Supplementary Notes 1. Family survey tool.

Section 1: Demographics

This section asks questions about yourself. This can be used to help understand if certain trends in responses are linked to being in a particular group. This could be categorised by location, age, or what language you speak.

Note: as some demographic data could identify individual respondents in areas with few acute care units, we will combine data for some analyses.

1) What is your gender? *Please select one option.*

❑ Male ❑ Female ❑ Other ❑ Prefer not to answer

a. If other, please specify……

2) What is your age? (years)……

3) What is your home postcode?……

4) What is the highest level of education you have completed? *Please select one.*

❑ Year 11 or below / Certificate 3
❑ Year 12 or equivalent / Certificate 4
❑ Bachelor degree
❑ Graduate degree
❑ Postgraduate degree
❑ Other (please specify)……

5) What is the combined income of all adults (including you) in your household per year before tax? *Please select one option.*

❑ Less than $4,900
❑ $5,000 - $9,999
❑ $10,000 - $19,999
❑ $20,000 - $29,999
❑ $30,000 - $39,999
❑ $40,000 - $49,999
❑ $50,000 - $59,999
❑ $60,000 - $69,999
❑ $70,000 - $79,999
❑ $80,000 - $89,999
❑ $90,000 - $119,999
❑ $120,000 - $149,999
❑ $150,000 or more
❑ Prefer not to answer

6) What is your current marital status?

❑ Married
❑ De facto (living with a partner)
❑ Divorced/separated
❑ Widowed
❑ Never married
❑ Other (specify)

a) If other, please specify…

7) How many children do you have?

❑ 0
❑ 1
❑ 2
❑ 3
❑ 4
❑ 5
❑ 6
❑ >6

a) How many of your children are 15 years or younger?

8) Do you have private health insurance?

❑ Yes ❑ No ❑ Unsure

9) **Is English the main language you use?** *Please select one option.*

❑ Yes ❑ No

a. **If no, do you need assistance reading English?**

❑ Yes ❑ No

10) **Do you plan on having more children?** *Please select one option.*

❑ Yes in the next 2 years ❑ Yes in more than 5 years ❑ I am not currently planning to have more children

❑ Yes in the next 5 years ❑ Yes but I'm not sure when ❑ Unsure

Section 2: Genomic Medicine and your Child's Family Report

We are gathering information about your experiences so far with genetic testing and genomic medicine. We will ask you to reflect and comment on your experiences with genomic testing, the layout of the family report, the information available on the family report, how you best understand information, and any recommendations you have to add to the report.

EXPERIENCES WITH GENOMIC TESTING

11) **Before the ultra-rapid genomic testing for your child, did you have any experience with genetic conditions, e.g. personal history, family history, general knowledge?** *Please select one option.*

❑ Yes ❑ No ❑ Unsure

a. **If yes or unsure, please describe, including the name of the genetic condition if you know it……**

12) **Before the ultra-rapid genomic testing for your child, did you have any experience with genetic testing, whether through a GP, specialist, genetics clinic, or an online test, e.g., non-invasive prenatal screening/testing (NIPS/NIPT), carrier/reproductive screening, ancestry testing, etc.?** *Please select one option.*

❑ Yes ❑ No ❑ Unsure

a. **If yes or unsure, which genetic testing have you had experience with?** *Please select all that apply.*

❑ Non-invasive prenatal screening/testing (NIPS/NIPT) → *reveal **question c.***

❑ Ancestry testing

❑ Pharmacogenomic testing

❑ Carrier/reproductive screening → *reveal **question c.***

❑ Nutrigenomic testing

❑ Other→*reveal **question b. and c.***

b. **If other, please list…**

c. **Have you paid for any of these tests?**

❑ Yes ❑ No ❑ Unsure

13) Which of the following best describes the outcome of your child's ultra-rapid genomic test? *Please select one option.*

- ❑ A genetic diagnosis was made
- ❑ More than one genetic diagnosis was made
- ❑ Not sure
- ❑ A partial genetic diagnosis was made (not all of my child's features were explained by the genetic diagnosis)
- ❑ A genetic diagnosis was not made

14) Do you recall receiving a family report with the outcome of your child's ultra-rapid genomic test? *Please select one option.*

- ❑ Yes
- ❑ No → *SURVEY STOP "This survey asks for feedback about the family report. Please contact [relevant Genetic Counsellor] for a copy of the report for your child's ultra-rapid genomic test."*
- ❑ Unsure → *SURVEY STOP "This survey asks for feedback about the family report. Please contact [relevant Genetic Counsellor] for a copy of the report for your child's ultra-rapid genomic test."*

LAYOUT OF THE FAMILY REPORT

15) How easy was it to find the result of your child's ultra-rapid genomic test in the family report? *Please select one option.*

- ❑ Not at all easy
- ❑ Not so easy
- ❑ Neutral
- ❑ Easy
- ❑ Very easy

16) How helpful was the family report in understanding the result of your child's ultra-rapid genomic test? *Please select one option.*

- ❑ Not at all helpful
- ❑ Not so helpful
- ❑ OK
- ❑ Helpful
- ❑ Very helpful

17) How satisfied were you with the general format (layout and style) of the family report? *Please select one option.*

- ❑ Not at all satisfied
- ❑ Not so satisfied
- ❑ Neutral
- ❑ Satisfied
- ❑ Very satisfied

18) How satisfied were you that the family report was structured in a logical manner? *Please select one option.*

- ❑ Not at all satisfied
- ❑ Not so satisfied
- ❑ Neutral
- ❑ Satisfied
- ❑ Very satisfied

19) How helpful were visual aids (e.g. pictures, bolded text, section headings, etc.) in helping you understand the information in the family report? *Please select one option.*

- ❑ Not at all helpful
- ❑ Not so helpful
- ❑ OK
- ❑ Helpful
- ❑ Very helpful

a. Please feel free to comment……

INFORMATION AVAILABLE IN THE FAMILY REPORT

20) How easy is it to understand the language used in the family report? *Please select one option.*

❑ Not at all easy ❑ Not so easy ❑ Neutral ❑ Easy ❑ Very easy

21) Where medical terms are used in the family report, were they explained in a clear manner? *Please select one option.*

❑ Yes ❑ No → *reveal **question a.*** ❑ Unsure → *reveal **question a.***

a. If no or unsure, please explain which terms weren't explained clearly and elaborate if you wish……

22) Does the family report contain any unnecessary information? *Please select one option.*

❑ Yes → *reveal **question a.*** ❑ No ❑ Unsure→ *reveal **question a.***

a. If yes or unsure, please give examples of what information was unnecessary……

[Q21 only for VUS or null result reports that include explanation of test limitations; piping from patient database:]

23) Did your family report explain any limitations of the test? *Please select one option.*

❑ Yes ❑ No ❑ Unsure

24) Did you find it helpful to have a list of which genetic health professionals (your genetic team) are involved in your child's care in the family report? *Please select one option.*

❑ Not at all helpful ❑ Not so helpful ❑ OK ❑ Helpful ❑ Very helpful

25) How easy was it to find sources of further information on the family report? *Please select one option.*

❑ Not at all easy ❑ Not so easy ❑ Neutral ❑ Easy ❑ Very easy

a. Please feel free to comment if there are other types of information you would find helpful……

26) Using the information in the family report, if you were to have another child, what is the chance that the condition would occur again? *Please select one option.*

❑ Less than 1 in 100 (less than 1%) ❑ Less than 1 in 10 (up to 10%) ❑ 1 in 4 (25%) ❑ 1 in 2 (50%) ❑ 3 in 4 (75%) ❑ Definite (100%)

[Q25 will not appear for respondents who chose 'I'm not currently planning to have more children' in Q8:]

27) Using the information in the family report, do you feel you have enough information for future family planning? *Please select one option.*

❑ Yes ❑ No ❑ Unsure

a. If no or unsure, please explain……

28) **Using the information on the family report, would you/have you felt confident explaining the result of your child's ultra-rapid genomic test to someone else e.g. family or friends?** *Please select one option.*

❑ Yes ❑ No ❑ Unsure

a. **If no or unsure, please explain……**

29) **Using the information in the family report, do you feel confident explaining to other family members whether or not they have a chance of being affected by the same condition as your child and/or having a child with the same condition?** *Please select one option.*

❑ Yes ❑ No ❑ Unsure

a. **If no or unsure, please explain……**

30) **Who have you have shared this report with?** *Please select all that apply and provide details of their relationship to you e.g. aunt, social worker. Please do **not** provide individual names or contact details.*

❑ Other Health Professionals ❑ Friends ❑ No one

❑ Family ❑ Other (specify)

a. **If other, please specify……**

HOW YOU BEST UNDERSTAND INFORMATION

31) **When something is explained to you, is it easier to understand fractions (e.g., 'there's a 1 in 2 (1/2) chance that the coin will be heads') or percentages (e.g., 'there's a 50% chance that the coin will be heads')?** *Please select one option.*

❑ Fractions ❑ Percentages ❑ No difference ❑ Not sure

32) **When reading or watching the news, how helpful do you find tables and graphs to explain parts of the story?** *Please select one option.*

❑ Not at all helpful ❑ Not so helpful ❑ OK ❑ Helpful ❑ Very helpful

33) **When you hear a weather forecast, do you prefer predictions using percentages (e.g., 'there will be a 20% chance of rain today') or predictions using only words (e.g., 'there is a small chance of rain today')?** *Please select one option.*

❑ Percentages ❑ Words ❑ No difference ❑ Not sure

34) **Do you have any suggestions or comments about the family report that would help us improve it?** ……

Supplementary Notes 2. Clinician survey tool.

Section 1: About you

1. **Where do you work?** *Please select all hospitals that apply.*
   - ❑ ACT - The Canberra Hospital
   - ❑ NSW - Children's Hospital Westmead
   - ❑ NSW - John Hunter Children's Hospital
   - ❑ NSW - Royal Prince Alfred
   - ❑ NSW - Royal Hospital for Women Randwick
   - ❑ NSW - Sydney Children's Hospital Randwick
   - ❑ NSW - Westmead Hospital (adult)
   - ❑ NT - The Royal Darwin Hospital
   - ❑ QLD - Queensland Children's Hospital
   - ❑ QLD - Royal Brisbane and Women's Hospital
   - ❑ SA - Women's and Children's Hospital
   - ❑ TAS - The Royal Hobart Hospital
   - ❑ VIC - Royal Children's Hospital
   - ❑ VIC - Monash Health
   - ❑ VIC - Royal Women's Hospital
   - ❑ WA - King Edward Memorial Hospital
   - ❑ WA - Perth Children's Hospital
   - ❑ Other
     a. **If other, please specify:**

2. **What is your primary role?** *Please select one option.*

   ○ Clinical Geneticist ○ Clinical Genetics trainee ○ Genetic Counsellor

3. **How many years of professional experience in clinical genetics do you have?** *Please select one option.*

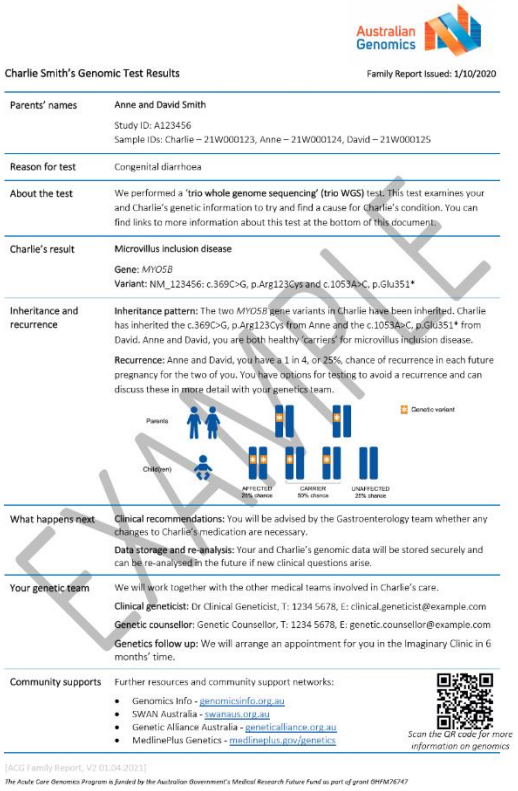
   ○ <5 ○ 6-10 ○ 11-15 ○ 16-20 ○ 21-25 ○ >25

4. **How many families have you seen as part of the Acute Care Genomics program <u>from 2020 onwards</u>?** *Please select one option.*

   ○ 1-2 ○ 3-5 ○ 6-10 ○ >10

Section 2: Your feedback on the plain language family report

This survey asks for feedback on the Acute Care Genomics plain language family report. See below for an example of the family report:



Charlie Smith's Genomic Test Results — Family Report Issued: 1/10/2020

Parents' names	Anne and David Smith Study ID: A123456 Sample IDs: Charlie – 21W000123, Anne – 21W000124, David – 21W000125
Reason for test	Congenital diarrhoea
About the test	We performed a **'trio whole genome sequencing' (trio WGS)** test. This test examines your and Charlie's genetic information to try and find a cause for Charlie's condition. You can find links to more information about this test at the bottom of this document.
Charlie's result	**Microvillus inclusion disease** **Gene:** *MYO5B* **Variant:** NM_123456: c.369C>G, p.Arg123Cys and c.1053A>C, p.Glu351*
Inheritance and recurrence	**Inheritance pattern:** The two *MYO5B* gene variants in Charlie have been inherited. Charlie has inherited the c.369C>G, p.Arg123Cys from Anne and the c.1053A>C, p.Glu351* from David. Anne and David, you are both healthy 'carriers' for microvillus inclusion disease. **Recurrence:** Anne and David, 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.
What happens next	**Clinical recommendations:** You will be advised by the Gastroenterology team whether any changes to Charlie's medication are necessary. **Data storage and re-analysis:** Your and Charlie'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 Charlie's care. **Clinical geneticist:** Dr Clinical Geneticist, T: 1234 5678, E: clinical.geneticist@example.com **Genetic counsellor:** Genetic Counsellor, T: 1234 5678, E: genetic.counsellor@example.com **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: • Genomics Info - genomicsinfo.org.au • SWAN Australia - swanaus.org.au • Genetic Alliance Australia - geneticalliance.org.au • MedlinePlus Genetics - medlineplus.gov/genetics *Scan the QR code for more information on genomics*

Parents; Child(ren); Genetic variant; AFFECTED 25% chance; CARRIER 50% chance; UNAFFECTED 25% chance

[ACG Family Report, V2 01.04.2021]

The Acute Care Genomics Program is funded by the Australian Government's Medical Research Future Fund as part of grant GHFM76747

EXAMPLE

USE OF THE FAMILY REPORT

5. **Have you used a family report with any families from the Acute Care Genomics program?** *Please select one option.*

○ Yes ○ No → ***EXIT:*** Thank you for your interest in our study but this survey is only for health professionals who have used the family report.

6. **How have you used the family report(s)?** *Please select all that apply.*
   - ❑ At the start of the result disclosure consultation, to guide the discussion
   - ❑ During the result disclosure consultation, to help families' understanding and recollection
   - ❑ At the end of the result disclosure consultation, to give families a written/visual record to take as a summary
   - ❑ Other
     a. **If other, please tell us more…**

7. **How helpful is the family report as part of the result disclosure process?** *Please select one option.*

○ Not at all helpful ○ Not so helpful ○ OK ○ Helpful ○ Very helpful

   a. **Please feel free to comment on the helpfulness of the family report…**

8. **Have you distributed a family report to anyone else apart from the family?** E.g., Intensive care team

○ Yes ○ No ○ Unsure

   a. **If yes, who did you distribute it to and why? …**

9. **Have you used the family report templates outside the Acute Care Genomics program?**

○ Yes ○ No ○ Unsure

   a. **If yes, please tell us more…**

LAYOUT OF THE FAMILY REPORT

10. **How easy was it to find the result of the ultra-rapid genomic test in the family report?** *Please select one option.*

○ Not at all easy ○ Not so easy ○ Neutral ○ Easy ○ Very easy

11. **How satisfied were you with the general format (layout and style) of the family report?** *Please select one option.*

○ Not at all satisfied ○ Not so satisfied ○ Neutral ○ Satisfied ○ Very satisfied

12. **How satisfied were you that the family report was structured in a logical manner?** *Please select one option.*

○ Not at all satisfied ○ Not so satisfied ○ Neutral ○ Satisfied ○ Very satisfied

13. **How helpful were visual aids (e.g., pictures, bolded text, section headings, etc.) as part of the result disclosure discussion?** *Please select one option.*

○ Not at all helpful ○ Not so helpful ○ OK ○ Helpful ○ Very helpful

14. **Please feel free to comment on any aspect of the layout…**

INFORMATION AVAILABLE IN THE FAMILY REPORT

15. **How easy do you think it is for families to understand the language used in the family report?** *Please select one option.*

○ Not at all easy ○ Not so easy ○ Neutral ○ Easy ○ Very easy

16. **Where medical terms are used in the family report, do you think they are explained in a sufficiently clear manner for families?** *Please select one option.*

○ Yes ○ No ○ Unsure

   a. **If no or unsure, please explain which terms aren't explained clearly and elaborate if you wish…**

17. **Does the family report contain any unnecessary information for families?** *Please select one option.*

○ Yes ○ No ○ Unsure

   a. **If yes or unsure, please give examples of what information was unnecessary……**

18. **Did you modify the family report beyond adding the details of the genetics team?** E.g., community supports, clinical recommendations.

○ Yes ○ No ○ Unsure

a. **If yes, how easy was it to modify the family report?**

○ Not at all easy ○ Not so easy ○ Neutral ○ Easy ○ Very easy

b. **What did you modify and why? …**

FINAL COMMENTS

19. **Please feel free to provide any other comments about the family report.** For example, any differences in how you used the report for informative or uninformative results.

Supplementary Table 1. Demographics of family respondents.

Gender (n=51)	**n (%)**
female	40 (78)
male	11 (22)
Age (n=49)	**mean (range)**
in years	34.9 (22-52)
Location (state/territory) (n=48)	**n (%)**
Australian Capital Territory	0 (0)
New South Wales	15 (31)
Northern Territory	0 (0)
Queensland	2 (4)
South Australia	4 (8)
Tasmania	1 (2)
Victoria	24 (50)
Western Australia	2 (4)
Highest level of education (n=50)	**n (%)**
secondary	16 (32)
post-secondary	34 (68)
Income (centiles) (n=49)	**n (%)**
0-20% (< AU$38,896)	5 (10)
20-40% (AU$38,897 to AU$69,524)	6 (12)
40-60% (AU$69,525 to AU$109,304)	11 (22)
60-80% (AU$109,305 to AU $168,688)	6 (12)
80-100% (>AU$168,689)	19 (39)
prefer not to say / missing	2 (4)
English as main language (n=51)	**n (%)**
yes	44 (86)
no	7 (17)
If English not main language – any assistance needed reading English (n=7)	**n (%)**
yes	0 (0)
no	7 (100)
Planning more children (n=51)	**n (%)**
Yes in the next 2 years	8 (16)
Yes in the next 5 years	3 (6)
Yes in more than 5 years	0 (0)
Yes but I'm not sure when	9 (8)
I am not currently planning to have more children	0 (0)
Unsure	9 (18)

Supplementary Table 2. Demographics of clinician respondents (n=57).

Location (state/territory)	**n (%)**
Australian Capital Territory	0 (0)
New South Wales	18 (32)
Northern Territory	0 (0)
Queensland	7 (12)
South Australia	3 (5)
Tasmania	2 (4)
Victoria	23 (40)
Western Australia	4 (7)
Years of professional experience in clinical genetics	**n (%)**
<5	22 (39)
6-10	10 (18)
11-15	15 (26)
16-20	5 (9)
21-25	3 (5)
>25	2 (4)
Number of families seen in ACG study	**n (%)**
1-2	9 (16)
3-5	15 (26)
6-10	21 (37)
> 10	11 (19)
prefer not to say / missing	1 (2)

Supplementary Table 3. Family and clinician responses to Likert scale questions regarding layout, content and use of family reports. Qualifying term represented by […] in column heading is in ***bold/italics*** within respective question; data reported as n (%).

	not at all […]	not so […]	neutral	[…]	very […]
Layout of family reports					
How ***easy*** was it to find the result of your child's ultra-rapid genomic test in the family report? (n=40 family respondents)	0 (0)	0 (0)	3 (8)	21 (53)	16 (40)
How ***satisfied*** were you with the general format (layout and style) of the family report? (n=40 family respondents)	0 (0)	1 (3)	2 (5)	26 (65)	11 (28)
How ***satisfied*** were you that the family report was structured in a logical manner? (n=39 family respondents)	0 (0)	0 (0)	3 (8)	23 (59)	13 (33)
How ***helpful*** were visual aids (e.g. pictures, bolded text, section headings, etc.) in helping you understand the information in the family report? (n=40 family respondents)	0 (0)	0 (0)	6 (15)	21 (53)	13 (33)
How ***easy*** was it to find sources of further information on the family report? (n=37 family respondents)	0 (0)	1 (3)	11 (30)	19 (51)	6 (16)
How ***easy*** was it to find the result of the ultra-rapid genomic test in the family report? (n=53 clinician respondents)	0 (0)	0 (0)	0 (0)	23 (43)	30 (57)
How ***satisfied*** were you with the general format (layout and style) of the family report? (n=53 clinician respondents)	0 (0)	0 (0)	0 (0)	21 (40)	32 (60)
How ***satisfied*** were you that the family report was structured in a logical manner? (n=52 clinician respondents)	0 (0)	0 (0)	0 (0)	21 (40)	32 (60)
How ***helpful*** were visual aids (e.g., pictures, bolded text, section headings, etc.) as part of the result disclosure discussion? (n=53 clinician respondents)	1 (2)	1 (2)	2 (4)	21 (40)	28 (53)
Content of family reports					
How ***helpful*** was the family report in understanding the result of your child's ultra-rapid genomic test? (n=39 family respondents)	0 (0)	2 (5)	5 (13)	24 (62)	8 (21)
How ***easy*** is it to understand the language used in the family report? (n=40 family respondents)	0 (0)	2 (5)	6 (15)	20 (50)	12 (30)
How ***helpful*** was it to have a list of which genetic health professionals (your genetic team) are involved in your child's care in the family report? (n=39 family respondents)	0 (0)	1 (3)	5 (13)	19 (49)	14 (36)
How ***easy*** do you think it is for families to understand the language used in the family report? (n=52 clinician respondents)	0 (0)	0 (0)	4 (7)	35 (67)	13 (25)
Use of family reports					
How ***easy*** was it to modify the family report? (n=24 clinician respondents)	0 (0)	0 (0)	1 (4)	16 (67)	7 (29)
How ***helpful*** is the family report as part of the result disclosure process? (n=53 clinician respondents)	0 (0)	0 (0)	3 (6)	21 (40)	29 (55)