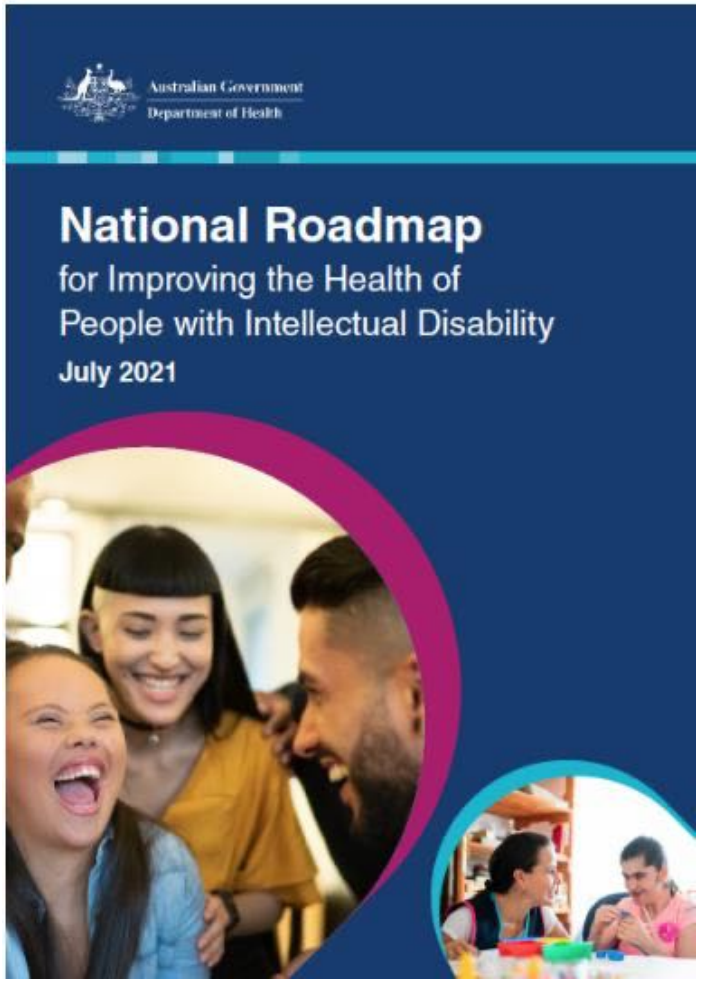
A little note about the slides

- These slides mainly follow Easy Read principles
- We use many images from www.photosymbols.com
- Publications, resources and links are on our website www.GeneEQUAL.com

Health Care Crisis



People with intellectual disability have

- X2 avoidable deaths
- X2 emergency and hospital admissions
- ⬆ physical and mental health conditions
- ⬇ lower rates of preventative healthcare

Australian Government: Department of Health. (2021). National Roadmap for Improving the Health of People with Intellectual Disability. https://www.health.gov.au/sites/default/files/documents/2021/08/national-roadmap-for-improving-the-health-of-people-with-intellectual-disability.pdf

October 2024
PANDDA Conference

GeneEQUAL

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NSW Genetics of Learning Disability (GOLD) Service

NSW GOVERNMENT

Hunter Genetics
Children, Young People and Families

UNSW SYDNEY

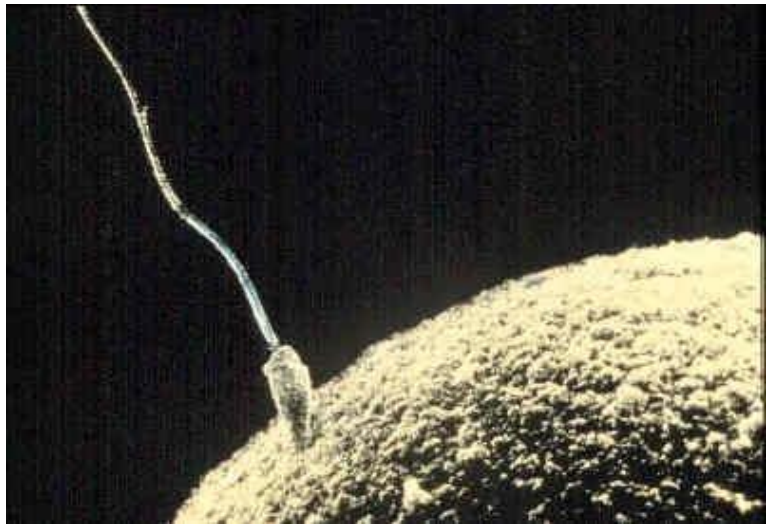
GeneEQUAL: Nothing about us without us!

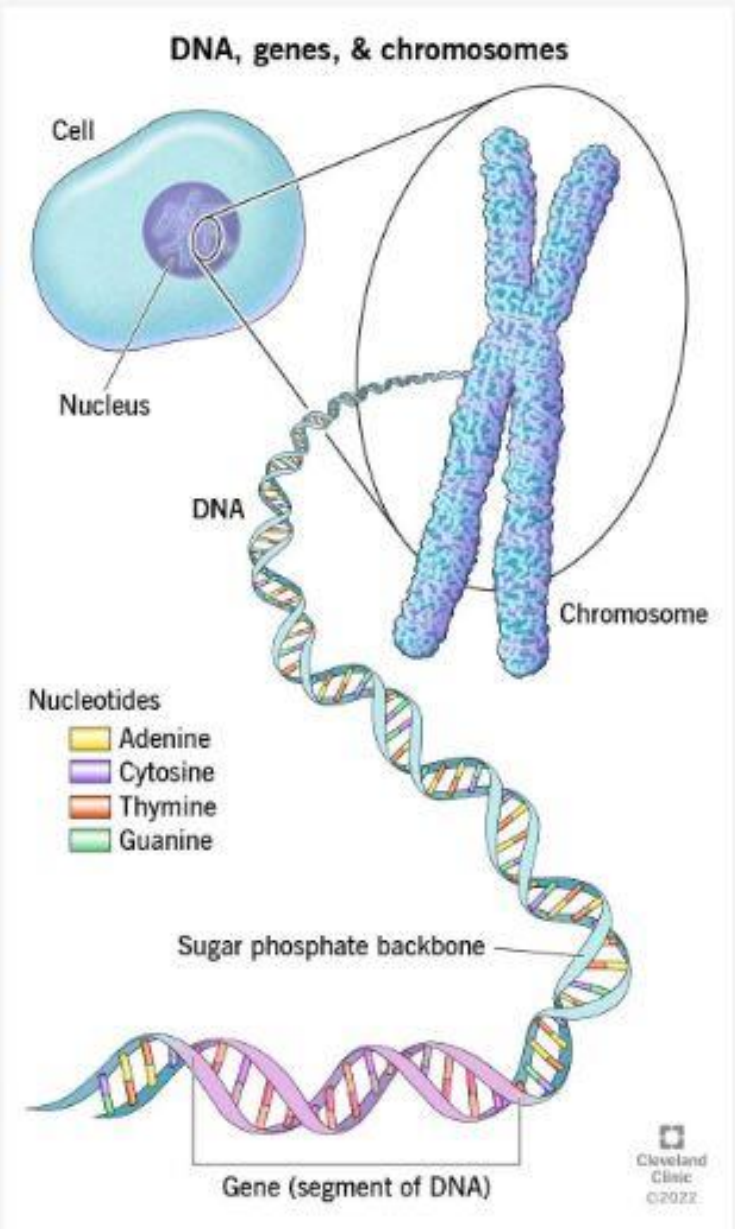
GeneEQUAL is an **inclusive research team** co-led by

- people with intellectual disability
- health care workers
- education, genetics, disability studies and ethics researchers

Genetic healthcare promises so much….

Basic Genetics



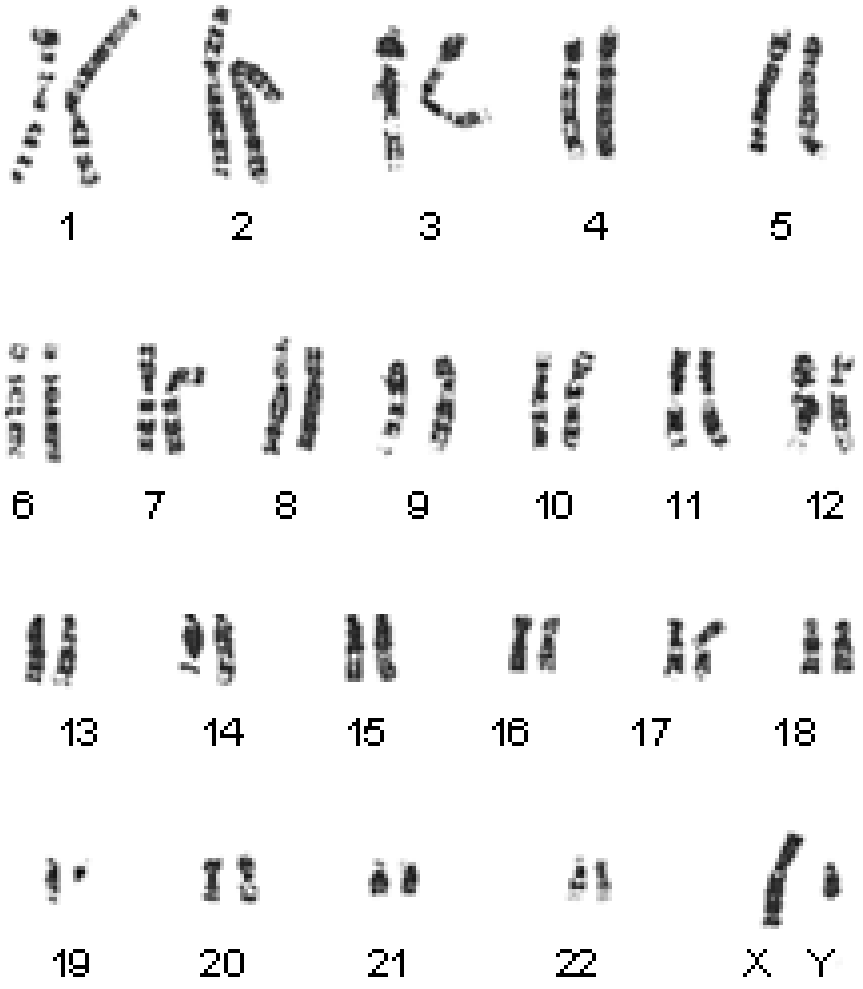


Genetic changes (pathogenic variants/mutations) can be inherited in the sperm/egg (germline) or develop during lifetime (somatic)

The evolution of genetic testing

Karyotype tests for:

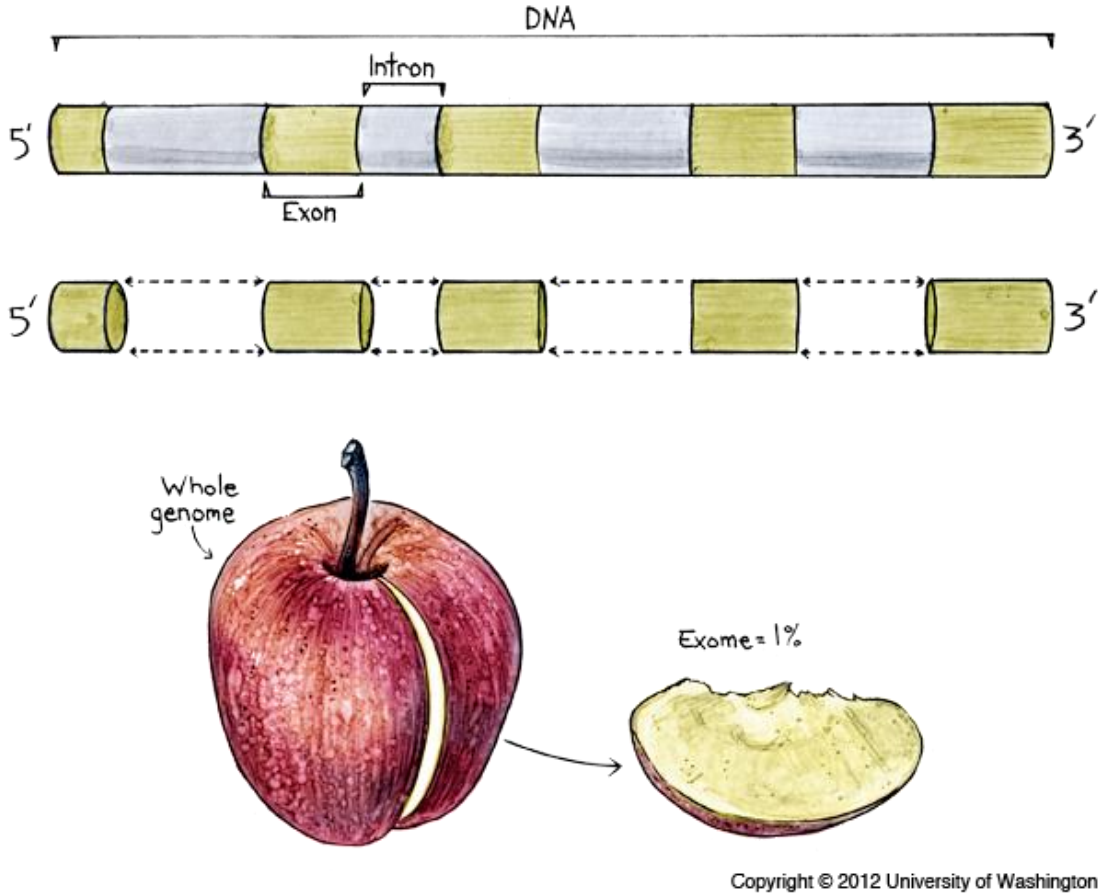
- Extra / missing chromosomes
- Extra / missing pieces of chromosomes



The evolution of genetic testing

- **Microarray** detects small deletions and duplications (copy number variants)
- **Single Gene testing** – may sequence an entire gene or look for common changes within the gene
- **Panels** – test a number of genes associated with a particular condition. Looking at most common changes within those genes

Whole Exome Sequencing



1% of genome is coding – 99% non coding (used to be referred to as junk!)…

Whole Genome Sequencing

We are now learning more about how genes are regulated

Connor A. Horton et al, Short tandem repeats bind transcription factors to tune eukaryotic gene expression, *Science* (2023).
DOI: 10.1126/science.add1250

What is the experience of people with intellectual disability with genetic healthcare?

Our early research told us:

ESHG www.nature.com/ejhg

ARTICLE OPEN

"I am not a number!" Opinions and preferences of people with intellectual disability about genetic healthcare

Iva Strnadová[1,2,3,4], Julie Loblinzk[1,3], Jackie Leach Scully[2], Joanne Danker[1], Michelle Tso[1], Karen-Maia Jackaman[1], Manjekah Dunn[5], Sierra Angelina Willow[1], Skie Sarfaraz[3], Vanessa Fitzgerald[6], Jackie Boyle[7] and Elizabeth Emma Palmer[5,8]

There is limited research exploring the knowledge and experiences of genetic healthcare from the perspective of people with intellectual disability. This study, conducted in New South Wales (Australia), addresses this gap. Eighteen adults with intellectual disability and eight support people were interviewed in this inclusive research study. The transcribed interviews were analysed using inductive content analysis. The findings were discussed in a focus group with ten adults with intellectual disability and in three multi-stakeholder advisory workshops, contributing to the validity and trustworthiness of the findings. Five main themes emerged: (i) access to genetic healthcare services is inequitable, with several barriers to the informed consent process; (ii) the experiences and opinions of people with intellectual disability are variable, including frustration, exclusion and fear; (iii) genetic counselling and diagnoses can be profoundly impactful, but translating a genetic diagnosis into tailored healthcare, appropriate support, peer connections and reproductive planning faces barriers; (iv) people with intellectual disability have a high incidence of exposure to trauma and some reported that their genetic healthcare experiences were associated with further trauma; (v) recommendations for a more respectful and inclusive model of genetic healthcare. Co-designed point-of-care educational and consent resources, accompanied by tailored professional education for healthcare providers, are required to improve the equity and appropriateness of genetic healthcare for people with intellectual disability.

European Journal of Human Genetics; https://doi.org/10.1038/s41431-023-01282-3

1. Many people with intellectual disability
   - Had bad experiences with genetic health care

2. There was a lack of Easy Read information
   - About genetic health care

3. Health professional education was needed

www.GeneEQUAL.com

Key recommendations from Royal Commission

Good health care for people with disability

People with disability have the same right to health care as people without a disability.

They should have the autonomy to manage their own health care.

But people with disability often don't get the health care they need.

This means they can have worse health than other people in the community.

Intellectual disability is a disability that slows down learning. We can learn if the way of teaching matches how the person learns. Intellectual disability is not an inability to think! We know what is going on around us, and we can feel what's going on too.

ROBERT STRIKE, AM

Improving genetic healthcare for people with intellectual disability

Co-production research is the best way to do this.

Co-production means doing research together.

People with disability are **included** in the research process.

Strnadová, I., Dowse, L., & Garcia-Lee, B. (2022). *Doing Research Inclusively: Co-Production in Action.* DIIU UNSW Sydney.

1

Starting research together
We decided to focus on meeting the **top three recommendations** from people with intellectual disability in Phase 1 of GeneEQUAL.

2

Planning research together
We agreed on a co-production and a research plan. We worked together to make sure our project would be accessible and treat everyone with respect.

3

Doing research together
We ran co-production and advisory workshops together. Some of our team worked more on a focus group with people with intellectual disability and others worked more on a health professional evaluation.

4

Understanding data together
We worked together to understand the main themes and findings of our co-production project.

5

Talking about our research together
We share our research together at conferences and by Easy Read newsletters, videos, podcasts and blogs.

6

Sharing what we learnt together
We have reflected on the co-production process together and thought of new ideas and recommendations for how we will work together in the future.

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Doing Research Inclusively. Co-production in Action.

Strnadová, I., Dowse, L., & Garcia-Lee, B. (2022). *Doing Research Inclusively: Co-Production in Action.* DIIU UNSW Sydney.

Co-researchers

Co-researchers add their expertise on

- Easy Read ethics documents
- Recruitment plans
- How to run interviews and focus groups
- And much more

They

- Bring their lived experience
- Are accessibility experts
- Know what will work and what will not

Improving genetic health care for people with intellectual disability – listen to the experts!

Listen to people with intellectual disability about

- their experiences at genetic appointments
- what they liked
- what they did not like
- what they recommend should change

Many people with intellectual disability experienced

- Bullying
- Sexual abuse
- Physical abuse
- Financial abuse

Seeing a new health service team can be frightening

UNSW SYDNEY

Strnadová, I., Loblinzk, J., Scully, J. L., Danker, J., Tso, M., Jackaman, K.-M., Dunn, M., Willow, S. A., Sarfaraz, S., Fitzgerald, V., Boyle, J., & Palmer, E. E. (2023). "I am not a number!" Opinions and preferences of people with intellectual disability about genetic healthcare. *European Journal of Human Genetics*

People made suggestions for how things could improve

- Explain things well
- Stop and listen
- Talk slowly
- Check understanding
- Use pictures
- Use Easy Read
- Give people choices
- Use language which makes people feel respected

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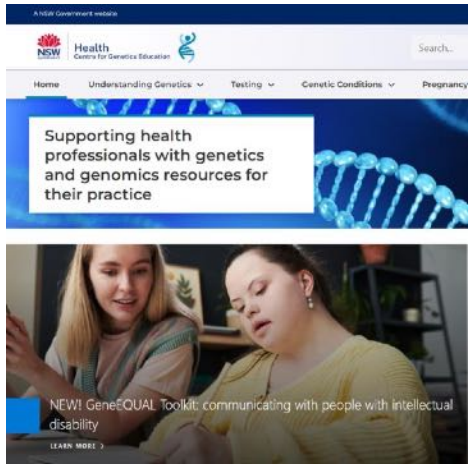
Our first project with NSW Health

We talked with people with intellectual disability about

- what is good or bad

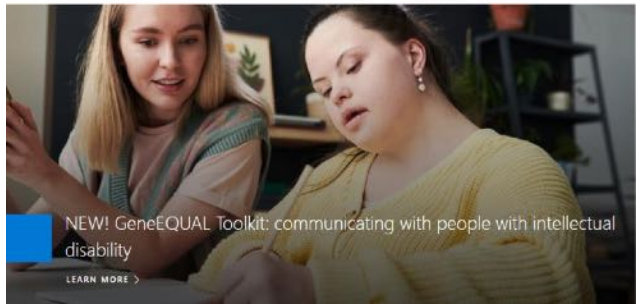
about genetic health care



Then together with people with intellectual disability

- we made an Educational Toolkit

GeneEQUAL Educational Toolkit

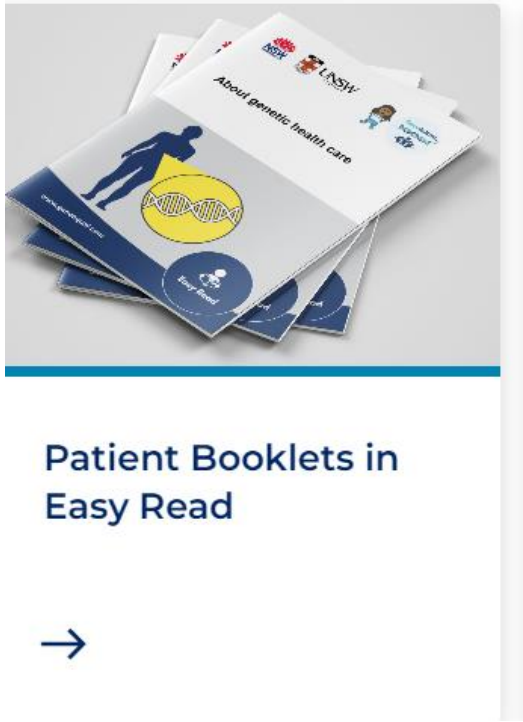


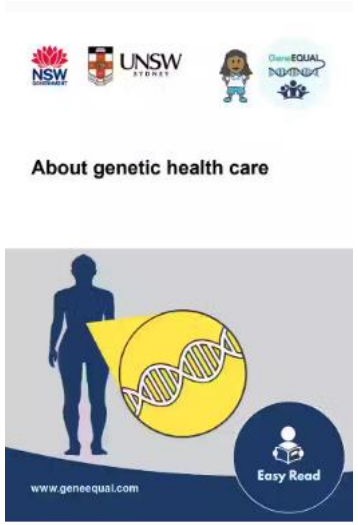
Videos for health professionals on how to provide

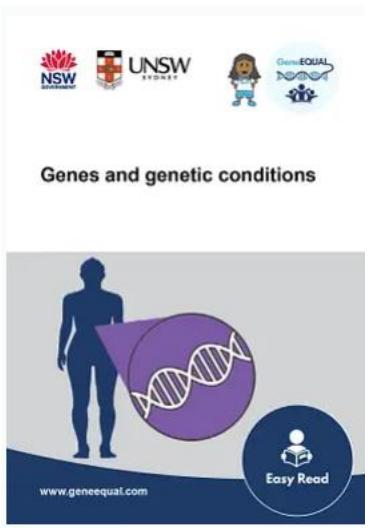
- Reasonable adjustments
- Person centered care
- Strengths based (trauma informed) care

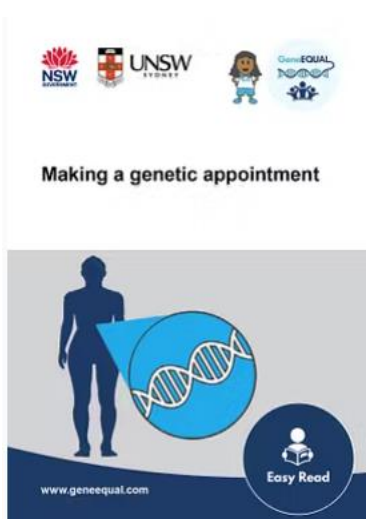
https://www.genetics.edu.au

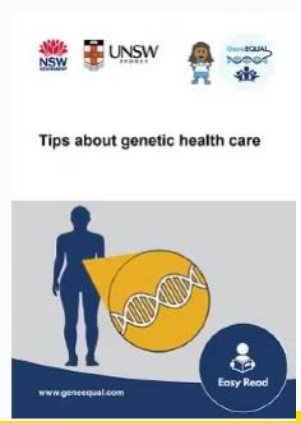
Easy Read booklets about genetics

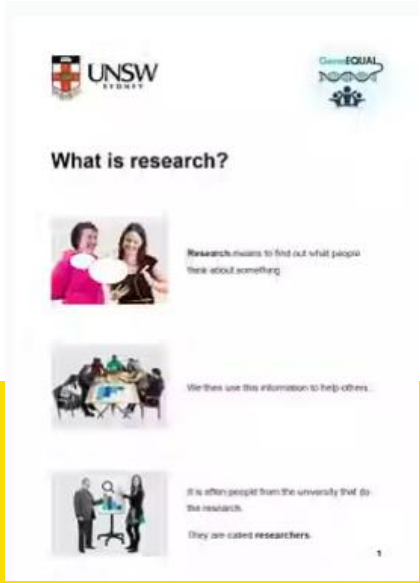












https://www.genetics.edu.au

NSW GOVERNMENT Health
Centre for Genetics Education

Understanding data together

We have been listening to feedback from

- People with intellectual disability
  - through a focus group
- Health care professionals
  - through interviews and a survey

- Manjekah and Chloe are students who lead this work

Our Educational Toolkit has had great feedback

You can find it at www.geneequal.com

Our toolkit has been visited

- over 10,000 times
- by people from 53 countries


OVER TO YOU
What did you like about this process?
How can we improve the way we work together?
What have you learned while working together?

What does co-production mean to us?

Doing research together

- People with a disability working alongside staff who work at universities

1

- Working out what jobs you want to do in Co-production

- Making sure research is about what is important for people with disability

GeneEQUAL's current big project

Australian Government
National Health and Medical Research Council

NHMRC

GeneEQUAL is now funded

- for 5 years
- for a project across Australia
- to include Aboriginal and Torres Strait Islander people
- by the National Health and Medical Research Centre

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NSW Genetics of Learning Disability Service

- Helping families find genetic reasons for intellectual disability
- Genetic appointments for families with 2 or more family members with intellectual disability
- Clinics across NSW
- Provide home visits if required

NSW GOLD Service

Phone: (02) 4985-3100 opt 1
Fax: (02) 4985-3105
Email: HNELHD-GOLD@health.nsw.gov.au