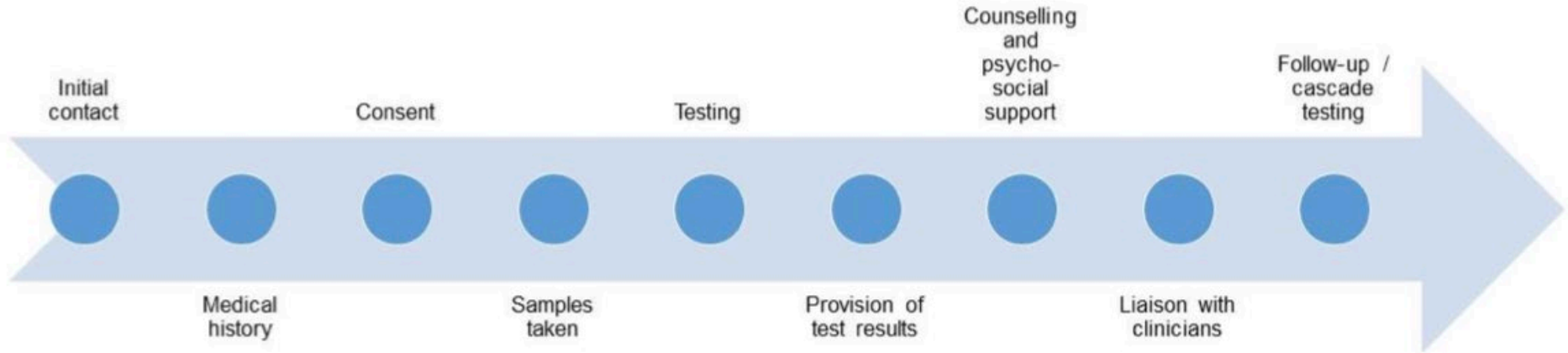


# Why might you see a Genetic Counsellor?

Living in the genomic era .... Genomics is more available than ever before and reaches across our entire life.



# What is the process of Genetic Counselling?



*The Changing Landscape of the Genetic Counsellor Workforce, Urbis consulting, 2017*

*Be empowered.*

*Questions you might ask your Genetic Counsellor.*

Does the genetic condition in question run in families?

If I have a genetic condition, are my family members at risk of getting it?

Is any kind of genetic testing available? If so, what are the benefits and limitations of the testing? Will I need to pay for it?

What are the benefits and limitations of testing?

How can I explain this to my family?

Am I exposing my family to any discrimination from testing?

If my family member has a genetic condition, might I get it?



Everybody has their journey.

Adaptation is a process, it takes time.

Genetic Counsellors are here to walk alongside you.

Thank you.