# Co-design, implementation, and evaluation of plain language genomic test reports

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# Gap analysis

## Current situation:

**Understanding and communicating genomic results can be challenging** for families and health professionals without genetic specialty training

## What do we need to do to get there?

Modify existing laboratory reports?
Create plain language reports?
**Opportunity for patient/family-centered approaches?**

## How do we do that?

**Co-design plain language report templates** with:
- patient groups
- plain language experts
- educators
- genetic health professionals

## Desired state:

**Better** patient/family and caregiver **understanding** and **communication** of genomic test **purpose**, **outcome**, and potential **clinical implications**

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# Co-design of plain language genomic test report templates

Scoping review and consultation with international experts identified common elements of plain language summaries:

- **synoptic reporting frameworks** (i.e. partitioning content under section headings)
- **patient narrative formats** (i.e. content reflecting the order of events that patients experience)

Three plain language genomic test report templates drafted:
***de novo* autosomal dominant, autosomal recessive, and uninformative results**

Feedback sought through consultation with:

- **specialists** (n=27), Acute Care Genomics study investigators
- **consumers** (n=19), Australian Genomics Community Advisory Group, Queensland Genomics Health Alliance Community Advisory Group, and Syndromes Without A Name Australia

Draft templates modified and improved through an iterative process incorporating suggestions regarding **layout, content, language and utility** alongside **plain language specialist review**

# Co-design of plain language genomic test report templates

**Eight plain language genomic test report templates** were developed for common genomic test outcomes in rare disease

**Templates for use where a diagnosis was achieved:**

- *de novo* autosomal dominant
- inherited autosomal dominant
- autosomal recessive
- X-linked inherited
- X-linked *de novo*
- mitochondrial

**Templates for use where a diagnosis was not achieved:**

- variant(s) of unknown significance with high clinical significance (i.e. strongly suspected to be causing the phenotype)
- uninformative results (i.e. no variants reported)

**plain language genomic test report templates available here:**

**https://bit.ly/3t6CUw4**

# Implementation of plain language genomic test report templates in acute paediatrics

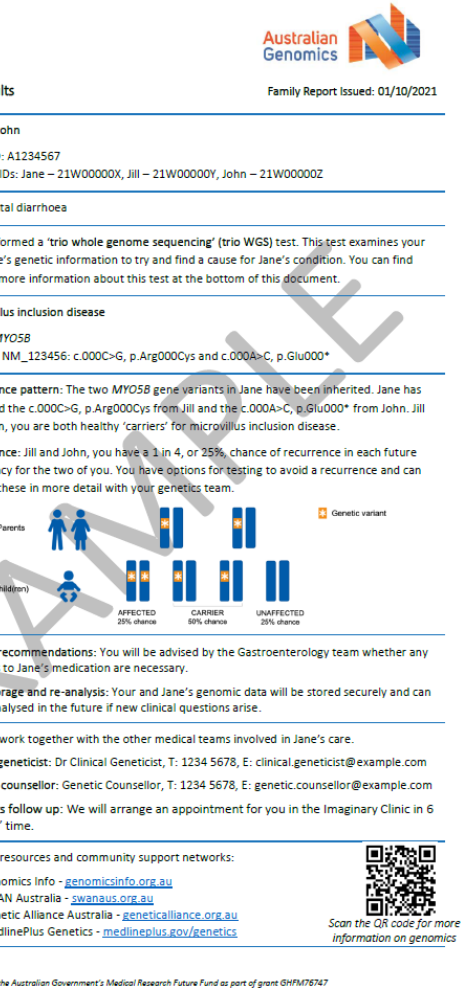
**national implementation** in
the Acute Care Genomics study**

pre-filled reports **electronically distributed to primary involved genetic health professionals** alongside laboratory reports

recipients encouraged to **replace pre-filled general information with personalised information**, including:

- genetics team contact details
- follow up arrangements
- community supports

recommended personalised 'family reports' be **provided to parents/guardians as part of result disclosure**



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**Jane Doe's Genomic Test Results** — Family Report Issued: 01/10/2021

| | |
|---|---|
| Parents' names | Jill and John<br>Study ID: A1234567<br>Sample IDs: Jane – 21W00000X, Jill – 21W00000Y, John – 21W00000Z |
| Reason for test | Congenital diarrhoea |
| About the test | We performed a **'trio whole genome sequencing' (trio WGS)** test. This test examines your and Jane's genetic information to try and find a cause for Jane's condition. You can find links to more information about this test at the bottom of this document. |
| Jane's result | **Microvillus inclusion disease**<br>**Gene:** *MYO5B*<br>**Variant:** NM_123456: c.000C>G, p.Arg000Cys and c.000A>C, p.Glu000* |
| Inheritance and recurrence | **Inheritance pattern:** The two *MYO5B* gene variants in Jane have been inherited. Jane has inherited the c.000C>G, p.Arg000Cys from Jill and the c.000A>C, p.Glu000* from John. Jill and John, you are both healthy 'carriers' for microvillus inclusion disease.<br>**Recurrence:** Jill and John, you have a 1 in 4, or 25%, chance of recurrence in each future pregnancy for the two of you. You have options for testing to avoid a recurrence and can discuss these in more detail with your genetics team.<br>Parents · Child(ren) · Genetic variant · AFFECTED 25% chance · CARRIER 50% chance · UNAFFECTED 25% chance |
| What happens next | **Clinical recommendations:** You will be advised by the Gastroenterology team whether any changes to Jane's medication are necessary.<br>**Data storage and re-analysis:** Your and Jane's genomic data will be stored securely and can be re-analysed in the future if new clinical questions arise. |
| Your genetic team | We will work together with the other medical teams involved in Jane's care.<br>**Clinical geneticist:** Dr Clinical Geneticist, T: 1234 5678, E: clinical.geneticist@example.com<br>**Genetic counsellor:** Genetic Counsellor, T: 1234 5678, E: genetic.counsellor@example.com<br>**Genetics follow up:** We will arrange an appointment for you in the Imaginary Clinic in 6 months' time. |
| Community supports | Further resources and community support networks:<br>• Genomics Info - genomicsinfo.org.au<br>• SWAN Australia - swanaus.org.au<br>• Genetic Alliance Australia - geneticalliance.org.au<br>• MedlinePlus Genetics - medlineplus.gov/genetics<br>*Scan the QR code for more information on genomics* |

EXAMPLE

[ACG Family Report, V2 01.04.2021]

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sections pre-filled by study team

sections for clinicians to update with personalised information

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# Evaluation of ‘family reports’ in Acute Care Genomics

Two **online surveys** were designed to elicit views on **report layout, content, and use**

see supplementary material for full survey tools and mapping of questions to constructs

**family survey** RR=33% (n=51/154)

- parents/guardians of ACG participants
- one survey per family
- invitations 12 weeks after result disclosure
- two survey reminders, at 2 and 4 weeks

**clinician survey** RR=53% (n=57/107)

- genetic health professionals caring for patients undergoing urGS via the Acute Care Genomics study
- surveyed at the end of the study
- three survey reminders over two months



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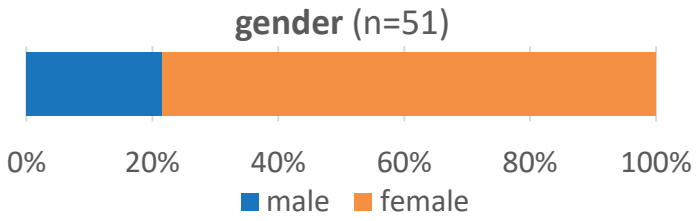
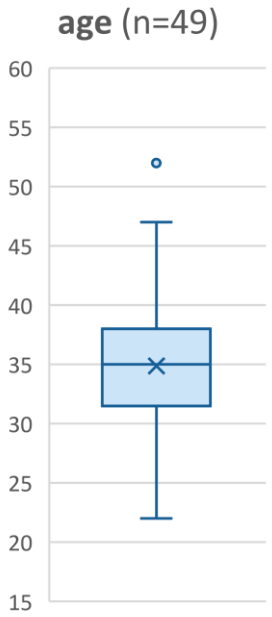
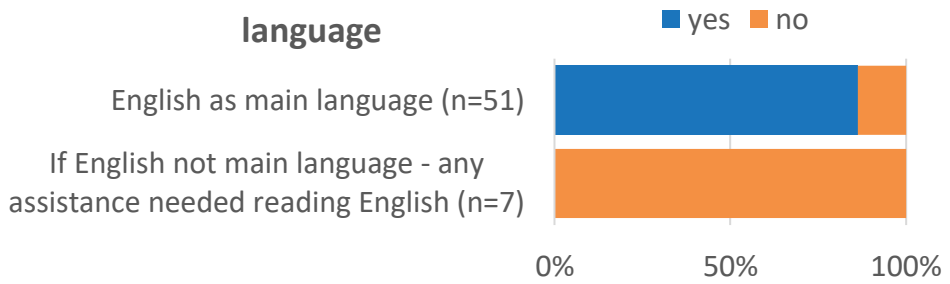
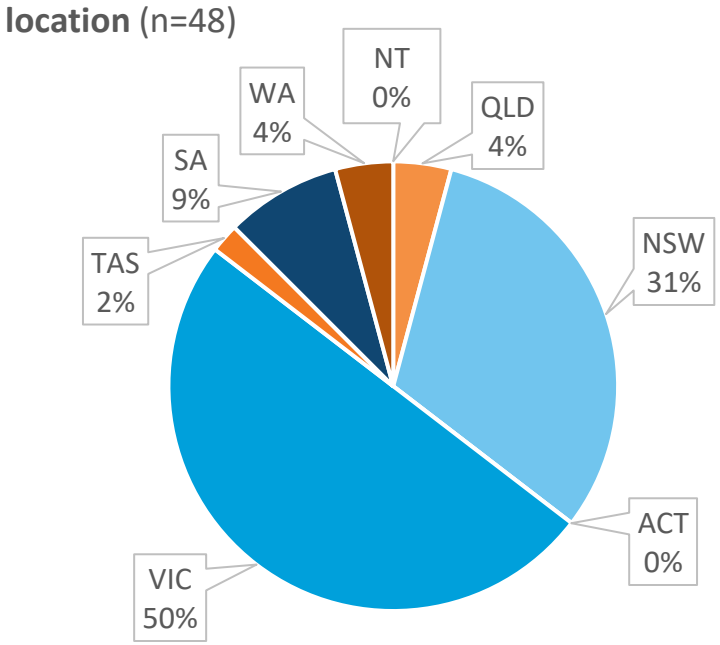
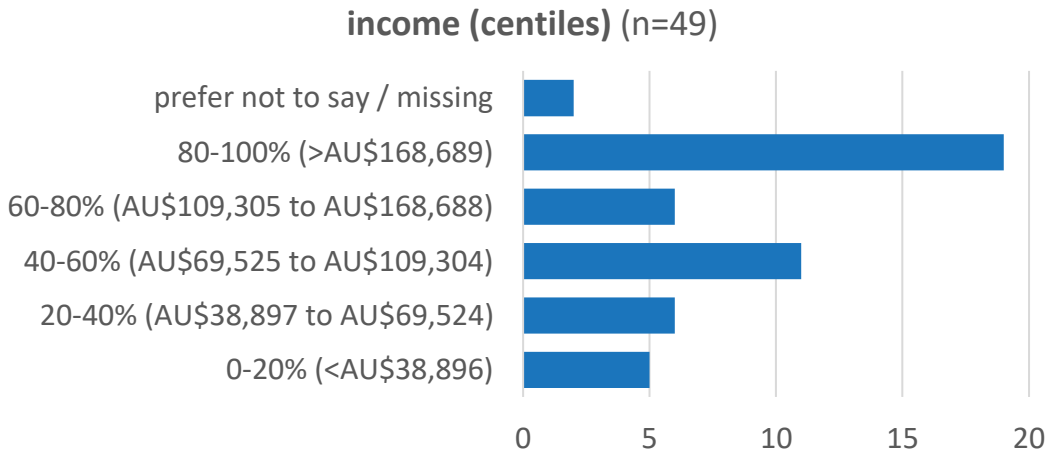
# Family survey respondent demographics

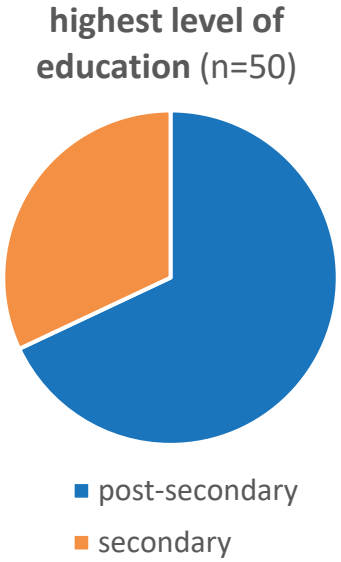
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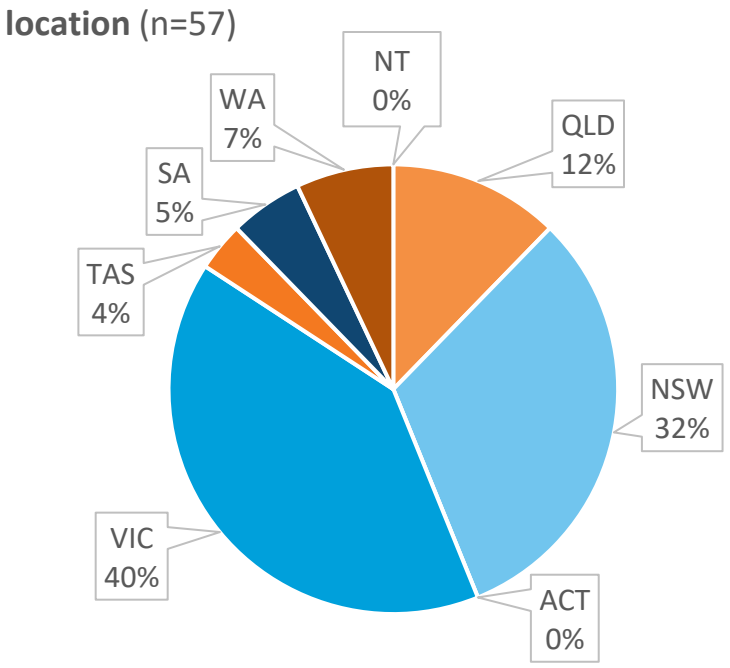
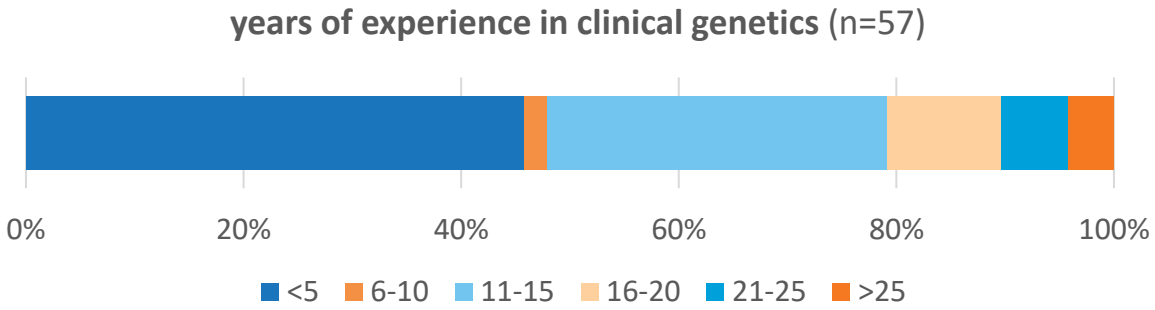
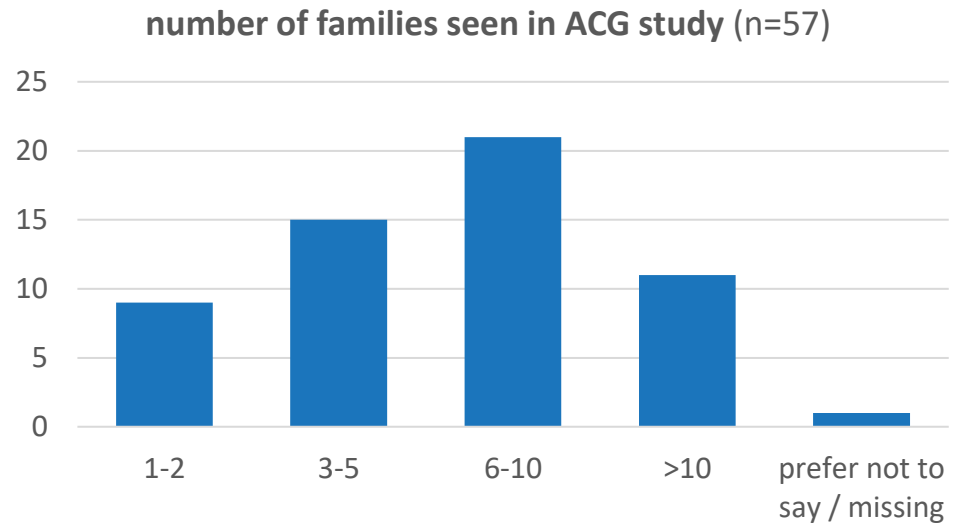
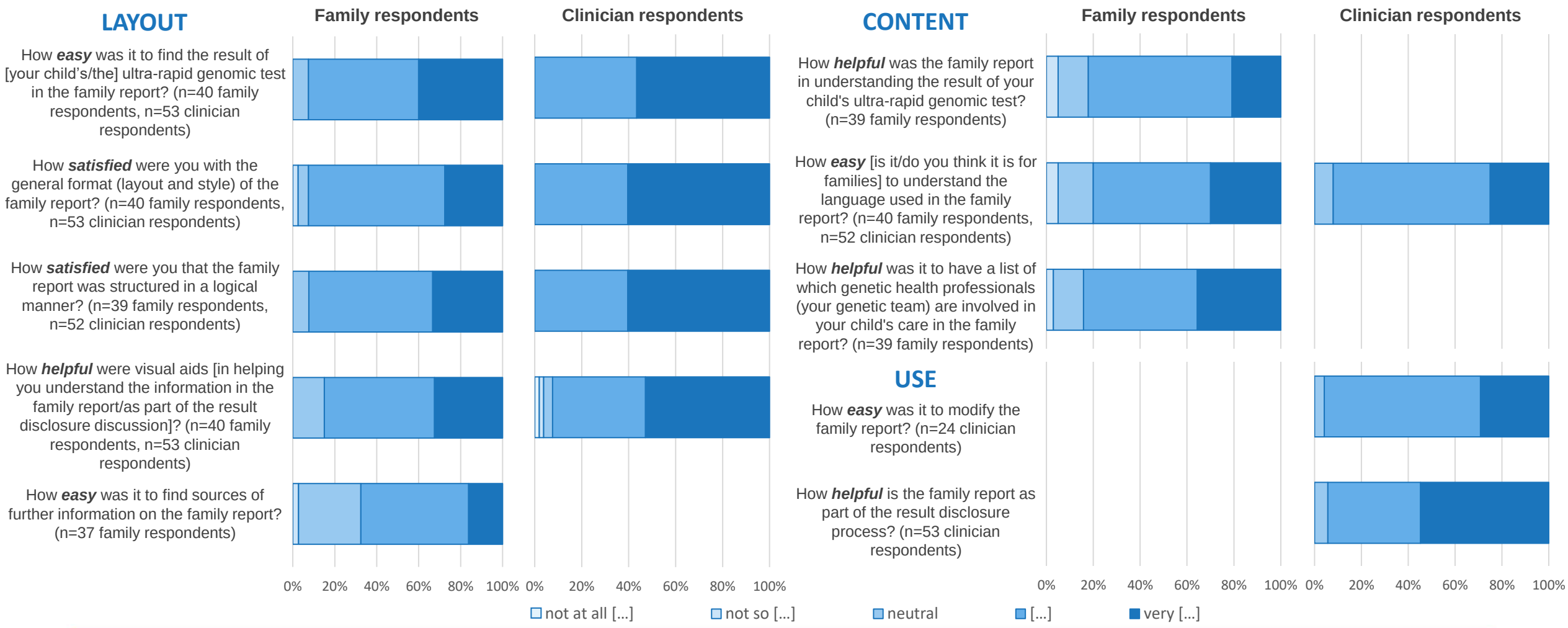
# Clinician survey respondent demographics



# Family and clinician survey responses

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## Family and clinician survey responses

*'We appreciated the extended medical report as well, and having that option was good for us. We shared the family report extensively with our friends and family who we have been updating on our child's condition. They are from diverse backgrounds and those who engaged with the report were generally understanding of its content'* [family respondent, diagnosis achieved]

**shared report with 50+ family/friends**

*'Very helpful to be able to leave the family with a clear, concise report after what is often a very complex appointment in a high stress situation for the family.'* [clinician]

*'bolded sections/headings helpful for key information.'* [family respondent, diagnosis achieved]

*'Much better than scribbling things on paper in front of the family, or trying to guide them through the complex language of laboratory reports.'* [clinician]

*'I understand that can be difficult to provide this information, but I would like to understand which genetic conditions my child was assessed against… I don't know which conditions have been excluded.'* [family respondent, no diagnosis]

## Conclusions & recommendations

- Successful implementation of plain language genomic test reports in a real-world, challenging clinical setting
- Widespread sharing by families and professionals, and adaptation of templates for broader clinical and research use
- **Through co-design, plain language genomic test reports implemented in a real-world acute paediatric setting may facilitate patient/family and caregiver understanding and communication of genomic test purpose, outcome, and potential clinical implications**
- Plain language genomic test reports are not intended to replace laboratory reports or result disclosure discussions with appropriately trained health professionals

**plain language genomic test report templates available here:**

**https://bit.ly/3t6CUw4**

*'I have taken inspiration from these reports to layout reports for other research projects. They have not been used as a direct template, but the flavour of the report has been helpful to generate other reports.'* [clinician]

*'I think these should be adopted much more broadly as they really help families to understand and retain the information in a way that is practical to them.'* [clinician]

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# Acute Care Genomics team

## Plain language genomic test reports:

**Working group and Evaluation:** Gemma Brett, Aisha Ward, Sophie Bouffler, Emma Palmer, Kirsten Boggs, Fiona Lynch, Amanda Springer, Amy Nisselle, Zornitza Stark

**Co-Design:** Australian Genomics Community Advisory Group, Queensland Genomics Community Advisory Group, Syndromes Without A Name

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**Special thanks:** Kim Boycott, Gabriel Recchia

**Australian Genomics:**

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**Project Officer: Sophie Bouffler**

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**NSW Laboratory Lead:** Bruce Bennetts

**SA Laboratory Lead:** Karin Kassahn

**QLD Laboratory Lead:** Ben Lundie

**plain language genomic test report templates available here:**

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**Ethics:** Julian Savulescu, Chris Gyngell, Danya Vears, Lynn Gillam, Fiona Lynch

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