Access to genomic testing for deafness: challenges and enablers of implementation

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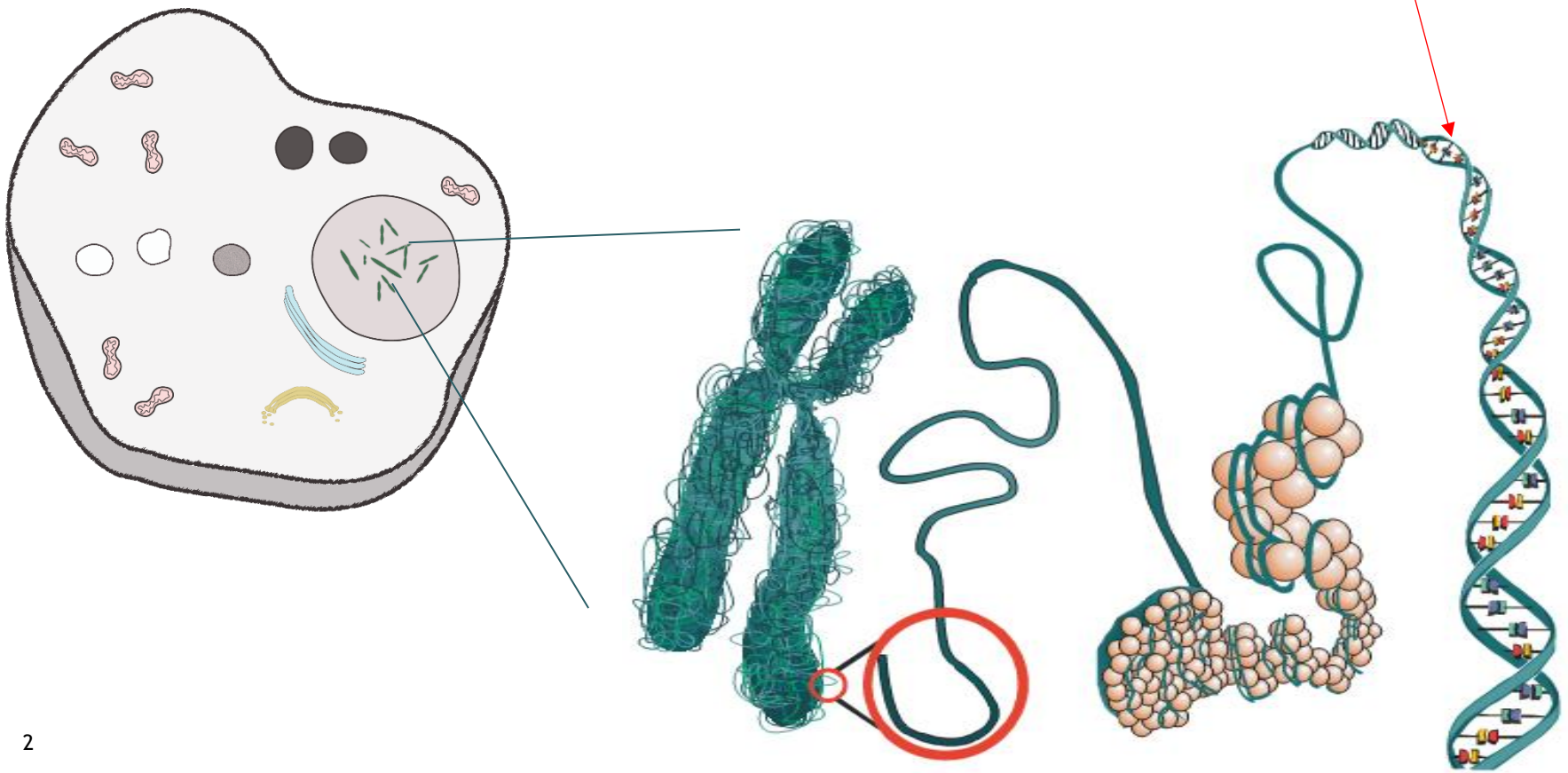
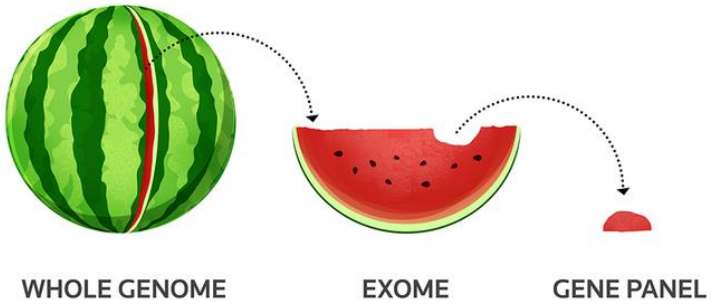
What is genomics?



Genomics and hearing health

Why is my child deaf?
What treatment pathway should we take?
How will my/my childs future look?
Will this happen again?



Background

2023

Anticipated: 200 patients a year
Actual: 18 tests ordered

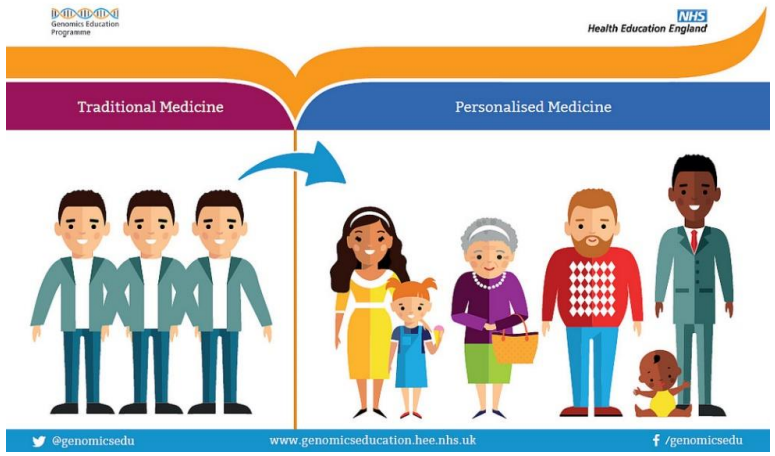
We have a federally funded test, why aren't we using it!

Who is eligible for testing?

- A person diagnosed with hearing loss in childhood with or without parents (singleton or trio)
- Reanalysis 2 years from original test
- Biological relatives
- Reproductive partners of relatives

Benefits of early molecular diagnosis…

- Personal utility
- More accurate human phenotyping
- Precision management
- Precision therapy



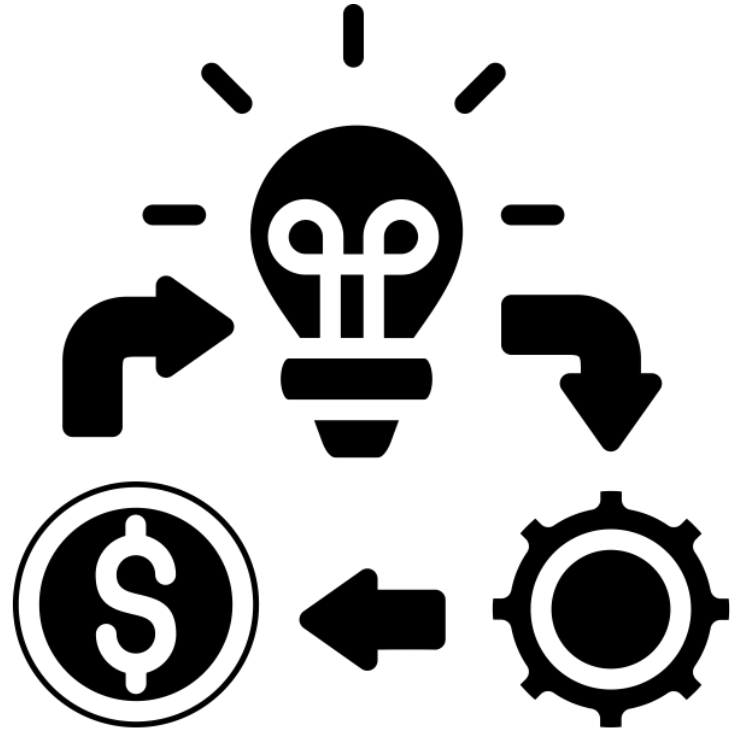
Melbourne Genomics Health Alliance

Alliance members

Supported by

Real world impact = implementation science

- Funded test vs funded service
- Laboratory cost recovery $1200
- Clinician comfort and education



Created by afian roc
from Noun Project

Melbourne genomics implementation fellowship

Genetics education for health professionals

- Online education session(s) via CHAMP network

Support materials

- Adapted from other disease areas (renal/childhood syndromes)

Laboratory support and access

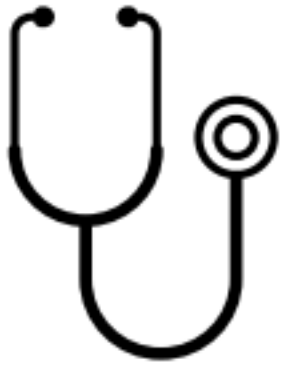
- Application to revise funding for the item number

Genetics support in hearing loss clinics

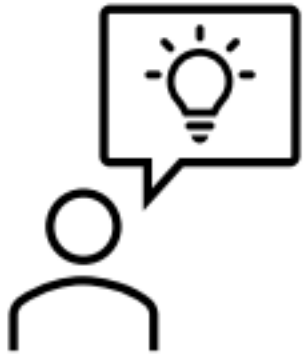
- Genetic counsellor
- Clinical geneticist
- Audit of impact

Barriers for implementation

Availability of genomic testing



Availability of clinicians



Different knowledge levels

Availability of genomic testing

What is happening with the Medicare item number?

- Pricing:
  - 73440 (singleton)
    - Fee = $1200
  - 73441 (trio)
    - Fee: $2100
- Public laboratories unanimously feel that the MBS fees are too low to provide a test

VCGS
leaders in genetic health

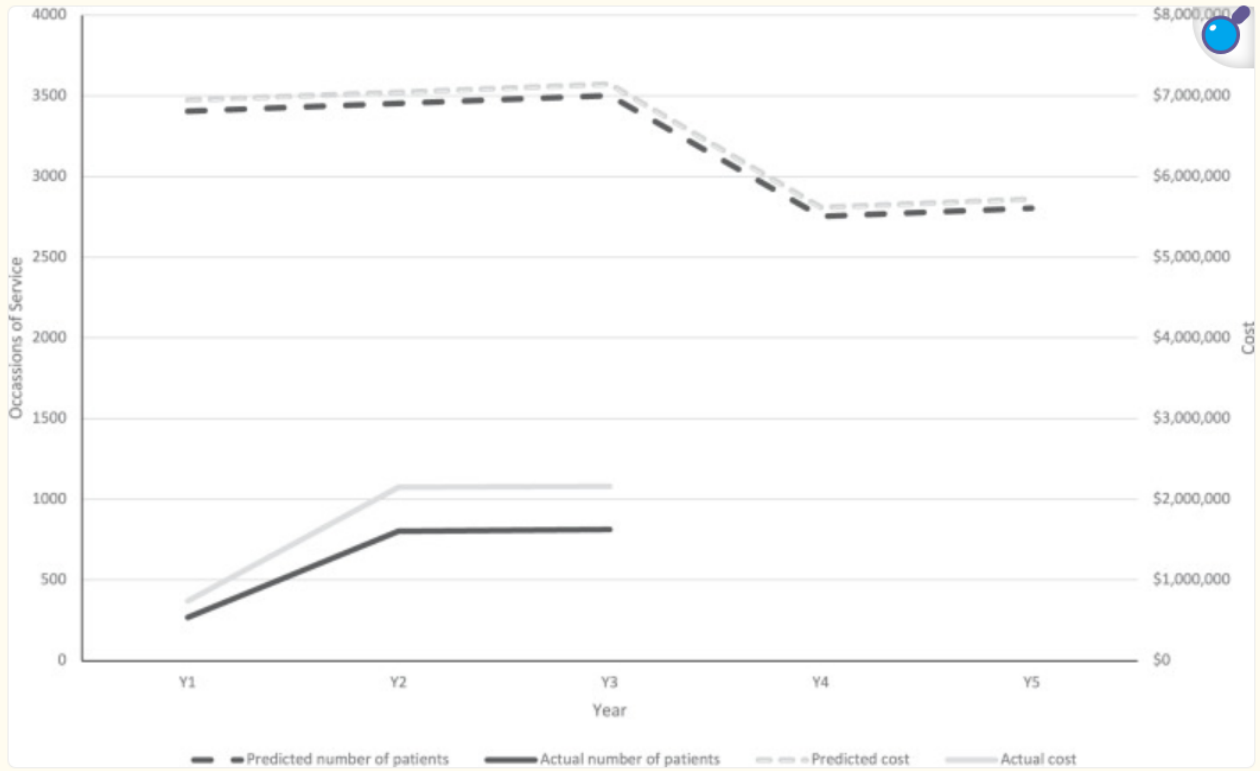
What is happening with the Medicare item number?

- **Tests currently available:**
  - Genomics for Life: custom panel of 175 genes, including CNV analysis of STRC and CATSPER2
  - PathWest: custom panel of 23 genes, including CNV analysis of STRC
  - Pathology SA: custom panel, only available for SA patients
- **VCGS collaboration with Illumina**
  - Using new technology to design test
  - Aiming for NATA accredited test to utilise Medicare funding and offer test with no out-of-pocket costs
- **Current options:** self-funded test, use Genomics for Life, referred to prenatal Genetics service if family planning, or wait for VCGS test

VCGS
leaders in genetic health

Availability of testing is not the only barrier

Fig. 1. Actual vs predicted services and reimbursement cost for childhood syndrome initial testing.



Mordaunt DA, Dalziel K, Goranitis I, Stark Z. Uptake of funded genomic testing for syndromic and non-syndromic intellectual disability in Australia. Eur J Hum Genet. 2023 Sep;31(9):977-979. doi: 10.1038/s41431-023-01417-6. Epub 2023 Jul 3. PMID: 37400487; PMCID: PMC10474079.

Mainstreaming

Availability of clinicians

- Establish a process to enable pediatricians and ENT surgeons to use the new Medicare Item number
- Provide opportunities for hearing loss clinicians to upskill in genomics.
- Streamline the experience of patients who do not require an assessment by a clinical geneticist.
- Provide timely diagnoses or prompt further investigations for syndromic causes of deafness.

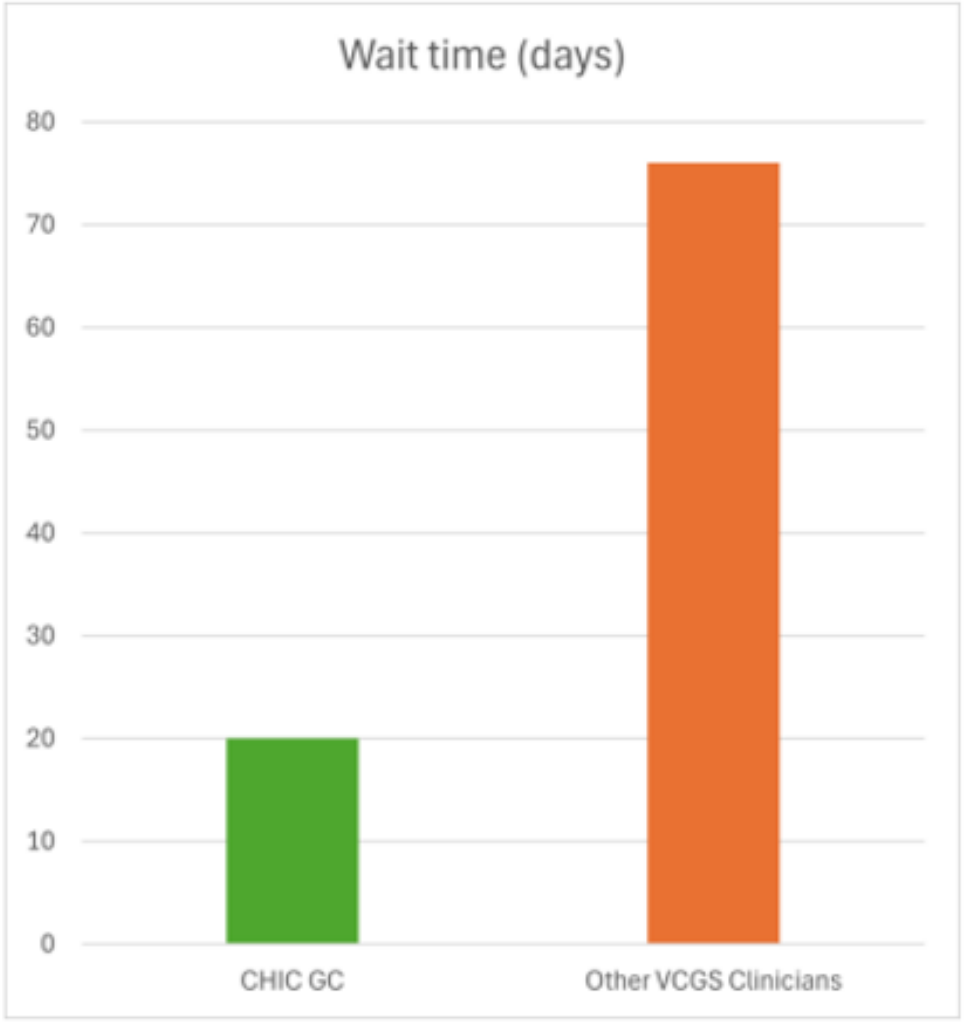
Integrating genetic counsellors into the clinic

- Triaging
- Intake call
- Sitting in with paediatricians
- Familiarity with genomic testing consent

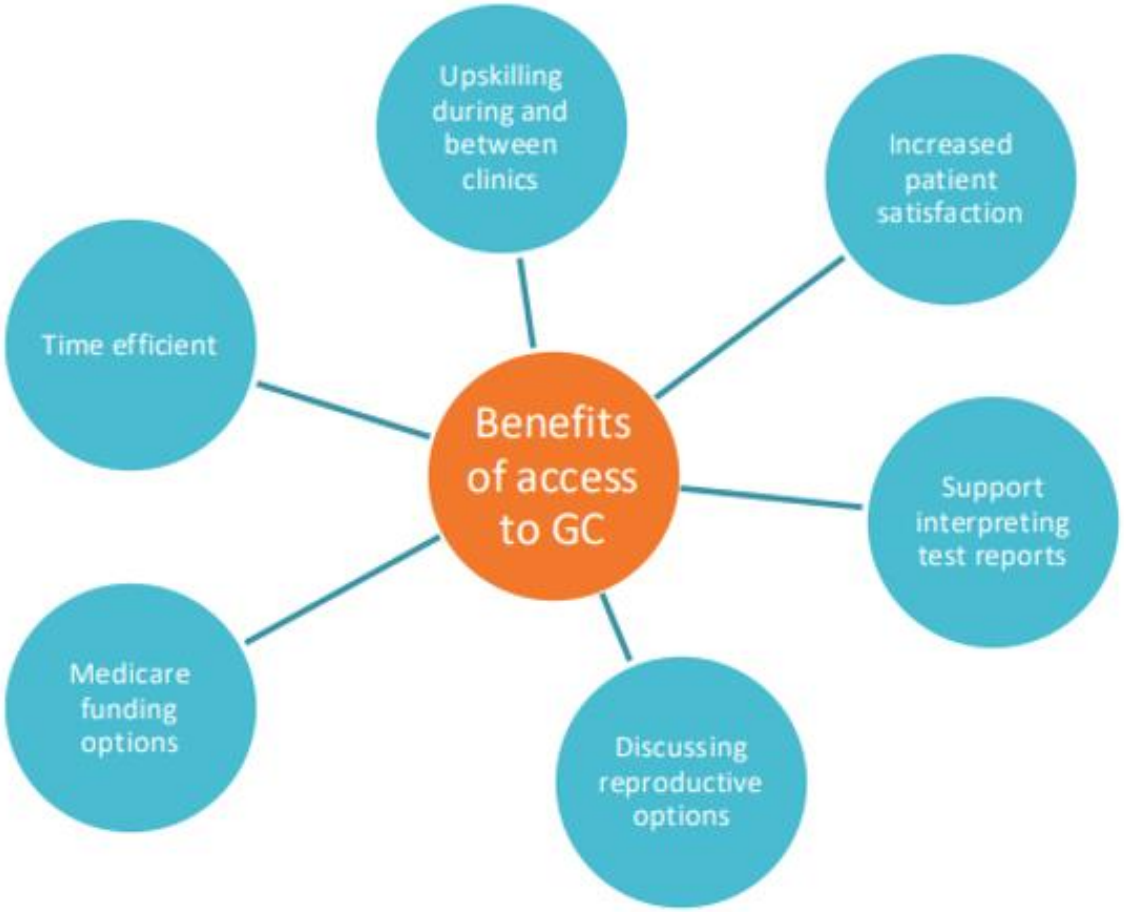
Focus groups with paediatricians and fellows

- Significant reduction in wait time for patients to access genetic health professional support
- Paediatricians ‘strongly agreed’ that access to a GC

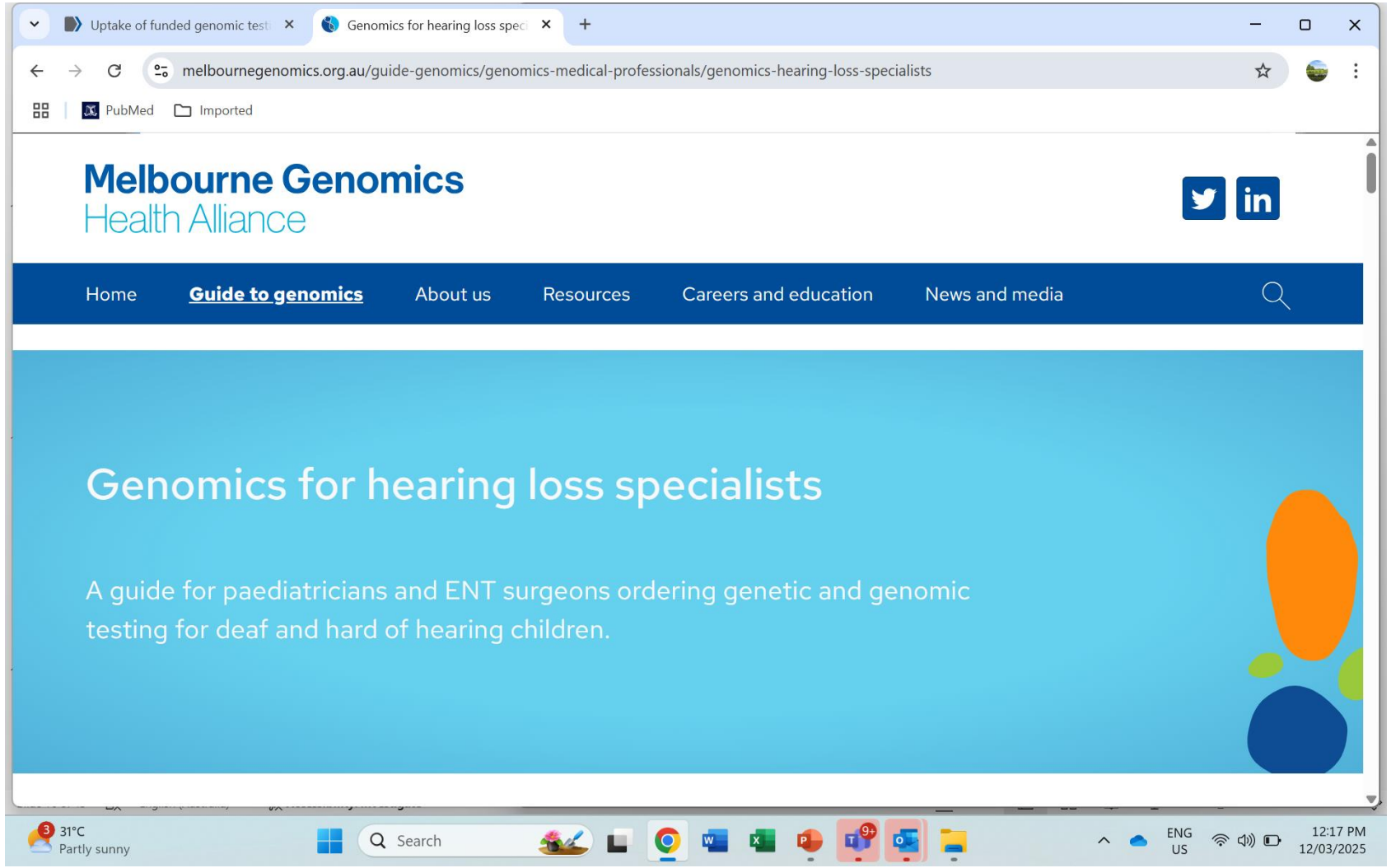
- improved their practice
- provided unscheduled learning opportunities
- essential for mainstreaming genomic testing
- highly utilized compared with use of the written and online resources



Benefits of the integrated clinic model



Different knowledge levels



Hearing loss can be caused by environmental or genetic factors. In around 60% of children with isolated (non-syndromic) hearing loss, a genetic cause can be identified by genomic testing.

This webpage is a **guide for paediatricians and otolaryngologists** who want to learn more about ordering genomic testing for children with non-syndromic hearing loss. **Bookmark it to keep it handy.**

In this document, we refer to individuals as 'deaf or hard of hearing' and use the term 'hearing loss' as a noun to describe the condition affecting individuals who are deaf or hard of hearing. This resource has been reviewed by the Australian Childhood Deafness Community Advisory Group.

***Please note**: This information is not intended to replace personalised patient advice from a genetics expert.*

This page is specific to genomics for hearing loss. If you are after general information, please refer to our genomics explainers.

Visual Genomics Report

The genomics test will examine if there is a genetic cause for your patient's condition. The report will have one of three outcomes.

A genetic cause:
Genetic diagnosis of autosomal recessive deafness (OMIM#600316)

No cause:
A genetic cause for this individual's phenotype has not been identified

A variant that is of uncertain significance (VUS):
The result is uncertain because of a lack of evidence pertaining to one or more of the examined variants

If your patient has no cause found, there may still be options to do a re-analysis. This may include expanding the gene panel that was looked at or routine re-analysis in a few years.

This document was designed to help Doctors explain the content of a genomics report to their patients, with the assistance of the included illustrations.

If your patient desires more information, you can refer them to this Melbourne Genomics page

Plain language genomics report templates are available to download and edit here:

Melbourne Genomics Health Alliance

This resource was created as part of a **Melbourne Genomics project** and was created with **Alternative Contracting** in collaboration with the **UWA Comic Contracting Project** and **Coventry Comics**
https://www.alternativecontracting.biz/
https://www.comicbookcontracts.com/

Top of the Report

The Variant

Your report will say:

Which test you have requested
eg. Clinical Exome Trio Analysis

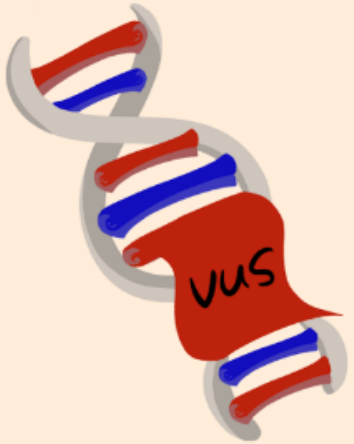
Reason for Referral
eg. Moderate bilateral sensorineural hearing loss

Which genes have been looked at
Gene List applied
eg. Deafness_IsolatedAndComplex v1.196

The classification of the variant:

If no diagnosis is found this information will help you decide if further testing is needed.

There are 5 possible classifications for a variant:



5. Pathogenic:
A cause of medical issues

4. Likely Pathogenic:
Likely the same except the databases contains too few example to be certain.

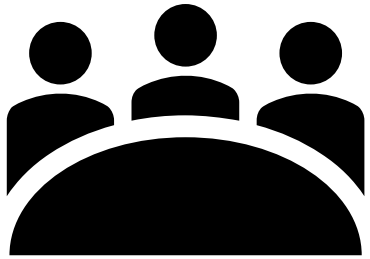
3. VUS:
a variant of uncertain significance

2. Likely benign:
Likely no negative effects.

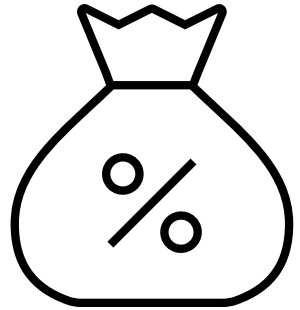
1. Benign:
No known negative consequences.

What have we achieved?

Resources tailored to hearing loss clinicians



Focus groups to generate evidence for genetic services to be integrated into clinic models



Strategies to increase the number of laboratories offering testing

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What does the future look like?

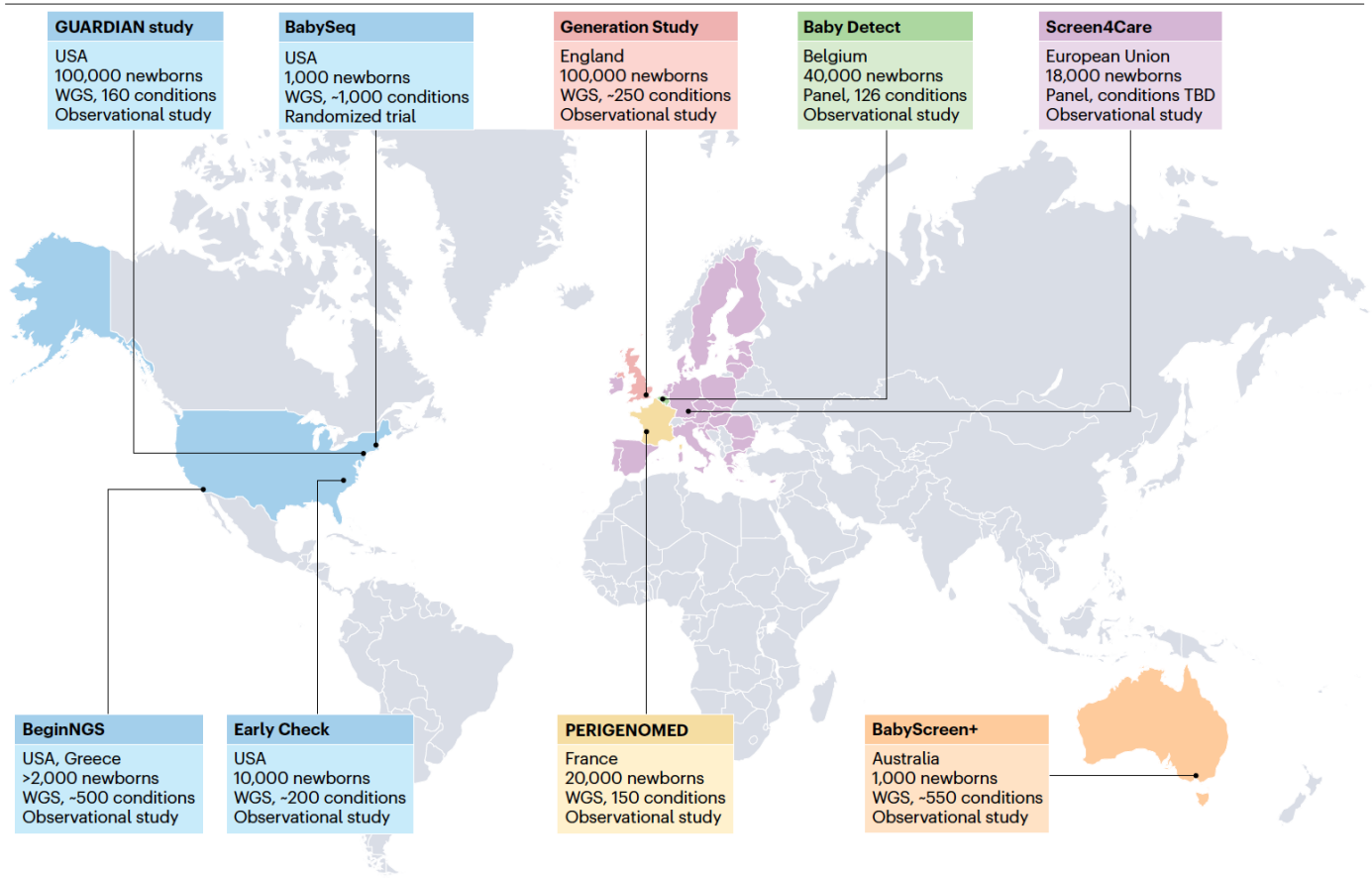
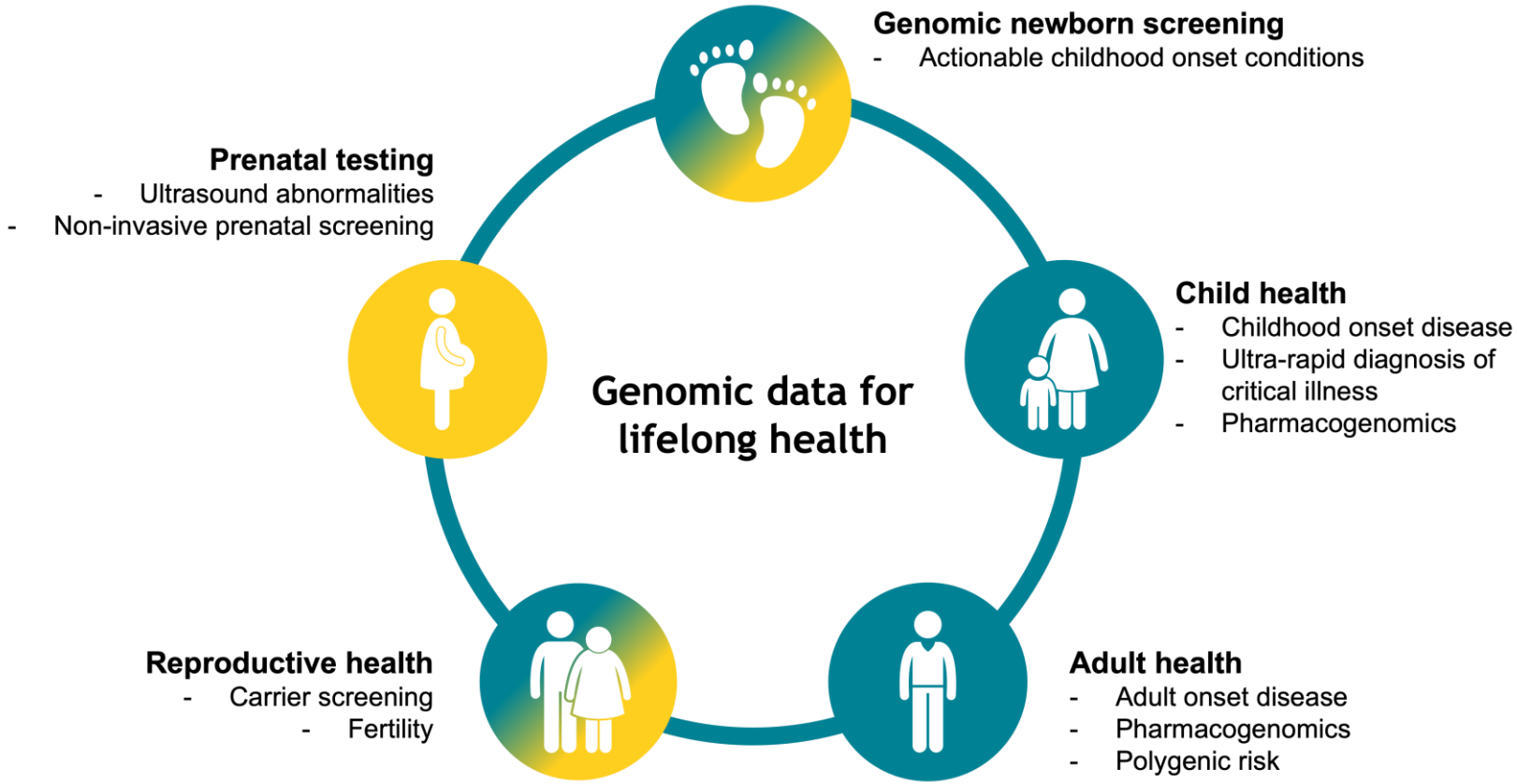


Fig. 1 | Large-scale genomic newborn screening studies launching internationally. Cohort sizes and proposed screening approaches are shown. TBD, to be determined; WGS, whole-genome sequencing.



Thank you

murdoch children's research institute

Melbourne Genomics Health Alliance

Global knowledge. Individual care.

Alliance members

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The Royal Children's Hospital Melbourne

The University of Melbourne

Walter+Eliza Hall Institute of Medical Research

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