

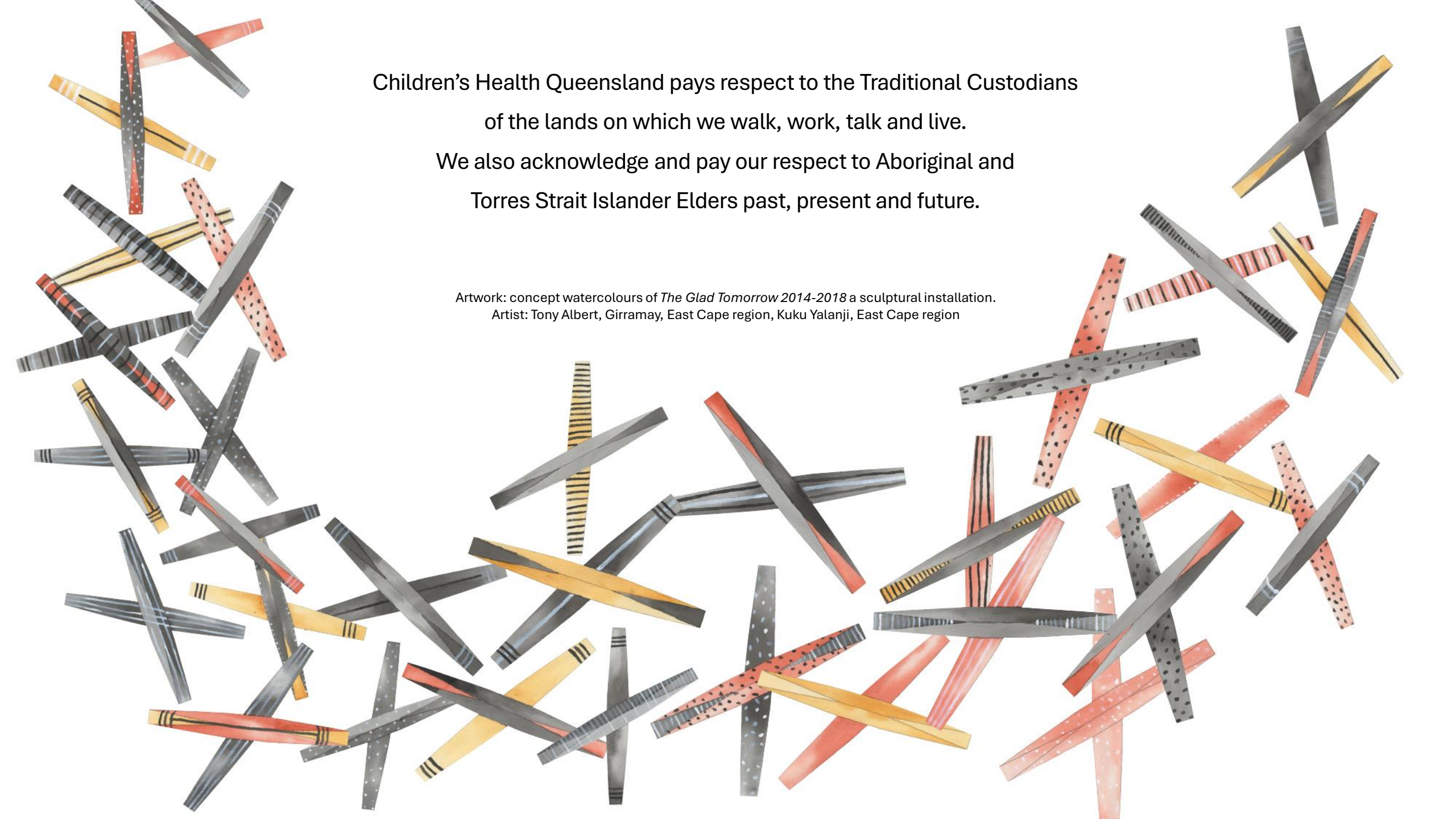
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What do clinicians and laboratory scientists think about Medicare-funded genomic testing for childhood deafness?

Dr Karen Liddle
Paediatrician and PhD Candidate

Children's Health Queensland pays respect to the Traditional Custodians
of the lands on which we walk, work, talk and live.
We also acknowledge and pay our respect to Aboriginal and
Torres Strait Islander Elders past, present and future.

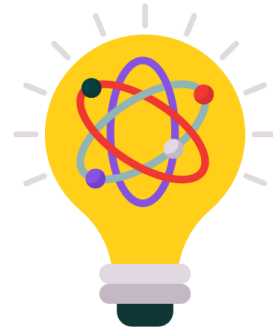
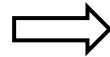
Artwork: concept watercolours of *The Glad Tomorrow 2014-2018* a sculptural installation.
Artist: Tony Albert, Girramay, East Cape region, Kuku Yalanji, East Cape region



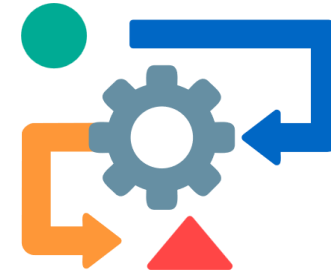
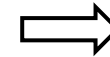
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Background



Aims



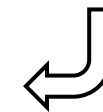
Methods



Conclusions



Results



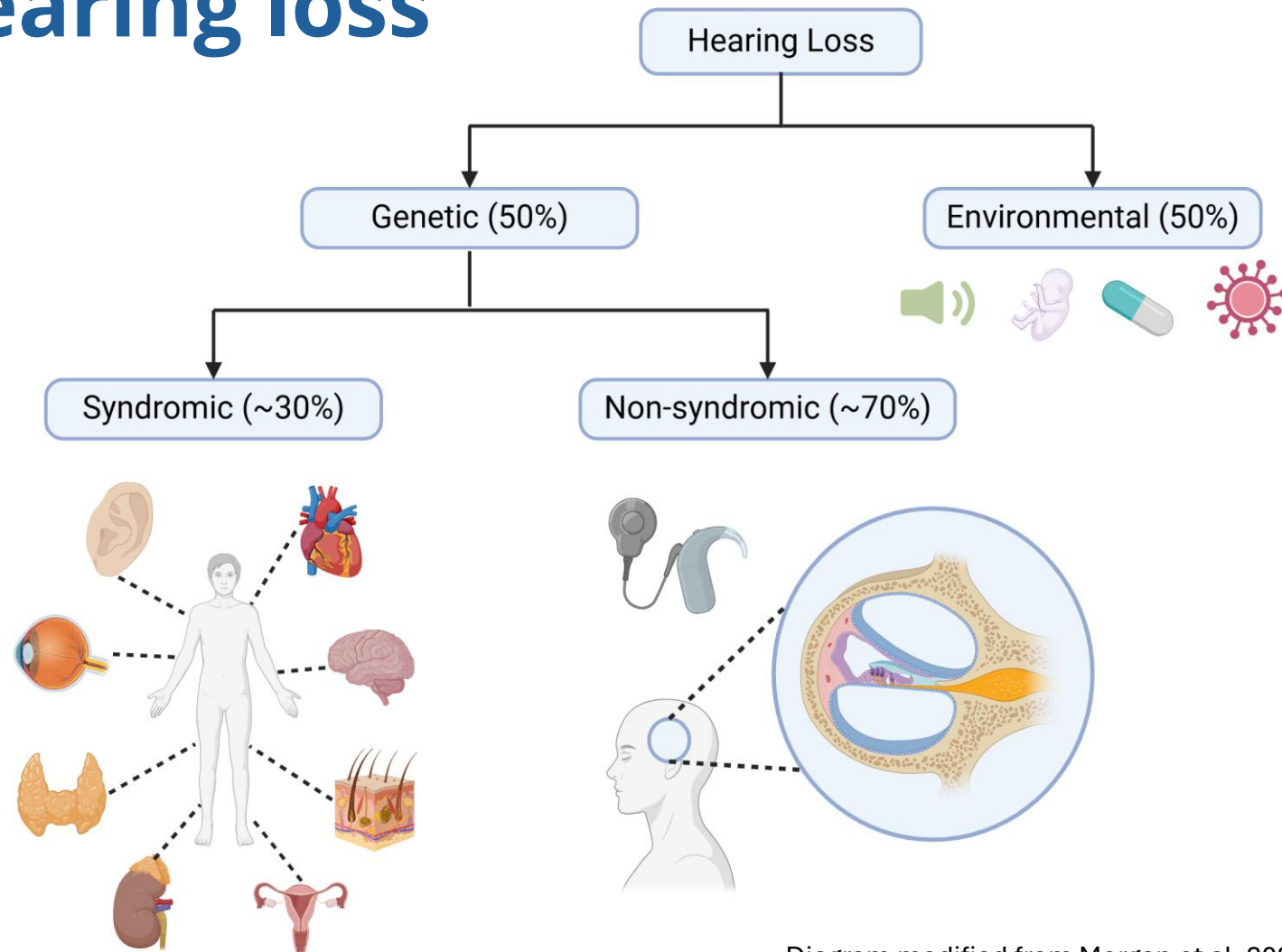
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Background



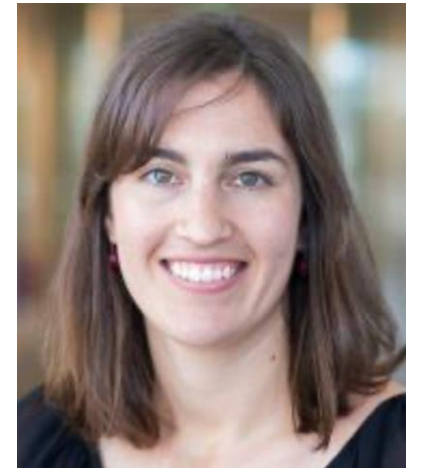
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Cause of hearing loss



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Medicare Services Advisory Committee application for funding



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Table 1 MSAC's supported MBS item descriptors

		Category 6 – PATHOLOGY SERVICES Group P7 – GENETICS
MBS item AAAA1	73440	
<p>Genomic testing and copy number variant analysis of genes known to be causative or likely causative of childhood hearing loss, if:</p> <ul style="list-style-type: none">(a) the characterisation is requested by a specialist or consultant physician experienced with childhood hearing loss; and(b) the patient has congenital or childhood onset hearing loss that presented prior to age 18 years and is permanent moderate, severe, or profound (>40 dB in the worst ear over three frequencies) and classified as sensorineural, auditory neuropathy or mixed; and(c) the patient is not eligible for a service to which items 73358 or 73359 apply;(d) the characterisation is not performed in conjunction with or following a service to which MBS item AAAA2 applies <p>Applicable once per lifetime.</p> <p>Fee: \$1,200.00 Benefit: 75% = \$900.00 85% = \$1,106.80</p>		

Genetic
Counselling
must be
offered

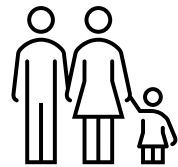
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Rebates and rules



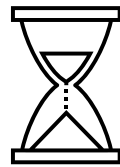
\$1200

'singleton'



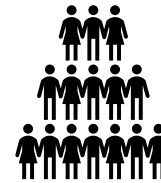
\$2100

'trio'



\$500

reanalysis



\$400

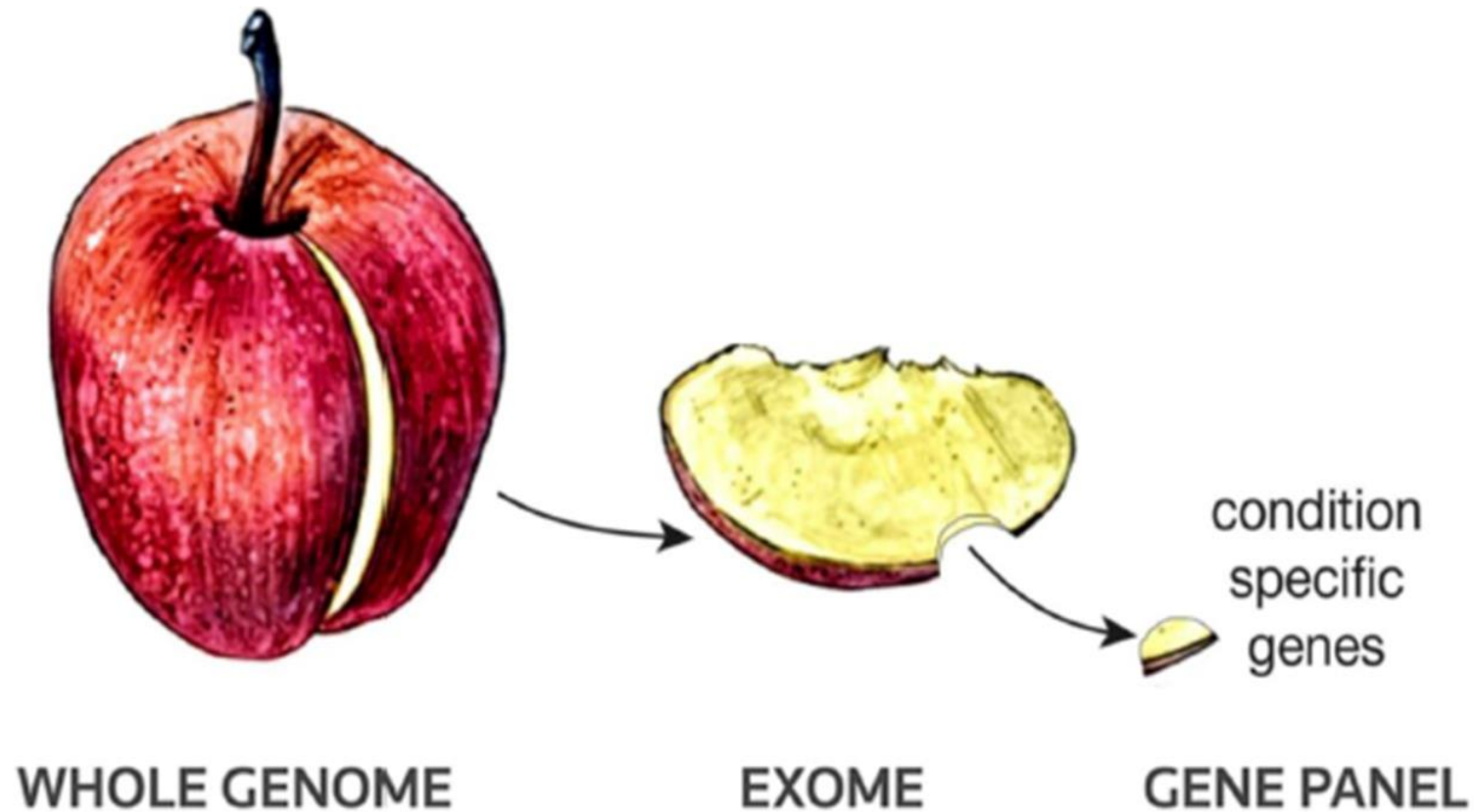
Family testing



\$900

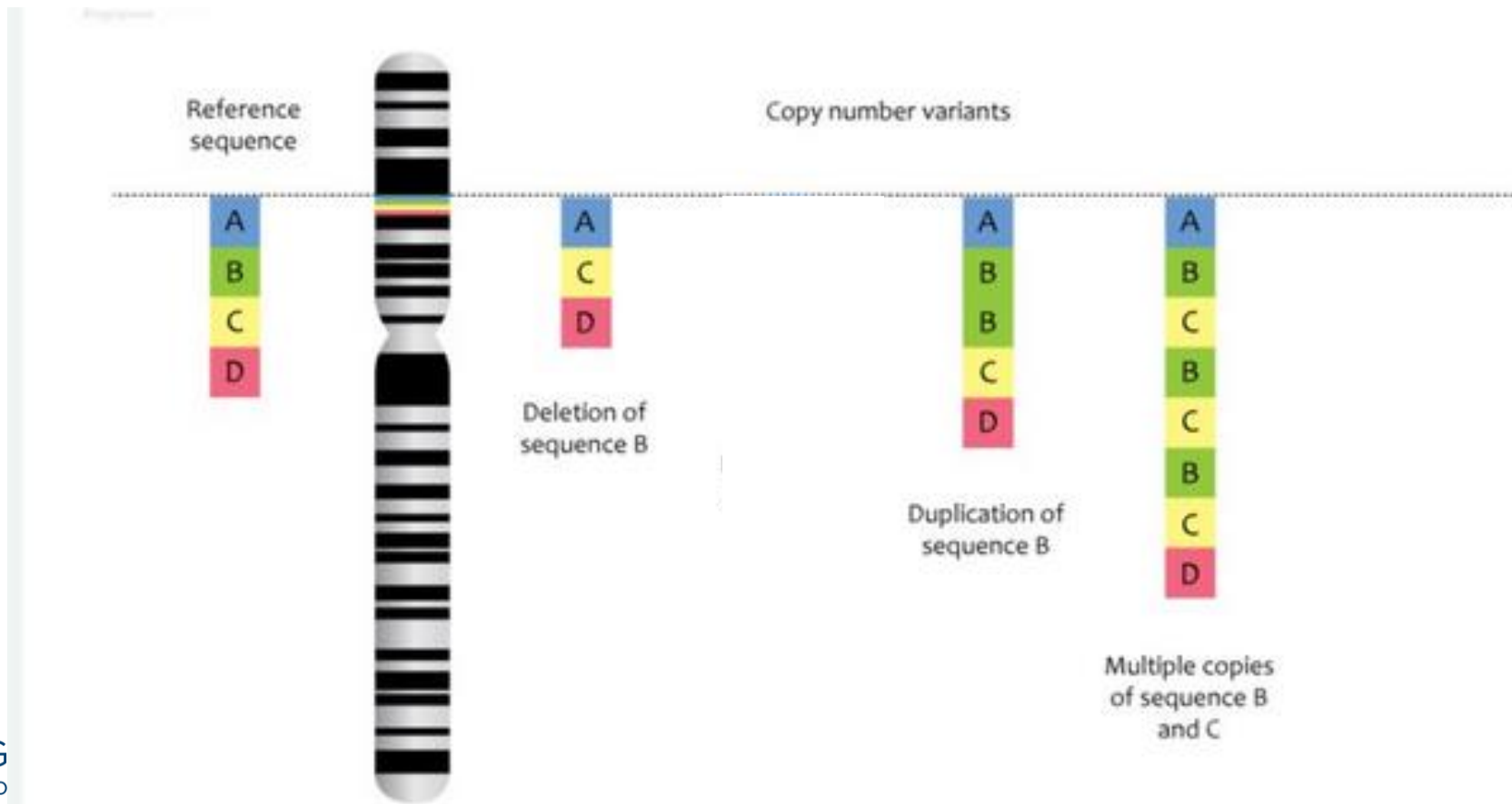
reproductive

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Copy number variants



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Expected uptake

Prospective:



300 eligible/ year;

expected uptake 60%

~180-200 per year

Retrospective:

3000 estimated eligible <18








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Are people ordering testing?

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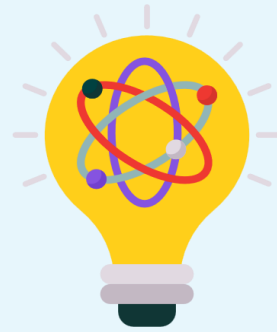
Services Australia data: Nov 2023-Jan 2025

Item number	State						Total
	NSW	VIC	QLD	SA	WA	TAS	
	12	18	3	13	9	2	57
	0	11	1	0	0	0	12
	0	0	0	0	0	0	0
	14	0	0	5	0	0	19
	0	0	0	0	0	0	0
Total	26	29	4	18	9	2	88

Family testing

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Aims



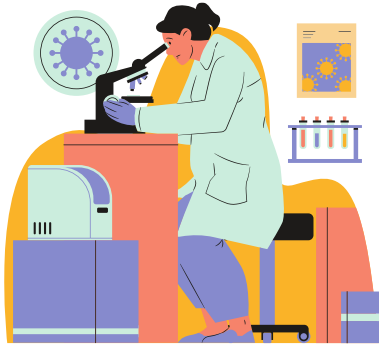
illumina



CREATE CHANGE

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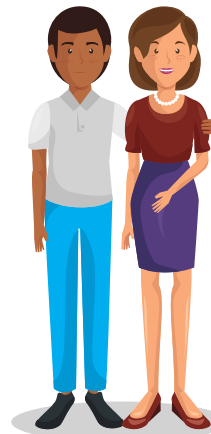
Aim: To capture the perceived feasibility and utility of genetic testing for hearing loss



**barriers and
facilitators for NATA
accredited testing**



**Clinicians'
perspectives on
offering genetic
testing**

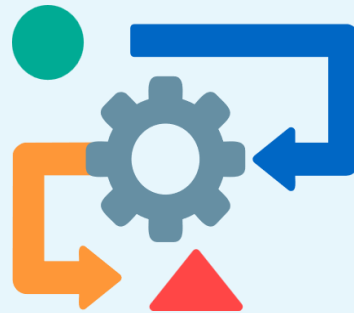


**young people
and parents'
perspectives**



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Methods



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Participants approached by email or EOI
at clinical meeting

Snowball sampling

Semi-structured interview via Zoom

Transcribed and thematically analysed

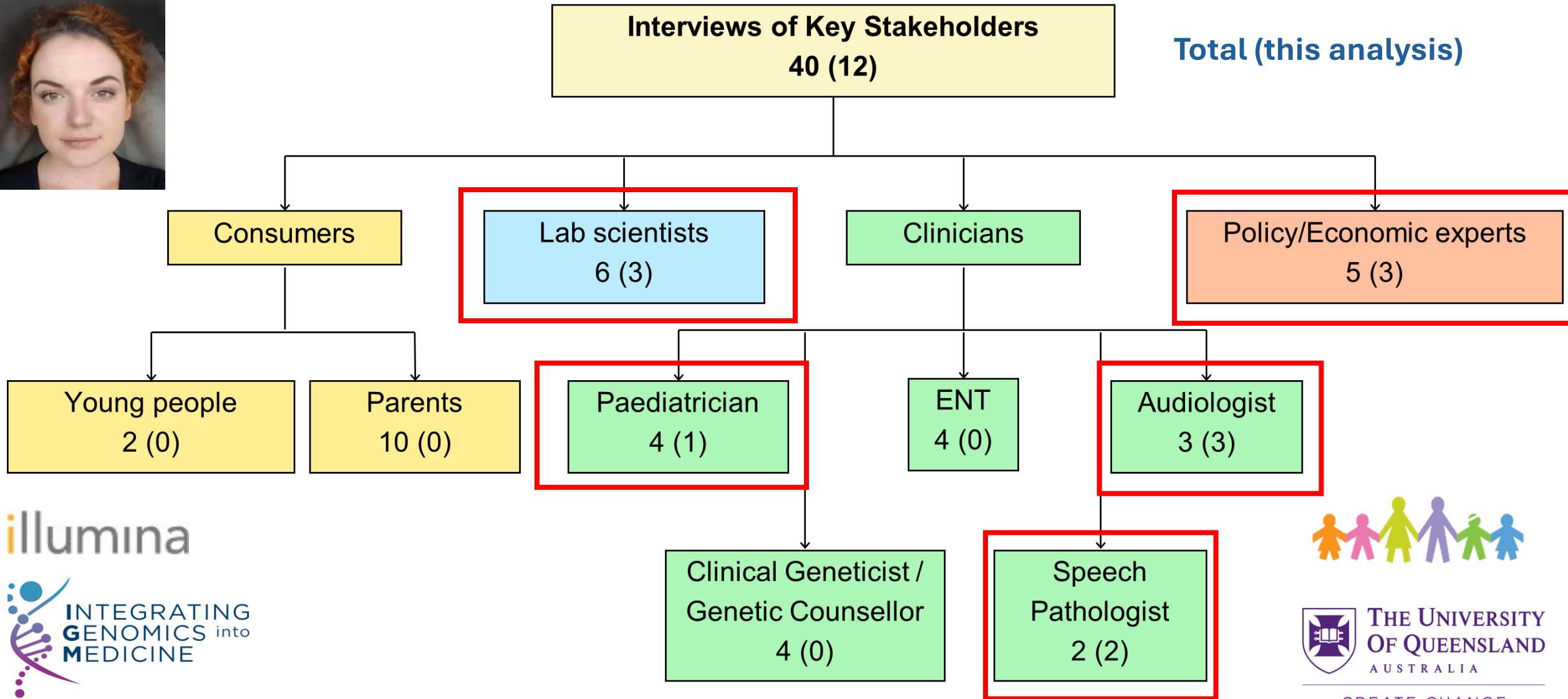


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Results



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Themes



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Clinical Utility: Parental reassurance



"My experience with most families is they're very, very grateful when they get an answer."

- Lucy, Speech Pathologist

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Clinical Utility:
Influences management



"It can give you information about the prognosis for the hearing loss...It helps you to know whether there can be associated medical features like, um, cardiac or renal or thyroid problems that you need to look for."

- Dianne, Paediatrician

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Clinical Utility:
Family planning



"It can help with genetic counselling. Um, it can for that child, for their own children, for the parents having subsequent children, for siblings."

- Dianne, Paediatrician

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Theme 2



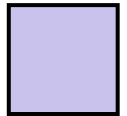
Clinical utility of genomic testing



Facilitators to offering genomic testing



Laboratories capability and capacity for genomic testing



Barriers to offering genomic testing



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Laboratories: Implications of chosen testing modalities



"The sensitivity for detecting small deletions and duplications in [the] genome is way higher than for exomes. And the confidence with which you can do it is way higher as well. So, I think really to me, the best method if you can do it for the right price, is genomes."

- Paul, Laboratory Scientist



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Laboratories: Ease of variant interpretation

“

"I'm not a deafness expert, I have to admit. Um, I'm not sure there's anything that is super unique to deafness when it comes to variant curation and interpretation"

-David, Laboratory Scientist

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Laboratories: Ease of variant interpretation



"A fair amount is known about the diseases as well as far as the, the cause and the molecular mechanism of disease. And so because of that, I think it makes it a little bit easier [to curate variants] as well. So I think, I think as far as diseases go, it's probably one of the easier ones

- Paul, Laboratory Scientist



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Laboratories: Diagnostic yield



"From my experience, the diagnostic yield associated with hearing loss panels is pretty good."

- Paul, Laboratory Scientist

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Laboratories: Prevalence of deafness



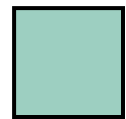
"So, if every deaf person was to access testing, then it would put quite a strain on the laboratories that are offering this testing ...so I think that's one of the challenges, the commonness of it."

- Paul, Laboratory Scientist



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Results: Theme 3



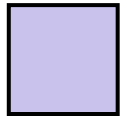
Clinical utility of genomic testing



Facilitators to offering genomic testing



Laboratories capability and capacity for genomic testing



Barriers to offering genomic testing



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Barriers: Rebate insufficient to cover cost



"...for the hearing loss...[the rebate] can be from 20 to 60% less than the actual cost."

- Edward, Health Economist



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Barriers: Inconsistency between rebates



"With Alport syndrome, the rebate is the same...which obviously assumes that the, the same amount of labour [is] involved in analysing 200 genes versus four genes, which is simply not true."

- Michael, Laboratory Scientist

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Barriers: Not just a simple panel



"...we should be performing copy number variant analysis for [particular] genes because that's a common cause of deafness...[although] we are unable to achieve that together with sequencing all of the genes for the fee that has been assigned."

- Sophie, Genomic Policy Expert



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Trio testing - economically feasible but not always practical



"It kinda gets tricky and becomes an ethical question as well, you know, saying no to singletons because dads passed away. Is that, how does that work? You know, it's not really fair, is it?"

- Michael, Laboratory Scientist



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Barriers: Clinician capacity



"So, once you've got private audiologists in play, doing hearing tests with kids in every shopping centre, you are not gonna be able to mandate for every diagnosis of a sensorineural loss to be referred back...to clinic [for genetic testing]...in [public hospitals] people just don't have the time in the, in a day.."

- Lucy, Speech Pathologist



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Barriers: Lack of clinician education



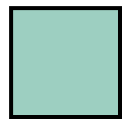
"...we need to increase education in [the clinician] space massively...genomic literacy needs to be much more pervasive in healthcare professions. "

- David, Laboratory Scientist



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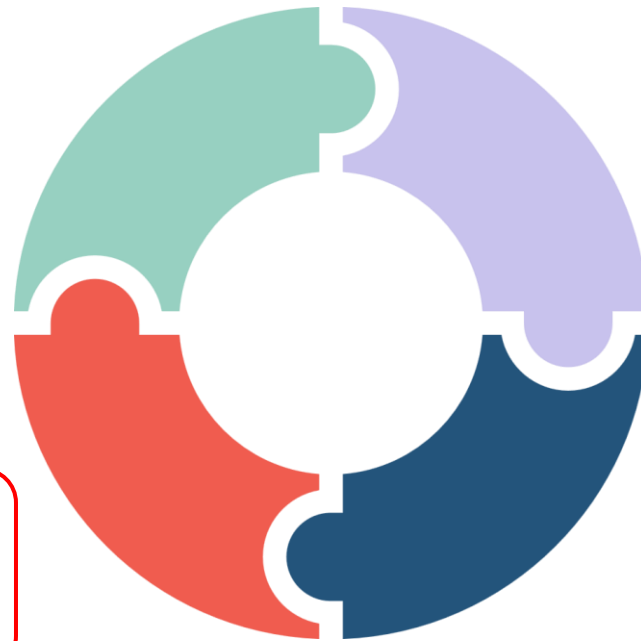
Theme 4



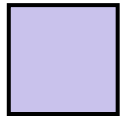
Clinical utility of genomic testing



Facilitators to offering genomic testing



Laboratories capability and capacity for genomic testing



Barriers to offering genomic testing



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Facilitators: Strong desire to offer test



"It's really heartbreaking for us to not be able to offer it."

- Sophie, Genomic Policy Expert

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Facilitators: Education and opportunities for clinicians



"Keep on educating us all about what's available, when and what is offered, and what would trigger, you know, that next level [for referral]...I think having an ongoing support, not just a once off [training] that would be incredibly useful."

- Bethany, Speech Pathologist



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Facilitators: Increasing resources and pathways



"You really need to be supporting a whole bunch of paediatricians...to be able to order it. And that needs a whole, a whole lot of work in terms of education, upskilling, creating the pathways, you know, deploying genetic counsellors to help with mainstreaming."

- Sophie, Genomic Policy Expert



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Facilitators: Multidisciplinary teams



"I think MDTs are worth their weight in gold...They're expensive, but...I'm quite a big fan of talk(ing) through some of these curly variants with people who have a relationship with the patient..."

- Paul, Laboratory Scientist



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Conclusion



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Conclusion

- Rebates significantly underutilized
- Participants felt testing was valuable
- Rebates do not cover laboratory cost
- Interest in upskilling

- Analysis of interviews is ongoing



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Acknowledgements



A/Prof Aideen
McInerney-Leo



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Sung



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Beswick



Ms Hannah
Byrne



Ms Lucy
Burbury



Australian Government
**National Health and
Medical Research Council**

Queensland Health

**Queensland Health Clinical
Research Fellowships**



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Thank you



