Ethics Exchange – Consent in challenging circumstances

Is there such a thing as informed consent? Lessons from obtaining consent for genetic and genomic testing

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Australian Genomics, Sydney Children's Hospital Network

28th June 2023

What do Genetic Counsellors do?

Talk to families about genetic conditions – work in adult, paed, prenatal, cancer and research settings

Help families adjust
Help families make difficult decisions

Take families through tailored consent process
Managing expectations
Discussing possible implications

How do we approach consent?

Genetics – full of very technical language
Need to balance information giving

Right amount for family to make decision on whether to proceed with testing – different environments call for different methods

Adult onset condition e.g. Huntington's disease vs. family with patient in NICU

What do we talk about when obtaining consent?

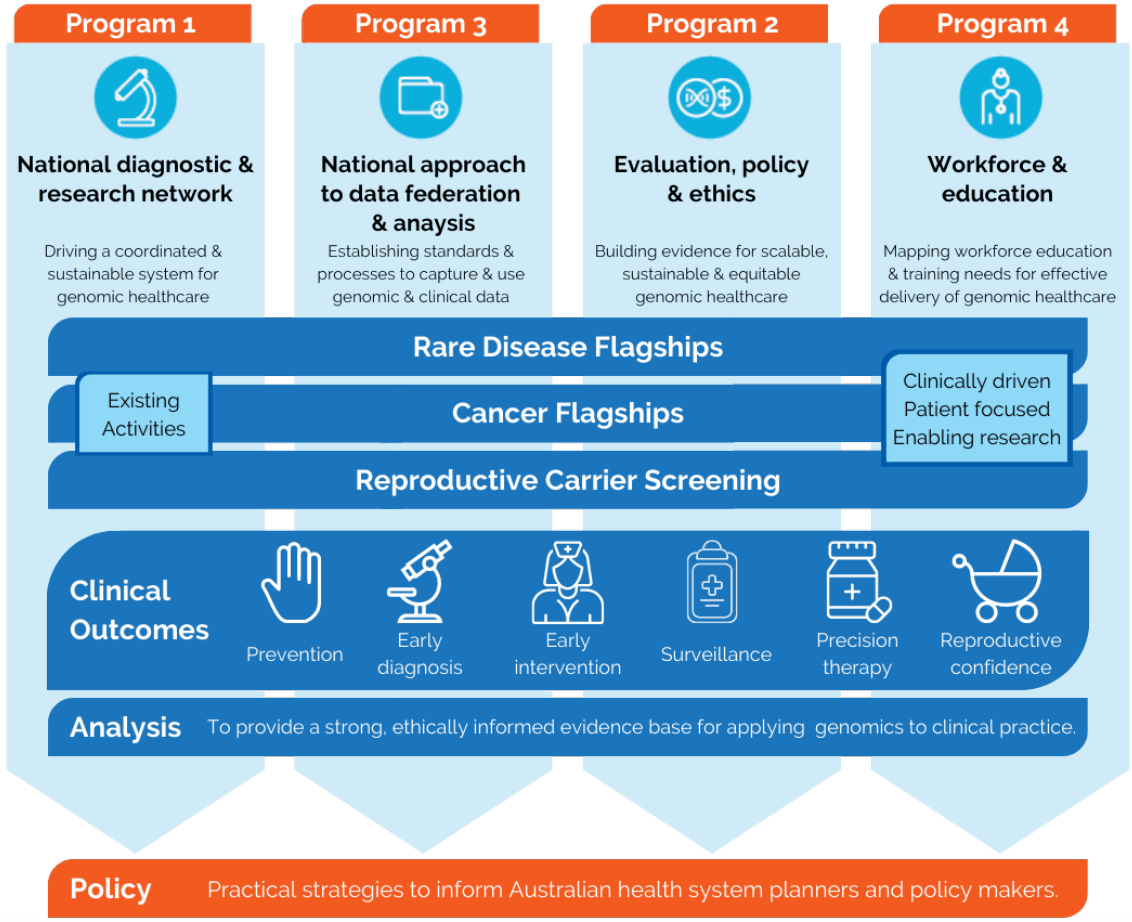
Genetics/biology lesson
Types of results – diagnosis, no diagnosis, VUS
Emotional impact of results
Unexpected family relationships - misattributed parentage, unknown consanguinity
Insurance implications
Data storage and privacy
Research implications

Australian Genomics

NHMRC GENOMICS Targeted Call – 2016-2023

Preparing Australia for Genomic Medicine – a health services research project

- Demonstrate how **patient benefit** could be maximized through application of genomic data in one or more human diseases.
- Provide evidence to inform analysis on the **cost effectiveness** of implementing genomic data into the Australian health system.
- A significant increase in the understanding of **practical strategies** that could be used by Australian health system planners and policymakers.
- Building Australia's research and **research translation capacity** in the area of genomics and healthcare.



A national network

5,477 Patients recruited / genomic testing

$99 million Research supported

129 Peer-reviewed papers
21 Reports

32 Recruitment sites

450 Collaborators & Investigators
103 Organisational partners

723 Presentations
54 Workshops & conferences

100% Results returned

117 Jobs created
55 PhD / Masters research students trained

23 Sub-projects in Genomic Health Service Delivery

115 Ethics & site submissions per year

10 Advisory Boards

13 Policy submissions
4 Position statements

21 Cancer & Rare Disease studies

Australian Genomics

How do we approach consent?

16 page PIS probably not helpful?

Understand ethical requirement – perhaps a summary document?

Tools AG has developed to help –
Acute Care study had 1 page consent summary + e-consent
CTRL online consent portal
Genetics Adviser
National standardised forms

Medical Research Future Fund
Rapid Applied Research Translation

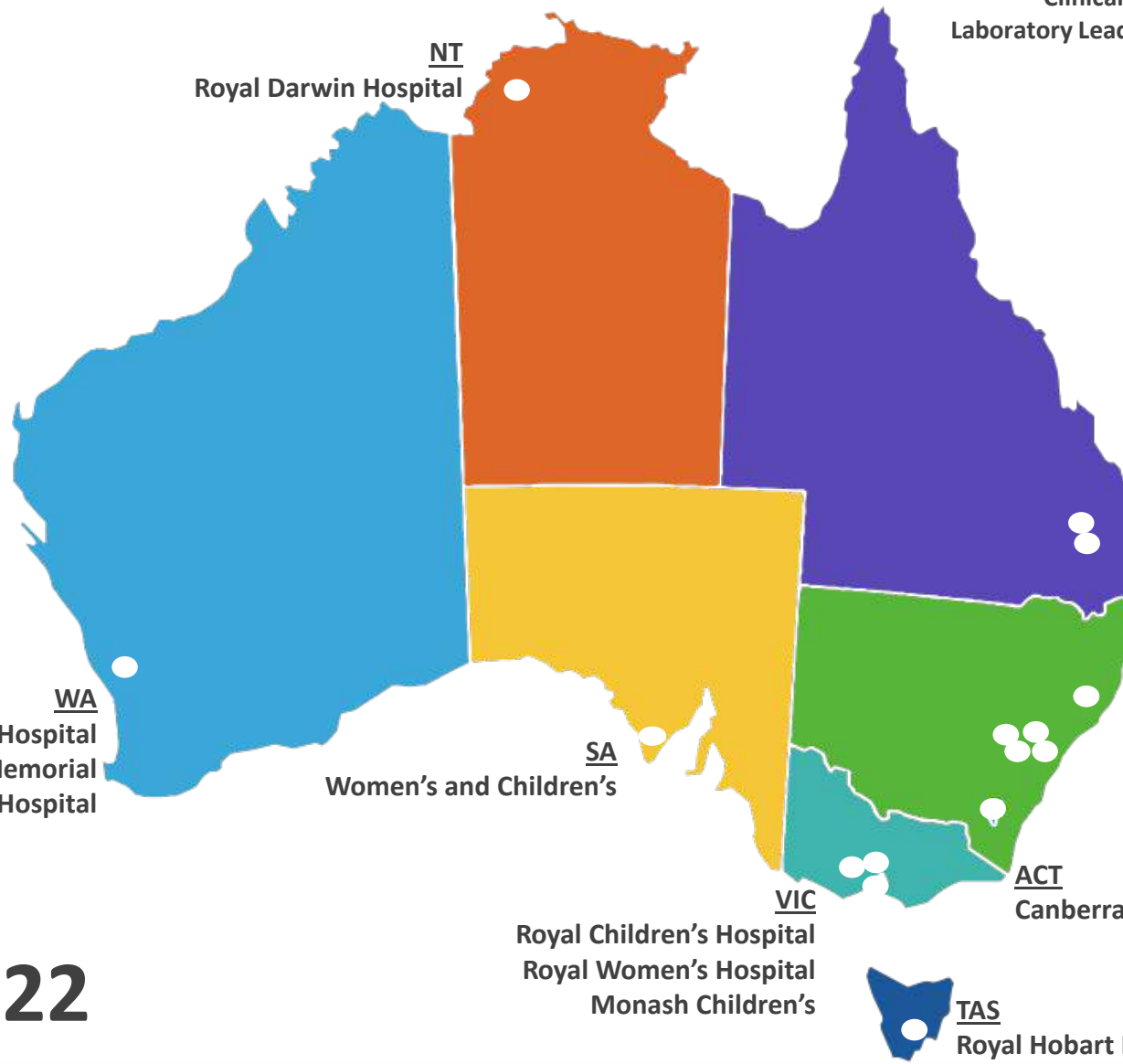
The Royal Children's Hospital Foundation

The Sydney children's Hospitals Network
care, advocacy, research, education

Queensland Genomics

CHILDREN'S RESEARCH FOUNDATION

Clinical Lead: Prof Zornitza Stark
Laboratory Lead: A/Prof Sebastian Lunke



NT
Royal Darwin Hospital

QLD
Royal Brisbane and Women's
Queensland Children's Hospital

NSW
Sydney Children's Hospitals Network
(Westmead and Randwick)
Westmead (Adult)
John Hunter Children's Hospital
Royal Hospital for Women
Royal Prince Alfred

ACT
Canberra Hospital

WA
Perth Children's Hospital
King Edward Memorial
Hospital

SA
Women's and Children's

VIC
Royal Children's Hospital
Royal Women's Hospital
Monash Children's

TAS
Royal Hobart Hospital

ACG: 2018-22

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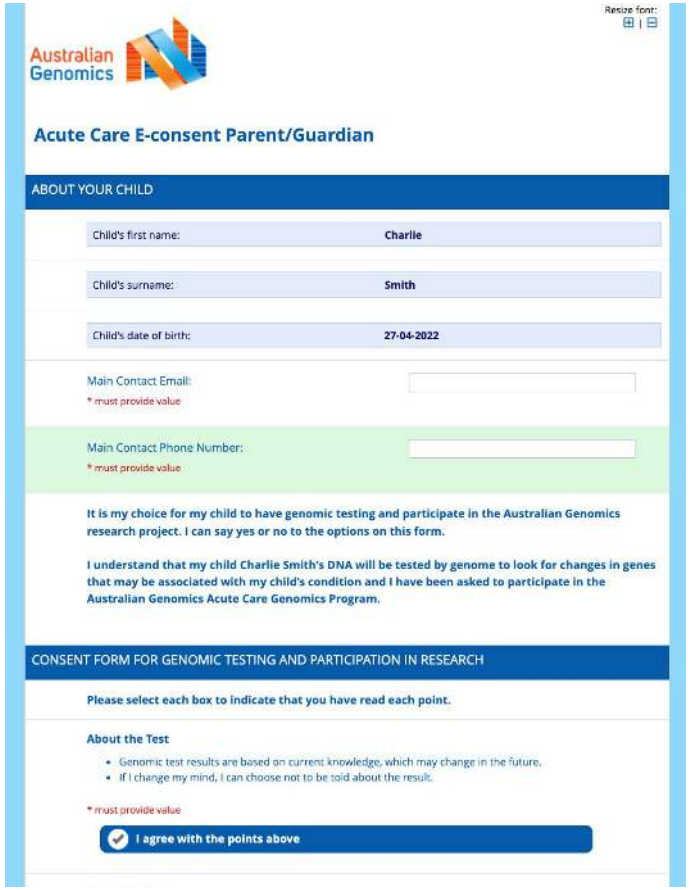
Supporting families: consent



Australian Genomics

Resize font:

Acute Care E-consent Parent/Guardian

ABOUT YOUR CHILD

Child's first name:	Charlie
Child's surname:	Smith
Child's date of birth:	27-04-2022

Main Contact Email:

* must provide value

Main Contact Phone Number:

* must provide value

It is my choice for my child to have genomic testing and participate in the Australian Genomics research project. I can say yes or no to the options on this form.

I understand that my child Charlie Smith's DNA will be tested by genome to look for changes in genes that may be associated with my child's condition and I have been asked to participate in the Australian Genomics Acute Care Genomics Program.

CONSENT FORM FOR GENOMIC TESTING AND PARTICIPATION IN RESEARCH

Please select each box to indicate that you have read each point.

About the Test

- Genomic test results are based on current knowledge, which may change in the future.
- If I change my mind, I can choose not to be told about the result.

* must provide value

I agree with the points above

Electronic consent

Australian Genomics

Acute Care Genomics Program

Parent/Guardian Information Statement

About this project

We are looking at how useful ultra-rapid genomic testing is for the diagnosis and management of critically ill babies and children with suspected genetic conditions. This study will help to find out when we should use ultra-rapid genomic testing and the most appropriate and cost-effective way to provide it in the future.

Important points

- Participation in this project is voluntary.
- Take the time you need to decide if you want to participate.
- You will have the chance to ask questions of the study team and other health professionals.
- Your and your child's participation in this research project will be recorded in your child's health records.
- You can withdraw at any time without affecting your child's care.

What does participation involve?

Meeting 1 – Information about the study and consent:

You will be asked if you are interested in participating in this study. Participation involves a test, called 'trio whole genome sequencing', of both your and your child's entire genetic code. The test is done on a sample of your and your child's blood, which will be stored according to laboratory guidelines. There will be no out-of-pocket expenses for this genomic test.

The test aims to check if your child may have a genetic condition to explain his or her health problems and we expect a result will be available within 5 days.

You may provide your consent on the day or later if you need more time to decide.

Meeting 2 – Return of results

The results of your child's genomic test will be provided to you. The test may or may not give a genetic diagnosis.

Other research

You will be contacted in the future regarding opportunities for further analysis of the genomic data. You can decide if you want to have further analysis at that time.

You may receive surveys asking your opinion about aspects of this study.

How will we collect information about your child?

As part of your and your child's participation in this project, we will gather information about your child's healthcare, including from their hospital records. This helps us to learn whether genomic testing is useful for your child, or those with the same condition.

What are the possible results of the genomic test?

- A cause for your child's condition(s) may be found.
- A cause for your child's condition(s) may not be found.
- A result of *'uncertain significance'* may be identified, which means it cannot be understood using current available information.

There is a small chance of **unexpected findings**, such as:

- Other unrelated medical conditions.
- Unexpected family relationships.

This test will not predict all future health problems.

The test result may affect your or your child's ability to obtain some types of insurance (such as income protection or life insurance). It does not affect health insurance.

What happens to information about me and my child?

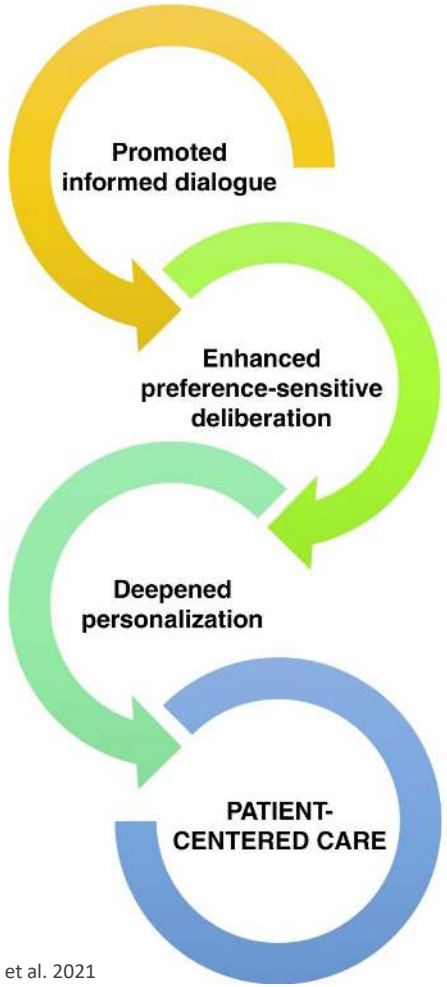
Samples, genomic data, and related health information that can identify you or your child, obtained in connection with this research:

- Will remain confidential, except as required by law.
- Will be stored securely in a study database. Only the doctors, researchers, and other personnel working directly with this study will have access to the database.
- May be shared to advance scientific knowledge. It will be shared in a way that protects your privacy ('de-identified'). This may include sharing on large national or international databases to help improve understanding of related conditions by comparing your and your child's results to those from other people.
- May go on to further research to try to find a diagnosis and/or understand your child's condition.
- May be released to genetic services to help with the care of other family members.

Australian Genomics

Ayres, S. et al. (2019) Genetic counseling in pediatric acute care: Reflections on ultra-rapid genomic diagnoses in neonates, J Genet Couns.
Lynch, F., et al. (2020) Rapid acute care genomics: Challenges and opportunities for genetic counsellors, J Genet Couns.

Collaboration with Genetics Adviser



- Promoted engagement during the course of the session
- Facilitated deliberation around the participants' preferences and perceived harms and benefits
- Enabled personal reflection on a wide set of experiences and circumstances

Shickh et al. 2021

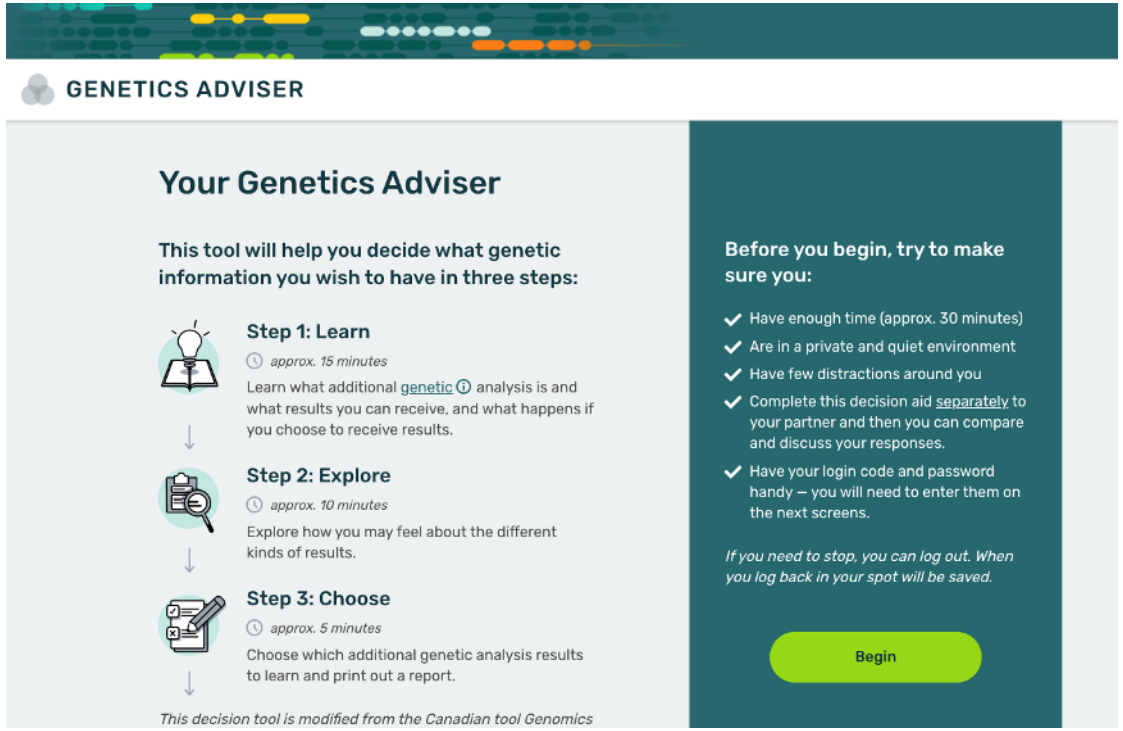
› Genet Med. 2021 Mar 2. doi: 10.1038/s41436-021-01112-1. Online ahead of print.

The role of digital tools in the delivery of genomic medicine: enhancing patient–centered care

Salma Shickh [#1,2], Sara A Rafferty [#3,4], Marc Clausen [2], Rita Kodida [2], Chloe Mighton [1,2], Seema Panchal [5], Justin Lorentz [6], Thomas Ward [5], Nicholas Watkins [5], Christine Elser [7,8], Andrea Eisen [6], June C Carroll [5,9], Emily Glogowski [10], Kasmintan A Schrader [11,12], Jordan Lerner-Ellis [13,14,15], Raymond H Kim [3,7,8], David Chitayat [3,4,5], Cheryl Shuman [3,4], Yvonne Bombard [16,17], Incidental Genomics Study Team

Collaborators, Affiliations + expand

PMID: 33654192 DOI: 10.1038/s41436-021-01112-1



Marc Clausen

Yvonne Bombard

What is dynamic consent?

"personalised, online consent and communication platforms that provide a consent mechanism as well as a means of ongoing communication… an information and communications technology (ICT) rather than paper-based tool… participant-centred, granular, flexible and can be adapted…" (Prictor et al., 2019)

Benefits to patients and research participants

- Engagement
- Tailored information
- Autonomy and transparency
- Understanding of medical services and research
- Trust in research and sharing of health data

Examples of dynamic consent in use

- CTRL (‘control’) Australian Genomics (AUS)
  - Cardiovascular Genetic Disorders Flagship
  - Mackenzie’s Mission
  - UDN-Aus
  - KidGen
- RUDY rare disease study (UK)
- PEER (Platform for Engaging Everyone Responsibly) Genetic Alliance (USA)
- CHRIS longitudinal study and biobank (ITA)

A role for dynamic consent in challenging circumstances

- When information delivery is best spaced out over time
- Participants have higher information needs
- Information should be tailored to individuals needs
- Participants are geographically distant from study site
- Participants have high privacy or data sharing concerns
- New or updated consent is required over time
- Participants may otherwise be lost to follow up

Thanks to Dr Matilda Haas, AG Research Projects & Partnerships Manager for these slides

National Consent Working Group

Julie McGaughran	QLD	Clinical Geneticist (Project Lead)
Keri Finlay	VIC	Project Coordinator
Laura Purcell	QLD	Project Officer
Ben Kamien	WA	Clinical Geneticist (paediatric/adult/prenatal)
Ainsley Newson	NSW	Research, bioethics/legal
Vanessa Fitzgerald	NSW	Genomics Principal Project Officer, NSW MoH
Alex Brown	SA	Research; Aboriginal and Torres Strait Islander Advisory Group
Nicola Poplawski	SA	Clinical Geneticist (cancer)
Danya Vears	VIC	Research, bioethics
Ivan Macciocca	VIC	Genetic Counsellor (paediatric/rare disease/cardiac)
Tony Roscioli	NSW	Clinical Geneticist (prenatal)
Chiyan Lau	QLD	Genetic Pathologist
John Cannings	NSW	Community Advisory Group
Elly Lynch	NT	Genetic Counsellor

Multi-disciplinary, cross-jurisdictional expert working group

- Including representatives from Community Advisory Group, Aboriginal and Torres Strait Islander Advisory Group, NSW MoH consent form project.

National Clinical Consent for Genetic and Genomic Testing

Key issues:

- States and territories have individualised approach to genetics and genomic testing consent
- May be a barrier to the flow of health information across states or jurisdictional borders as families dispersed across country
- Inconsistency in the patient experience, outcomes and management and data sharing

What has been happening in the last few years?

- There has been attempts to create a national form, which have been implemented with varying degrees of success
  - Australian Genomics and NSW Ministry of Health consent materials
- Has been developments in data/sample storage and sharing, data re-analysis, patient re-contact, withdrawal of consent, future research options
  - Current forms need updating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Thank you!

Questions?

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