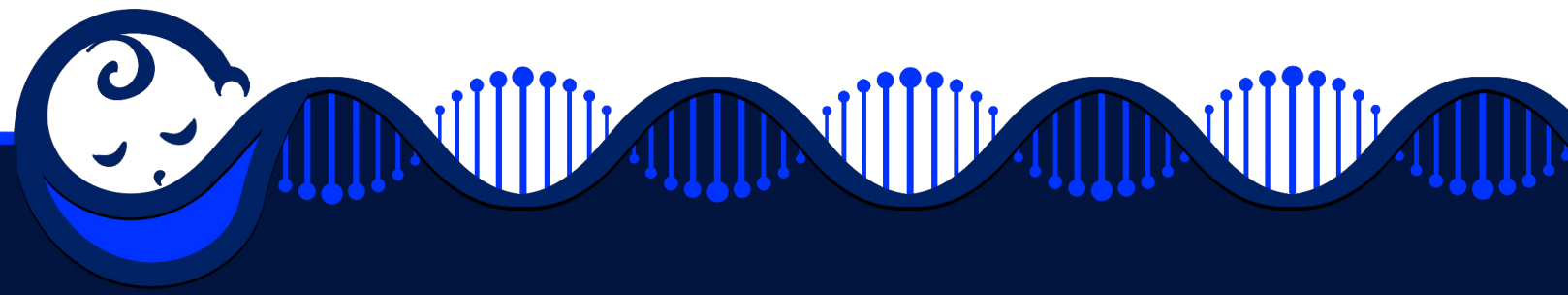
UNIVERSITY OF WOLLONGONG AUSTRALIA

gEnomics4newborns

THE UNIVERSITY OF SYDNEY

National Australian citizens' jury on using genomics in newborn bloodspot screening

INFORMATION BOOKLET



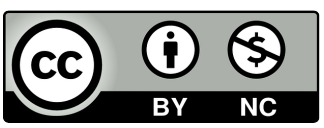
AUSTRALIAN CENTRE FOR HEALTH ENGAGEMENT EVIDENCE AND VALUES, UNIVERSITY OF WOLLONGONG

National Australian citizens' jury on using genomics in newborn bloodspot screening: Information Booklet. 2025.

Australian Centre for Health Engagement, Evidence and Values (ACHEEV), University of Wollongong, NSW, Australia.

ACHEEV was established in January 2019. Our work focuses on health: the health of people, other animals, society and the planet, and how all of these things are connected. Our research is rigorous, interdisciplinary and independent. We are not afraid to ask difficult questions, and we have the skills, knowledge and experience to generate meaningful answers that can guide policy and practice. Our mission is to make health systems more inclusive and democratic. In everything we do, we ask how we can work towards greater justice and equity.

https://www.uow.edu.au/the-arts-social-sciences-humanities/research/acheev/



FOREWORD

Welcome to the National Australian citizens' jury on using genomics in newborn bloodspot screening. We are grateful that you have accepted our invitation to be part of this national event. Newborn bloodspot screening is a trusted and effective program in Australia. Genomic technologies are developing rapidly. Some experts argue genomics should be used in newborn bloodspot screening in future; others say we should be cautious. This jury will show what an informed group of Australians, a group including many kinds of people, think about these possible changes. This jury will answer the question:

> **Under what circumstances, if any, should Australia use genomics in the newborn bloodspot screening program, to ensure the program remains trustworthy and effective?**

We look forward to meeting you online on 9 March 2025, and in person on 28 March 2025 in Canberra.

WHAT IS THE PURPOSE OF THIS BOOKLET?

To help you prepare for the event, we have put together this information booklet. It explains what a citizens' jury is. It also introduces you to newborn screening, genomics, and the changes to newborn screening government is currently considering. You can use this booklet before, during and after the event.

You don't need any prior knowledge to take part in the jury. Just read this booklet and start to think about what we are asking you to consider. You and your fellow participants are not expected to be experts on this topic. You will most likely have further questions after reading this booklet. We encourage you to bring your questions to the jury, along with your insights and views. These are all critical to the discussions you will have with your fellow jurors.

ACKNOWLEDGEMENT OF COUNTRY

We acknowledge the Ngunnawal people as traditional custodians of the land we meet on in-person. We pay our respects to their Elders past and present, and to the children of today who are the Elders of the future.

We acknowledge all Aboriginal and Torres Strait Islander peoples across the Australian continent. From their lands, we will join the citizens' jury remotely. We pay respects to all Aboriginal and Torres Strait Islander people, their Elders past and present, and to their children of today who are the Elders of the future.

We wish to acknowledge the Aboriginal and Torres Strait Islander people who have had their genetic material taken or their genetic data used, without their consent. We pay respect to our Aboriginal and Torres Strait Islander colleagues who have guided us in this project and thank them for their generosity.

ACKNOWLEDGEMENT OF FAMILIES OF CHILDREN WITH RARE DISEASES

We wish to acknowledge the parents of babies and children with rare health conditions. The uncertainty and challenges you face whilst continuing to support your child's growth and development shows your strength and love. To parents who have lost their children due to rare health conditions, we acknowledge your pain and grief.

ACKNOWLEDGMENT OF SUPPORTING ORGANISATIONS AND INDIVIDUALS

Our thanks to members of the Expert Reference Group for the Citizens' Jury and the Consumer Advisory Panel for the gEnomics4newborns project for their guidance.

Our thanks to the expert witnesses for their invaluable contributions: Dr Kaustuv Bhattacharya, Professor Bruce Bennetts, Dr Kristen Nowak, Professor Zornitza Stark, Professor Kristi Jones, Professor Margaret Otlowski and Professor Ainsley Newson. Special thanks to the parents

who have shared their life stories with us. Thanks also to all the Chief and Assistant Investigators on the gEnomics4newborns research project and their teams who have contributed to the design of the jury and the text in this booklet.

Thanks also to the researchers from the Australian Centre for Health Engagement, Evidence and Values (ACHEEV) at the University of Wollongong who are supporting the delivery of the jury as facilitators and moderators: Associate Professor Chris Degeling, Dr Yves Saint James Aquino, Lucy Carolan, Dr Patti Shih, Ms Emma Frost, Ms Belinda Fabrianesi, Ms Saniya Singh, and Ms Kathleen Prokopovich.

This document was prepared by Diana Popic, Lucy Carolan, Dr Yves Saint James Aquino and Professor Stacy Carter from ACHEEV. Professor Stacy Carter is the guarantor and takes final responsibility for the contents.

1. Table of contents

1. The citizens' jury

1.1. WHAT IS A CITIZENS' JURY?

A citizens' jury is a democratic research process used all over the world. It brings together a randomly selected group of people who roughly represent an entire community. The people who attend (citizens or community members) learn about an issue. They discuss the issue with one another, and then make recommendations about what should happen, and whether and how policy should change. These recommendations are carefully recorded. Then, the research team report the group's recommendations to people and organizations that can make decisions about the topic (policymakers). They will also share the recommendations publicly, often through the form of news articles, presentations and academic papers.

In this citizens' jury, you are part of a randomly selected group of people who represent the entire Australian population. Our research team will report the jury's recommendations to people who make decisions about the newborn bloodspot screening program. These people will include the Australian Government Minister responsible for health, and the Commonwealth Department responsible for health policy.

1.2. HOW DO CITIZENS' JURIES WORK?

To help jurors learn about the jury topic, researchers invite experts to share information with the jury. Participants can ask experts questions about the information and discuss it with each other. The researchers will allow time for this discussion.

In this citizens' jury, experts in newborn screening and genomics will provide you with written information and video presentations. Experts will explain different perspectives in their topic areas. You will have two opportunities to meet the experts and ask questions. If you have any more important questions after that, the researchers will do their best to find answers for you.

The jury process will allow you to explore what matters to you, and what matters to the other jurors. We will then ask you to think about all the different perspectives together and consider how best to balance the information. A citizens' jury is about respecting the diversity of perspectives and finding ways we can live together. The information you read and hear may inform your opinions, and your opinion may (or may not) change. We will support you and your fellow participants as you discuss issues and make recommendations together.

There may be times when you and your fellow participants do not all agree on a recommendation. Whether you agree or not, we will ask you for the reasons behind your opinions. Your reasons are always very useful to decision-makers as they consider what to do.

1.3. YOUR ROLE AS A JUROR

This project is an opportunity for Australians to be directly involved in democratic decision-making. Your involvement will tell us whether using genomics in newborn screening is acceptable to Australians. It will tell us if there are any situations where genomics should not be used in newborn screening. It will tell us if Australians expect any rules or supports to be put in place if genomics is used.

Your role as a juror is to represent your fellow Australians. It is also important that your discussion and recommendations are well-informed. So you need to use this booklet, and your conversations with the experts, to get the information you need to understand the issues and make decisions. It is important that you work as a group, each bringing different life experiences and perspectives. Understanding these differences will help your recommendations reflect the different views of the Australian population.

You can find a list of key terms on citizens' juries on the last pages of this booklet.

1.4. ABOUT THE JURY

This jury process is in two parts.

Part 1 happens online, so you can participate from home. You will be required to join three online meetings of about two hours to be held on:

- Sunday 9 March 2025, 3:00-5:05pm Sydney time
- Sunday 16 March 2025, 3:00-5:00pm Sydney time
- Sunday 23 March 2025, 3:00-5:00pm Sydney Time

During Part 1 you will meet the other 29 jurors. Together, you will watch videos, and then be able to ask questions of the experts who recorded them. You will be able to discuss the videos with your fellow jurors. In between, you will be able to access online information provided by the experts, comment on the videos, and read and respond to comments from other jurors. We will ask you to complete a short online survey at the beginning and end of Part 1, so that we can track the views of the jury over time. We will send you links to both surveys. Part 1 will take a total of approximately 8 and 1/2 hours.

Part 2 happens face to face (in person), over three days, from 1pm Friday March 28 to 4pm Sunday March 30, 2025, at the Mercure Hotel in Canberra, Braddon, ACT. Part 2 will take about 19 hours. Travel time to and from Canberra could take between 7 and 29 hours depending on where you live. On the Friday evening you will be invited to meet-and-greet drinks. You will have free time on Saturday evening.

Part 2 will include a final opportunity to meet the experts and ask them questions. Working together as a jury, you will first develop recommendations on issues that policy makers are especially concerned about. You will then work together to decide what other issues are important, and develop recommendations for policymakers around those issues. Facilitators will support all participants through this process. On the Sunday afternoon, participants will have the opportunity to present their recommendations to the research team and organisations interested in the outcomes of the jury. You will be required to complete a short survey at the end of Part 2 so we can track the views of the jury over time. You will also be asked to complete another survey about your experience of being on the jury.

1.5. THE PROCESS: A ROADMAP

START HERE

PART 1: ONLINE

Zoom Session 1

Sunday March 9th
3pm – 5:05pm Sydney time

Opening whole group session
Welcome and introductions, why we are here and why is the jury process important?

Relationship & skill building Meeting one another and skill building exercise.

Evidence videos 1 & 2.
Watch evidence video together, followed by Q&A session with expert.

Monday March 10th - Sunday March 16th

Evidence videos 1 to 5 posted online - Bulletin Board activated.

During this week, watch the videos and engage with your fellow jurors online.

Zoom Session 2

Sunday March 16th
3pm – 5pm Sydney time

Opening whole group session
Welcome back & update

Evidence videos 3, 4 & 5. Watch evidence videos together, followed by small group discussions & whole group Q&A session with experts.

Monday March 17th - Sunday March 23rd

Evidence videos 6 & 7 posted online

During this week, watch the videos & read the information provided – feel free to interact with other participants and ask questions.

Zoom Session 3

Sunday March 23rd
3pm – 5pm Sydney time

Opening whole group session
Welcome back & update

Evidence videos 6 & 7. Watch evidence videos together, followed by small group discussions & whole group Q&A session with experts.

Monday March 24th - Thursday March 27th

Let us know if you have any questions and we will go looking for answers for you, for when we meet in person!

NEXT UP CANBERRA

IN PERSON

PART 2: FACE TO FACE

Carlton Room, Ground Floor, Mercure Canberra

DAY 1: Friday 28th March 2025, 12:30pm – 7pm
12:30 lunch, 1pm start

Welcome Lunch

Available for participants (outside the Carlton Room)

1pm Opening Ceremony

Getting started – Ground Rules – Working together and developing skills

Afternoon tea break

Reviewing hopes, concerns and final questions about genomics in NBS

Expert panel session – Closing conversation

6pm - 7pm Light supper

Drinks and canapes

DAY 2: Saturday 29th March 2025, 8:45am – 5.30pm
8:45am coffee, 9am start

9am Start

Small group deliberation on three important issues

Morning tea break

Plenary discussion of three important issues, and developing recommendations

Lunch break

Whole group discussion of one more important issue. What other issues are important?

Afternoon tea break

Prioritise topics for recommendations

Begin drafting in small groups

Get feedback from the whole group

Closing conversation

5.30pm Close

(Free evening)

DAY 3: Sunday 30th March 2025, 8:45am – 4pm
8:45am coffee, 9am start

9am Start

Drafting recommendations in small groups

Morning tea break

Finalise recommendations together and voting

Lunch

Practice presenting recommendations

Afternoon tea break

Closing ceremony: Jurors present recommendations to policymakers, policymakers respond

Closing conversation

Final survey

We say goodbye and close

4pm Close

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1.6. ONLINE PLATFORMS FOR PART 1

Part 1 will occur on Zoom and VisionsLive.

Zoom is a video conferencing platform that can be used on a computer desktop or a tablet. If you need to borrow a tablet, we can help.

VisionsLive is an online research platform that is used to host research activities. It works a bit like a Facebook page, but it is private and secure, and will only be available to the jurors and researchers. You can watch videos, read information, post comments, and reply to other participants' comments on VisionsLive. To access VisionsLive you will need a computer or tablet. Mobile phones are not suitable for the activities we will do on VisionsLive. If you need to borrow a tablet, we can help. You will receive email messages and links to access the activities on VisionsLive. You will simply need to click on the links to join.

If you haven't used Zoom before or would like **support** to use Zoom or VisionsLive, please contact our research team and we can make a time to talk it through with you. Contact details can be found on page 17 section 1.10.

1.7. VENUE FOR PART 2, IN-PERSON

For Part 2 you will need to travel to Canberra, ACT. Here you will meet with the other participants. You will also meet the researchers, experts, and staff from organisations interested in the jury.

ACCOMMODATION, BREAKFAST AND DINNER

Everyone will stay at the **Mercure Canberra,** located at the corner of Ainslie & Limestone Avenues, **Braddon** ACT 2612, Phone +61 (2) 6243 0024. The hotel is 1km (10 – 15 minute walk) from Canberra city centre.

Hotel check-in is from 2pm. It may not be possible to check-in before the event starts at 1pm. We have scheduled a break in the program on Friday afternoon to allow you to check in and put your luggage in your room. **Hotel check-out** on the Sunday is required by 11am.

All participants will have their own **queen room** with a private bathroom.

A buffet **breakfast** is provided to all hotel guests, with no need for vouchers. Breakfast is available from 7am on the Saturday and Sunday.

Before the start of the event, the research team will give you a VISA gift card with funds to cover **dinner** for Friday and Saturday nights* valued at $80 ($40 per night). If you have any questions about how to use the VISA gift cards just ask someone from the research team. If you are staying more than two nights in Canberra due to availability of flights/ travel restrictions etc, we will provide you with additional dinner vouchers.

VENUE FOR CITIZENS' JURY AND LUNCH

The jury will take place in the **Carlton Room on the ground floor of the Mercure Canberra**. The Carlton Room is large and well-ventilated.

On the Friday, lunch will be available outside the Carlton Room (in the pre-function area) from 12.30pm. There will be an afternoon tea break and light supper and drinks served at the end of Friday. Morning tea, lunch and afternoon tea will be provided for you on the Saturday and Sunday. If you have any dietary requirements, please let the research team know in advance.

The **jury begins at 1pm on Friday 28 March 2025**. The UOW research team will be in the Carlton Room from 11am setting up if you need to speak to one of us.

Hand sanitiser will be available. You can ask a member of the research team for a mask if you would like to wear one.

1.8. TRAVEL COSTS

All participants' travel costs will be covered by the research project. We will be in contact with you prior to the event to discuss the best way for you to travel to and from Canberra. If you are travelling by plane, train, or bus we can book your tickets for you. If you are driving to the venue we will cover your petrol costs.

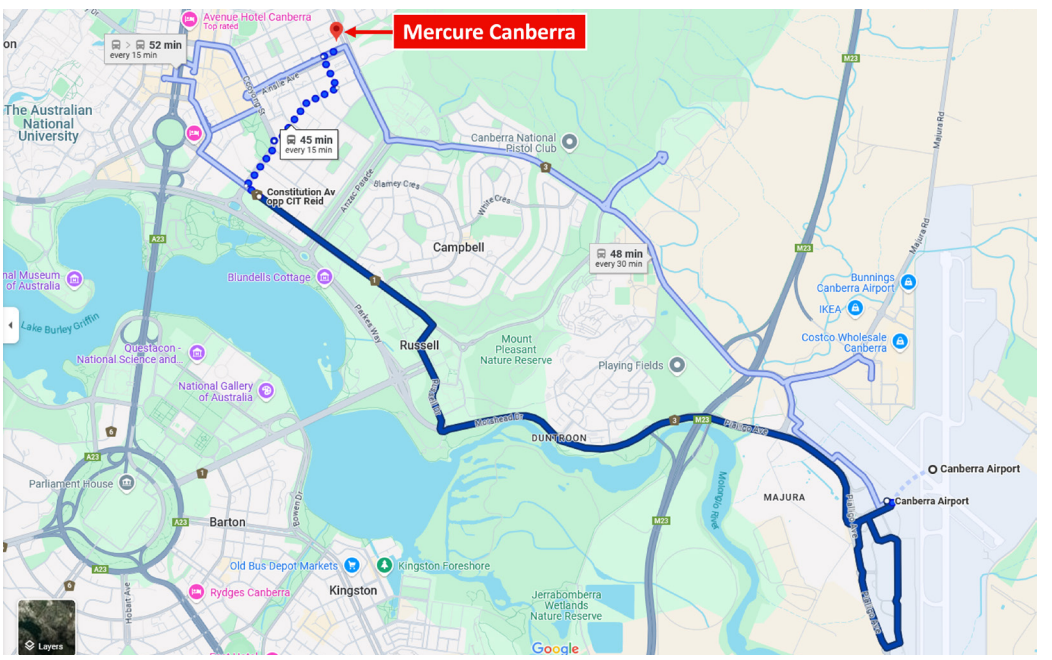
In addition to the $80 dinner card mentioned above, we will provide all

participants with a $100 VISA gift card to cover travel incidentals (taxi fares, parking charges, tolls, petrol etc) incurred between your home and the venue. These cards will be posted to you before you need to travel. If you think your travel incidentals will cost more than $100 in total, please discuss this with the research team before you travel, keep your receipts, and we can organise reimbursement.

1.9. TRANSPORT TO MERCURE HOTEL, CANBERRA

BY PLANE

If you are flying, when you arrive at Canberra Airport, you will need to make your own way to the venue. Taxis and rideshares are available from the airport. A taxi fare from the airport to the hotel is usually less than $30. Rapid 3 Bus can take you from the Canberra Airport to the City interchange (see map below).



Rapid 3 Bus Route from Airport to Mercure Hotel, Braddon

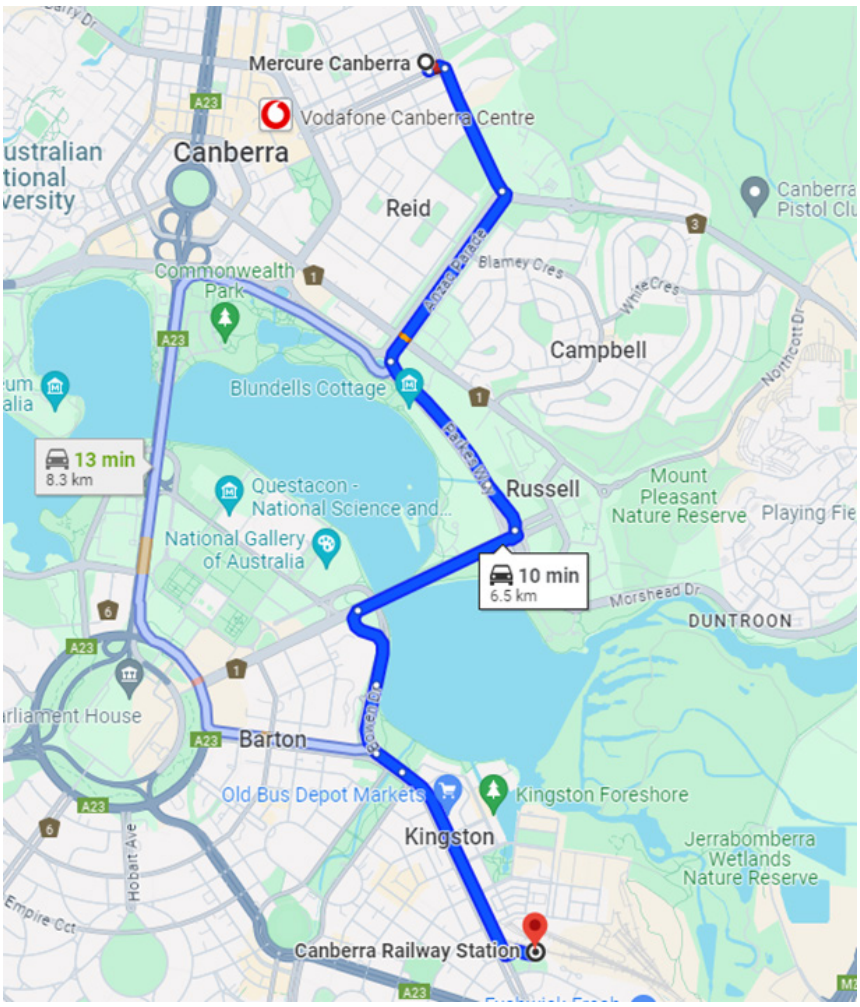
BY BUS/COACH

If you are catching a bus/coach, when you arrive at the City Interchange, you will need to make your own way to the venue. You could take a taxi or rideshare from the City Interchange to the hotel or it is about a 20-minute walk to the Mercure Canberra in Braddon. Buses run from the City Interchange that can drop you closer to the hotel. One route is the ADFA Loop via Campbell & Reid which costs under $5.

BY TRAIN

If you are catching a train, when you arrive at Canberra Railway Station (see map below), you will need to make your own way to the venue. It is a 10-minute journey by taxi or ride share (which should cost approximately $22 - $28), to the Mercure Canberra in Braddon. Alternatively, you could catch a Line 2 bus to the City.

Canberra is 4 hours 10-minutes by train from Sydney Central Station. Canberra is also accessible by train from several regional towns in New South Wales and Victoria.



Taxi/rideshare route from Canberra Railway Station to Mercure Hotel, Braddon.

BY CAR

If you are driving to Canberra, parking at the Mercure Hotel is free, but it is subject to availability at the time of arrival. Entry to the back car park is via Batman Street and the gates are operated by a keypad. (The research team will provide you with a code closer to the event).

1.10. IF YOU NEED SUPPORT OF ANY KIND

If you have any questions or need support during your time on the jury, please talk to the research staff. You can see who the research staff are on page 18**.**

For questions about the **study or jury program,** please contact: Professor Stacy Carter, ph. 02 4221 3243, e. citizensjury-genomicsNBS@uow.edu.au

For questions about **transport, accommodation, and online (Zoom or VisionsLive) support**, please contact: Lucy Carolan, ph. 0488 746 163, e. citizensjury-genomicsNBS@uow.edu.au

It is possible that you might become distressed during the jury process. For example, if someone close to you has a genetic condition, or if talking in larger groups causes you some anxiety. In the first online, and the first face to face sessions, we will introduce the research staff whose role it is to support anyone who feels distressed. If you experience any distress during the jury, and feel comfortable to, please talk to Saniya Singh. She has the skills to assist you and will provide you with immediate assistance.

If you do not want to use this support, but have any concerns about your how you are feeling, or about your mental health, you can seek support from one of the services listed below:

beyondblue	ph: 1300 22 4636	www.beyondblue.org.au
Lifeline	ph: 13 11 14	www.lifeline.org.au
13YARN	ph: 13 92 76	www.13yarn.org.au

1.11. IF YOU DEVELOP COLD OR FLU SYMPTOMS

Please note that you must not travel to the event if you are unwell with cold or flu like symptoms, or have an active viral infection. If you are unsure, please call the research team to discuss.

2. The research team, expert witnesses and supporting organisations

2.1. WHO IS RUNNING THE EVENT?

The Australian Centre for Health Engagement, Evidence and Values (ACHEEV) specialises in connecting health decision-makers to the Australian public through research processes like citizens' juries. Our mission is to make health systems more inclusive and democratic. We identify real-world problems faced by health systems. We then support Australians to learn about these problems so they can provide advice to decision-makers.

WHO WE ARE: THE RESEARCH TEAM

PROFESSOR STACY CARTER

Role during the jury: Lead facilitator

Stacy is the Director at ACHEEV. Stacy's training is in public health, research methods, and ethics. She has done a lot of research about screening and testing in health. Stacy has led many citizens' juries across Australia.

ASSOCIATE PROFESSOR CHRIS DEGELING

Role during jury: Co-facilitator

Chris is an Associate Professor at ACHEEV. His research explores public health ethics and public health policymaking on issues where humans, animals and ecosystems meet. Chris has led many citizens' juries across Australia.

LUCY CAROLAN

Role during jury: Participant Support Officer (Logistics)

Lucy is a research assistant at ACHEEV working on a range of health-related projects including AI in healthcare and the sharing of general practice data for research. Lucy has supported participants in numerous citizens' juries.

DR SANIYA SINGH

Role during jury: Participant Support Officer

Saniya is a qualified psychologist with mental health first-aid training. She has supported diverse people across numerous settings, including in hospitals and community treatment centres. She recently completed her PhD at ACHEEV on the impact of emotions on medical decision-making.

DR YVES SAINT JAMES AQUNO

Role during jury: Participant Support Officer

Yves is a clinician and philosopher working as a research fellow at ACHEEV. His research expertise includes philosophy of medicine, medical ethics, ethics of cosmetic surgery and ethics of artificial intelligence in healthcare. He was a team member on the ACHEEV citizens' jury on using AI in healthcare.

EMMA FROST

Role during jury: Small group facilitator

Emma is doing her PhD at ACHEEV. Emma's research focuses on Australians' views on the use of Artificial Intelligence in healthcare, and was a team member on the ACHEEV citizens' jury on using AI in healthcare.

BELINDA FABRIANESI

Role during jury: Small group facilitator

Belinda is a senior research assistant at ACHEEV. Her current research focus is the social and ethical considerations in sharing and linking large datasets for secondary purposes. Belinda has supported numerous citizens' juries as a small group facilitator.

DR PATTI SHIH

Role during jury: Small group facilitator

Patti is a Research Fellow and lecturer at ACHEEV. She is a sociologist specialising in social and cultural aspects of healthcare. Her current research is on public engagement in healthcare. She has years of experience in qualitative and deliberative research in public health services.

KATHLEEN PROKOPOVICH

Role during jury: Small group facilitator

Kathleen is doing her PhD at ACHEEV. Her PhD research explores engaging Culturally and Linguistically Diverse (CALD) communities in school-based vaccination for Human papillomavirus (HPV). She is interested in participatory research methods.

2.2. THE EXPERT WITNESSES

In the online process, you will hear important background information from a range of experts. This will include experts with different training and experience. They may know about genetic medicine, rare diseases, paediatrics, newborn screening, health policy, bioethics, or law. We encourage you to ask the experts any questions you have about what they have said. You can ask them during Zoom meetings, or during the expert panel on 28 March. Eight experts will be available to answer your questions.

WHO ARE THE EXPERTS FOR THIS CITIZENS' JURY?

BACKGROUND AND TECHNICAL INFROMATION

CONJOINT ASSOCIATE PROFESSOR KAUSTUV BHATTACHARYA

Conjoint Associate Professor Bhattacharya (University of NSW) is a specialist at Sydney Children's Hospitals Network. He is currently president of Australasian Society of Inborn Errors of Metabolism. He is passionate about novel treatments for rare conditions. He has published novel approaches to treat several metabolic diseases, some of which can be fatal in a baby's first year of life. Kaustuv participates in scientific advisory panels for national and international charities including Rare Voices Australia. He has been on Human Genetics Society of Australasia NBS advisory committee for over 10 years.

Conjoint Associate Professor Bhattacharya will present background information to you. You will meet him online on 9 March 2025, at 4:00pm. Conjoint Associate Professor Bhattacharya will explain:

- What population screening is
- What the Newborn Bloodspot Screening (NBS) program is, what health conditions it tests for, and why

- Why the program is delivered differently in each state and territory, and how this impacts Australians living in each state and territory
- How successful the program has been so far
- Why governments in high-income countries are considering using genomics is newborn screening programs now

Conjoint Associate Professor Bhattacharya will talk about issues covered on page 34 in this booklet.

PROFESSOR BRUCE BENNETTS

Professor Bennetts is a clinical scientist at the Children's Hospital at Westmead. He has over 30 years of experience in diagnosing rare genetic disorders. His department offers a range of molecular testing for many genetic disorders. He is currently leading a Medical Research Future Fund grant exploring the role of genomic sequencing in newborn screening. His research team includes clinicians, pathologists, molecular geneticists, newborn screeners, genetic counsellors and midwives.

Professor Bennetts will present technical information to you. You will meet him online on 9 March 2025, at 4:00pm. Professor Bennetts will explain:

- Different childhood-onset health conditions, including rare, genetic and metabolic conditions.
- What genetics and what genomics is, and the difference between them in the context of screening newborn babies.
- What biochemical tests are and how the current NBS program uses mostly biochemical tests, with genetic testing being used for a small number of health conditions.
- How genetics and genomics could be used in the NBS program.

Professor Bennetts will talk about issues covered on page 37 in this booklet.

POLICY INFORMATION AND FIRST-HAND EXPERIENCE OF RARE GENETIC HEALTH CONDITIONS

DR KRISTEN NOWAK

Dr Nowak is Director, Population Health Genomics, Western Australian (WA) Department of Health. Her policy team aids translation of evidence-based genomic and screening technologies and knowledge into health systems. Nationally, Kristen is a member, Health Technology and Genomics Collaboration; and Scientific and Medical Advisory Committee, Rare Voices Australia. Previously she was Chair of the Newborn Bloodspot Screening Program Management Committee, and member of the Standing Committee on Screening.

Dr Nowak will present the policy context to you. You will meet her online on 16 March 2025, at 3:00pm. Dr Nowak will talk about the different ways genomics could be used in the Australian NBS program.

She will explain what the use of genomics in the NBS would mean for all newborn babies. But also, what resources would be needed to deliver genomic tests. Finally, she will ask you to consider if and how Australia's screening principles would be maintained should each test be introduced.

Dr Nowak will talk about issues covered on page 41 in this booklet.

LOUISE HEALY, RARE VOICES AUSTRALIA

Louise Healy is Education and Advocacy Manager at Rare Voices Australia (RVA). RVA is the peak body for Australians living with a rare disease and led the collaborative development of the National Strategic Action Plan for Rare Diseases. RVA's focus is policy and system advocacy, including newborn bloodspot screening expansion. Louise has more than 10 years of rare disease advocacy experience.

In the video presentation for evidence package 2, three parents will share their lived and living experience of having a child with a rare health condition, and their experience of screening and/or testing for rare health conditions.

Ms Healy will answer any questions you may have about child and parent experiences of rare health conditions and screening through the NBS programs. You will meet her online on 16 March 2025, at 3:00pm.

BENEFITS – WHY MIGHT WE WANT TO DO THIS?

PROFESSOR ZORNITZA STARK

Professor Stark is a clinical geneticist at the Victorian Clinical Genetics Services (VCGS) and Clinical Lead at Australian Genomics. She completed her medical studies at the University of Oxford. She trained in paediatrics at the Royal Children's Hospital in Melbourne and in clinical genetics at VCGS. Zornitza has lead numerous translational genomics projects through Melbourne Genomics, Australian Genomics, and the Genomics Health Futures Mission. All projects aim to accelerate rare disease diagnosis.

Professor Stark will present evidence package 3. You will meet her online on 16 March 2025, at 4:00pm. Professor Stark will explain some reasons why we may want to use genomics in newborns screening. She will explain the:

- Potential benefits to the child, including earlier diagnosis and access to care.
- Potential benefits to the child's family, including less psychological distress and information for future family planning.
- Potential benefits to the health system, including more research and better reference data.

Professor Stark will talk about issues covered on page 45 in this booklet.

RISKS, CHALLENGES OR UNINTENDED CONSEQUENCES – WHY MIGHT WE BE CONCERNED ABOUT DOING THIS?

PROFESSOR KRISTI JONES

Professor Jones is a clinical geneticist and leads the Department of Clinical Genetics at the Children's Hospital, Westmead. Clinical interests are broad, but include neurogenetics and preimplantation genetic testing. She is Clinical Professor at the University of Sydney and is an active clinical researcher. She has led clinical trials of advanced therapeutics in Duchenne muscular dystrophy since 2008, including recent gene therapy trial in 2-4 year old boys.

Professor Jones will present evidence package 4. You will meet her online on 23 March 2025, at 3:00pm. Professor Jones will explain some reasons we may be concerned about using genomics in newborn screening. She will explain how if genomics is used in the NBS program babies and their families could experience the NBS in many different ways, depending on their health condition. She will explain the reasons why families are unlikely to have the same experience of the one program.

Professor Jones will also describe the potential for inaccurate genomic screening tests depending on the health condition. She will outline some of ways an inaccurate test result could impact on families. Lastly, Professor Jones will explain that using genomics in NBS may mean we tell families that their child will develop a health condition well into the future. She will outline how this may impact on families' wellbeing.

Professor Jones will talk about issues covered on page 48 in this booklet.

WAYS OF THINKING (TOOLS TO HELP YOU THINK THROUGH THE INFORMATION)

PROFESSOR MARGARET OTLOWSKI

Professor Otlowski is Professor of Law at the University of Tasmania and Director of the Centre for Law and Genetics. Her research expertise is in health law focusing on law and genetics/genomics including issues of regulation, privacy, consent, discrimination and data sharing.

She is a Fellow of the Australian Academy of Law and Patron for Tasmanian Women Lawyers. She is also a Commissioner on the Tasmanian Voluntary Assisted Dying Commission.

PROFESSOR AINSLEY NEWSON

Professor Newson is Professor of Bioethics at the University of Sydney. She has 25 years of research experience in the ethical aspects of genetics, including a longstanding interest in newborn screening.

Ainsley is interested in how genomics may be introduced at this life stage while also maintaining trust and effectiveness of the current screening program. A decade ago, Ainsley helped write Australia's current newborn screening policy framework. She has two children.

Professor Otlowski (your legal expert) and Professor Newson (your ethics expert) will together present different ways of thinking about the topic. You will meet them online on 23 March 2025, at 4:00pm.

They will talk about how using genomics in the NBS program may:

- Require changing the rules that direct population screening
- Reveal extra information not currently part of the program and why it's important to think about this now
- Make 'patients-in-waiting' that could create uncertainty for families and risks to health services
- Impact on public trust in the program and why this matters
- Make consent more complicated
- Impact on public attitudes on diversity and disability

Professor Otlowski and Professor Newson will talk about issues covered on page 55 in this booklet.

2.3. FUNDING

MEDICAL RESEARCH FUTURE FUND (MRFF)

This citizens' jury is part of the gEnomics4newborns research project, which is funded by the MRFF's Genomics Health Futures Mission - 2021 Genomics Health Futures Mission Grant Opportunity. MRFF is an ongoing research fund set up by the Australian Government in 2015 to support Australian health and medical research.

2.4. SUPPORTING ORGANISATIONS

LEEDER CENTRE FOR HEALTH POLICY, ECONOMICS AND DATA

The gEnomics4newborns research project is conducted by the Leeder Centre for Health Policy, Economics and Data located at the Faculty of Medicine and Health, University of Sydney. The Leeder Centre conducts health policy research, education, analysis and advice to improve health outcomes through the practical implementation of policy innovations.

ADVISORY GROUP

The citizen's jury was developed in consultation with an advisory group consisting of representatives from government departments, the

academic sector, profesisonal organisations, industry, and consumer representatives. The members are:

- Professor Kees van Gool, professor of health policy and systems at the Menzies Centre for Health Policy and Economics, University of Sydney.
- Associate Professor Azure Hermes is from the Gimuy Walubara Yidinji people, traditional custodians of the Cairns area, and works at the National Centre for Indigenous Genomics, Australian National University.
- Belinda Burns, Department of Health, Government of Western Australia
- Lauren Hunt, Chief Operating Officer, Human Genetics Society of Australasia
- Dr Erin Evans, Chief Executive Officer, InGeNA: Industry Genomics Network Alliance
- Julie Cini, community leader and patient advocate
- Professor Jackie Leach Scully, Professor of Bioethics and Director of the Disability Innovation Institute, University of New South Wales
- Professor Julie McGaughran, Director of Genetic Health Queensland
- Klair Bayley, patient advocate and a qualified nurse and midwife working at the Office of Population Health Genomics, Department of Health, Government of Western Australia
- Kym Mina, Genetic Pathologist and Fellow with the Royal College of Pathologists of Australia
- Dr Natasha Heather, Chair of the Human Genetics Society of Australasia's Newborn Screening Committee

3. Critical thinking and cognitive bias

3.1. WHAT IS CRITICAL THINKING?

Most of the time, we think quickly, emotionally and intuitively to make decisions. For example, if we feel cold, we put on a jumper and don't think about whether putting on a jumper was the right thing to do. Less often, we think slowly and carefully about the best course of action. For example, when we decide where to live, when we write a will, or when we consider committing to a volunteer role in a community organisation. For these bigger decisions which have significant consequences, we look for information, ask questions of people who know more than we do, consider many viewpoints, and weigh-up pros and cons. We think slowly and carefully. Critical thinking is a lot like this.

Critical thinking is when we bring together data, facts, observations, life experiences and arguments, and then think carefully and slowly about them. But it also involves thinking about a problem with the needs and rights of other people in mind, and asking what would be best for everyone. When we think critically, we improve our ability to find quality solutions to complex problems.

So how do we think critically?

1. We learn to do it, and do it ourselves.
2. We think beyond our own and our local community's interests. We consider the interests of people we know little about.
3. We ask questions, are open-minded, reflect on our own assumptions, and test our own conclusions.
4. We draw on different ways of thinking to broaden our views.

To help you think critically during your time on the jury, there are some key thinking skills and questions we recommend you use on the next page. During our online session on 23 March 2025, two expert witnesses

will discuss different ways of thinking about all the information you've heard. You can also read about these different ways of thinking on page 55 section 5.9 of this booklet.

3.2. CRITICAL THINKING TOOLS AND QUESTIONS

Critical thinking for	Why is this important?	Example questions to ask
CLARITY 	When a statement is clear, we can tell if it's both accurate and relevant.	**Can you elaborate? Can you give me an example?**
ACCURACY 	When a statement is considered correct by an agreed standard. A statement can be clear but inaccurate.	**How can we verify or test that? Is there alternative evidence that may contradict the claim?**
RELEVANCE 	Relevance is clear connection between the statement and the problem. A statement may be clear and accurate, but irrelevant to the issue.	**How is that related to this issue? What's the link between the information and the problem?**
DEPTH 	Statements have depth when they deal with the complexities of the issue.	**Are they considering the complexity of the issue? What details of the problem are we missing?**
BREADTH 	Breadth is when an argument considers many/all viewpoints and perspectives.	**What other points of view might be missing? Do we need to look at this in other ways?**
LOGIC 	Thinking is logical when a combination of thoughts make sense together and support one another.	**How is it possible to be both X and Y? Is there a contradiction?**
SIGNIFICANCE 	When there's lots of information or views to consider, we need to focus our thoughts to make our way to a solution.	**What's the most important problem to consider? What facts are most important?**
FAIRNESS 	When we consider all arguments, and don't show favouritism to one side/idea.	**Do I have any personal interests in this issue? Are they sympathetically representing the viewpoints of others?**

Source: Mosaic Lab and Miniature Guide to Critical Thinking

3.3. WHAT IS COGNITIVE BIAS?

Cognitive bias is fast thinking that stops us from seeing information accurately and limits our understanding of complex topics. Cognitive biases make it more difficult for us to draw on the evidence or really hear other people's perspectives. We all have cognitive bias. It is human nature. It is also something we can become aware of and try to prevent in our own thinking. To help you understand cognitive bias further, we will be talking about it in our **introductory online session on 9 March 2025**. Below are descriptions of **six types of cognitive bias**, and guidance on how to avoid them during your time on the jury.

Six types of cognitive biases

Anchoring bias

Being **overly reliant** on the first and last pieces of information you hear or see. Example: The first thing you hear at the start of the presentation and the last person you hear in a day will create a lasting memory – Be mindful of these.

! **Review all the information you have received.**

Bandwagon effect

The probability of one person adopting a belief increases based on the number of people who hold that belief. This is a powerful form **groupthink** and is the reason why meetings are often unproductive.

! **Be clear on your own position. Be wary of getting swept along.**

Blind-spot bias

Failing to recognise your own cognitive biases is a bias in itself. People notice biases much more in others than in themselves.

! **Encourage others to watch for your blind spots and you can watch for theirs.**

Confirmation bias

We tend to listen only to information that confirms our **preconceptions**. This is also true for believing things more from those people we most closely relate to. This can therefore lead to **stereotyping**.

! **Take time to consider all the evidence and different positions.**

Information bias

The tendency to **seek information even when it will not affect action.** More information is not always better. With less information, people can often make more accurate predictions.

! **Check that more information is essential. Don't delay a decision for too long to find more information.**

Authority or Anti-Authority bias

A tendency to **give greater weight or importance to the opinion of an authority figure or organisation** and be more influenced by them. The flip side is a 'blanket' opposition to authority, disregarding their knowledge.

! **Carefully consider the evidence. Remain open to all kinds of relevant expertise.**

Source: Mosaic Lab

4. Our question for you (the remit)

4.1. BACKGROUND

The purpose of the newborn bloodspot screening program is to find babies who have specific rare, serious and treatable health conditions, so that care can be provided early to greatly improve a baby's health. The newborn bloodspot screening program works very well. It is one of the most trusted population screening services in Australia. Australia tests 99% of babies through the program.

Since it began, the newborn bloodspot screening program has relied on tests that look at the natural chemicals in in a baby's blood. Recently, in some states and territories, a small number of genetic tests have also been used. There are now proposals to start using genomics in the newborn bloodspot screening program.

There are always debates about whether to add new tests to the screening program. This jury is not about whether or not we should test for more conditions. It is about whether or not we should use genomics in newborn bloodspot screening. Genomics technologies may help to find more health conditions in babies, but it will also introduce complex challenges that are important to think through before Australia makes any final decisions.

4.2. OUR QUESTION FOR YOU:

Under what circumstances, if any, should Australia use genomics in the newborn bloodspot screening program, to ensure the program remains trustworthy and effective?

4.3. SCOPE OF THE JURY

This is about screening the **whole population** of Australian newborn babies soon after birth.	This is **not** about using tests to work out what is wrong with a baby who **has symptoms**.
The NBS already screens **>99%** of Australian babies with their parents' **consent**.	Most parents consent to NBS **in the first days after** the birth.
This is **not** about ensuring the NBS tests for the same conditions in all Australian **states and territories** (work to achieve that is already underway).	This is not about whether we should **expand** the NBS to cover more conditions **in general** (that is already being considered).

Genetic or genomic testing ranges from:

single gene tests ⟷ **gene panels (sequencing DNA regions or chromosomes)** ⟷ **sequencing whole genomes**

(less data) - (more data)

There are lots of questions:

What types of genomic testing should be done in the NBS?

How much of the genetic information from testing should be **extracted**, and how much should be **reported**?

How much of the genetic information should be **stored**?

The answer to these questions may or may not be the same

Notice that the **technology** used, the **genes (or genome) tested**, and the **health conditions tested** for are different, but are connected.

Scientists are still working to understand the relationship between our genes and different health conditions.

This means a lot of the information from genomics right now is uncertain. But this could change.

We are asked to make recommendations about **using genomics** in NBS.

We are **not** being asked to provide the names of health conditions that should or should not be tested for.

There is **no clear agreement** on how much of the DNA we should sequence, what genetic information should be reported, or what to do with information that is uncertain now, but could be helpful in future.

This means your considered recommendations about how to use genomics in NBS are **very important** to inform policymaking.

5. Information on the topic

5.1. WHAT IS THE NEWBORN BLOODSPOT SCREENING PROGRAM?

The Australian **newborn bloodspot screening program (NBS)** was set up in the 1960s to identify babies who would otherwise develop rare but serious health conditions that can be treated. The NBS program aims to find these babies early, before they have symptoms. The program relies largely on checking a blood sample for different biochemicals (naturally occurring chemicals in the body) that show that a condition is present. This is referred to as a *biochemical test*. Babies found to have, or be at risk of developing, a health condition after screening can get medical care. Early care can reduce the effects of the condition (some of which are life-threatening) and improve the baby's health.

Image by Luma Pimentel | Unsplash

There is a difference between screening in a program like the NBS, and diagnostic testing done in a clinic. *Screening* means checking people who seem fine, to find out if they might have a health condition. *Diagnosis*, on the other hand, usually refers to identifying a condition by examining symptoms and conducting tests.

The first health condition screened for in Australia was phenylketonuria (PKU), a disorder that can affect the brain. This condition has no obvious symptoms at birth. Each month without treatment, a baby loses about

four IQ points. Babies with phenylketonuria can have a special diet, supplements, and sometimes medicines. There is good evidence that if babies with phenylketonuria are found early and treated, they can avoid intellectual disability and health problems.

The NBS program has grown since it started. Australia currently screens babies for 32 **health conditions** that are likely to cause serious harms to *babies*. They are also all conditions where immediate treatment can help, even if the condition can't be cured. Overall, the NBS program is very beneficial for babies who have these conditions.

More information: https://www.health.gov.au/our-work/newborn-bloodspot-screening

5.2. PRINCIPLES FOR NEWBORN BLOODSPOT SCREENING IN AUSTRALIA

Australia's approach to screening is based on principles that have been accepted around the world. The key principle is that the potential benefits of screening (earlier identification and treatment of a health condition) outweigh the potential harms of screening (such as the possibility of 'false alarms' where a person might go on to receive tests or treatments they don't need).

You can see the full text of the criteria here: https://www.health.gov.au/resources/publications/population-based-screening-framework

The NBS National Policy Framework describes extra things that need to be considered when undertaking screening in newborns (compared with, for example, cancer screening for adults).

As we learn more about a condition we might think about adding it to the NBS program. The framework includes criteria that can be used to decide whether a health condition should be added (or removed) from the NBS program (see Table 1 on the next page for a summary of the criteria).

Table 1: NBS Policy Framework Decision-Making Criteria

(The text was adapted from the original criteria, which can be found at the link below this table.)

NBS Policy Framework Decision-Making Criteria	
The condition	1. The condition should be a serious health problem that leads to significant illness or early death.
	2. There should be a benefit from screening the newborn baby (rather than waiting until they are older).
	3. Scientists and doctors should understand how the condition develops, including how and when symptoms might appear.
The screening test	4. There should be an agreed way to test for the condition.
	5. This way of testing should be socially and ethically acceptable to health professionals and the public.
The intervention	6. Health services must be ready to provide diagnostic tests (to confirm if the condition is present), and management if needed, to babies who receive a positive screening test.
	7. There should be an agreed way to treat or manage babies diagnosed with the condition.
Additional considerations	8. The benefit of testing for extra conditions in the program must be weighed against the impact on the program as a whole.
	9. For any decision, there may also be other relevant information that should be considered.

For more information about the NBS Policy Framework: https://www.health.gov.au/resources/publications/newborn-bloodspot-screening-national-policy-framework?language=en

In recent years a new process has been set up to decide if conditions should be added or removed from the NBS program. This process involves the experts who oversee and deliver the NBS program, as well as the federal Department of Health and Aged Care and all the State and Territory governments.

The process includes a step where the **Medical Services Advisory Committee (MSAC)** looks at all the available evidence and estimates how much it would cost the Australian health system to add the condition to the NBS program. The assessments by MSAC are conducted according to the Committee's technical guidelines and also consider the criteria in the NBS National Policy Framework.

For more information about MSAC: https://www.msac.gov.au/

For more information about the NBS decision-making process: https://www.health.gov.au/resources/publications/newborn-bloodspot-screening-nbs-our-national-decision-making-pathway-fact-sheet?language=en

5.3. A POTENTIAL CHANGE IN THE KINDS OF TESTS USED IN THE NBS

Since it began, NBS has relied on *biochemical tests:* tests that measure the levels of specific natural chemicals in a baby's blood. Recently, specific genetic tests have started being included in the NBS program to detect a few extra health conditions.

In this jury, we are considering a big potential change in NBS: the use of *genomic testing*. The difference between genetic and genomic testing is explained in the following sections.

5.4. GENES, GENETICS AND GENOMICS

The science of genetics is complicated! Every ***cell*** in the human body has a *nucleus* that contains all of the information the cell needs to function and to make our bodies work. This information is stored as

DNA (deoxyribonucleic acid), which is made up of biochemical building blocks called *nucleotides* (which are labelled A, C, G or T).

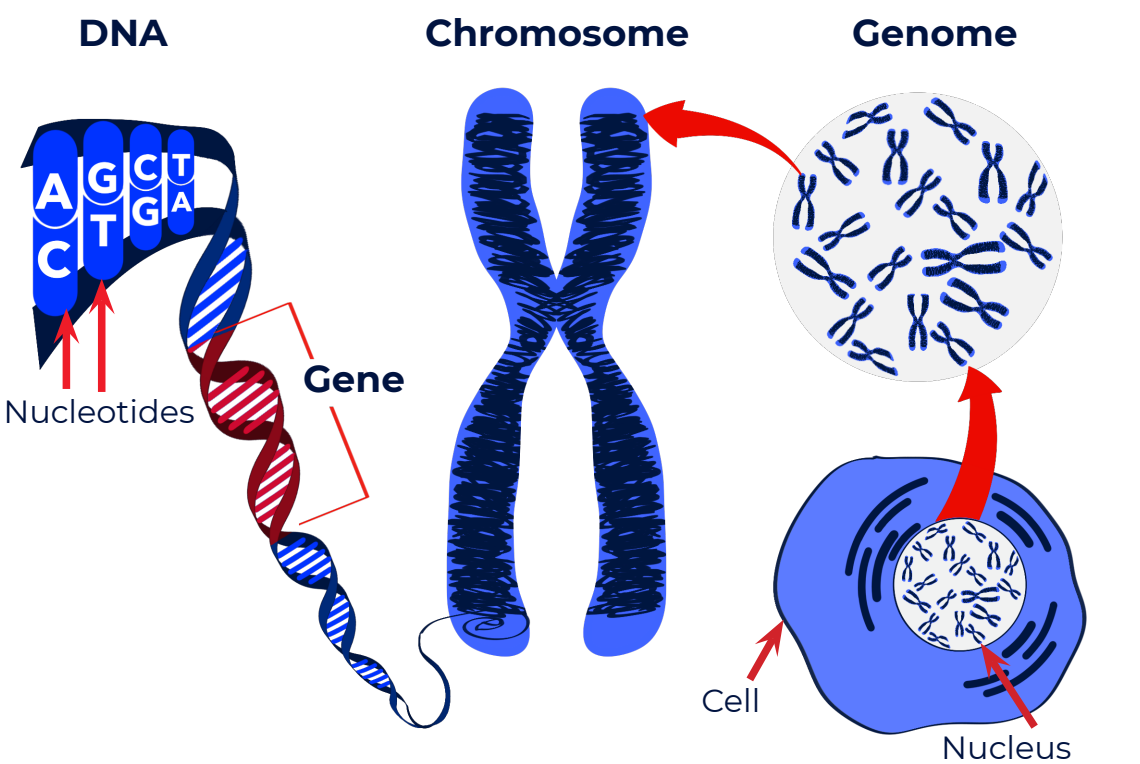
A ***gene*** is a string of DNA that codes for something, such as a protein that is used to form muscle. Humans have approximately 20,000 genes.

Genes and other parts of the DNA are arranged into ***chromosomes***. Humans usually have 46 chromosomes altogether. We inherit one set of 23 chromosomes from our biological mother, and the other set of 23 chromosomes from our biological father.

The ***genome*** is a person's entire genetic code - all of their DNA and chromosomes.

The figure below shows the relationship between DNA, genes, chromosomes, and the genome.

Figure 1: How DNA (strings), genes and chromosomes are organised into the genome inside the cell



Think of a *person's genome as a large cookbook*, and a chromosome as a chapter in the cookbook (see Figure 2 on the next page). A gene in this analogy is like a recipe that provides our cells instructions on how to make proteins to build more parts of the body and make the body function properly.

Sometimes, there are mistakes or "typos" in the recipe that lead to what we call genetic variants. Some genetic variants have typos that do not affect the body, but others have a bigger impact on how the body works. There are different types of typos. For example, imagine an instruction that is supposed to read:

'Dice two ripe tomatoes.'

Typos in the gene could result in the instruction being read as:

'Dice two ripe toes.'

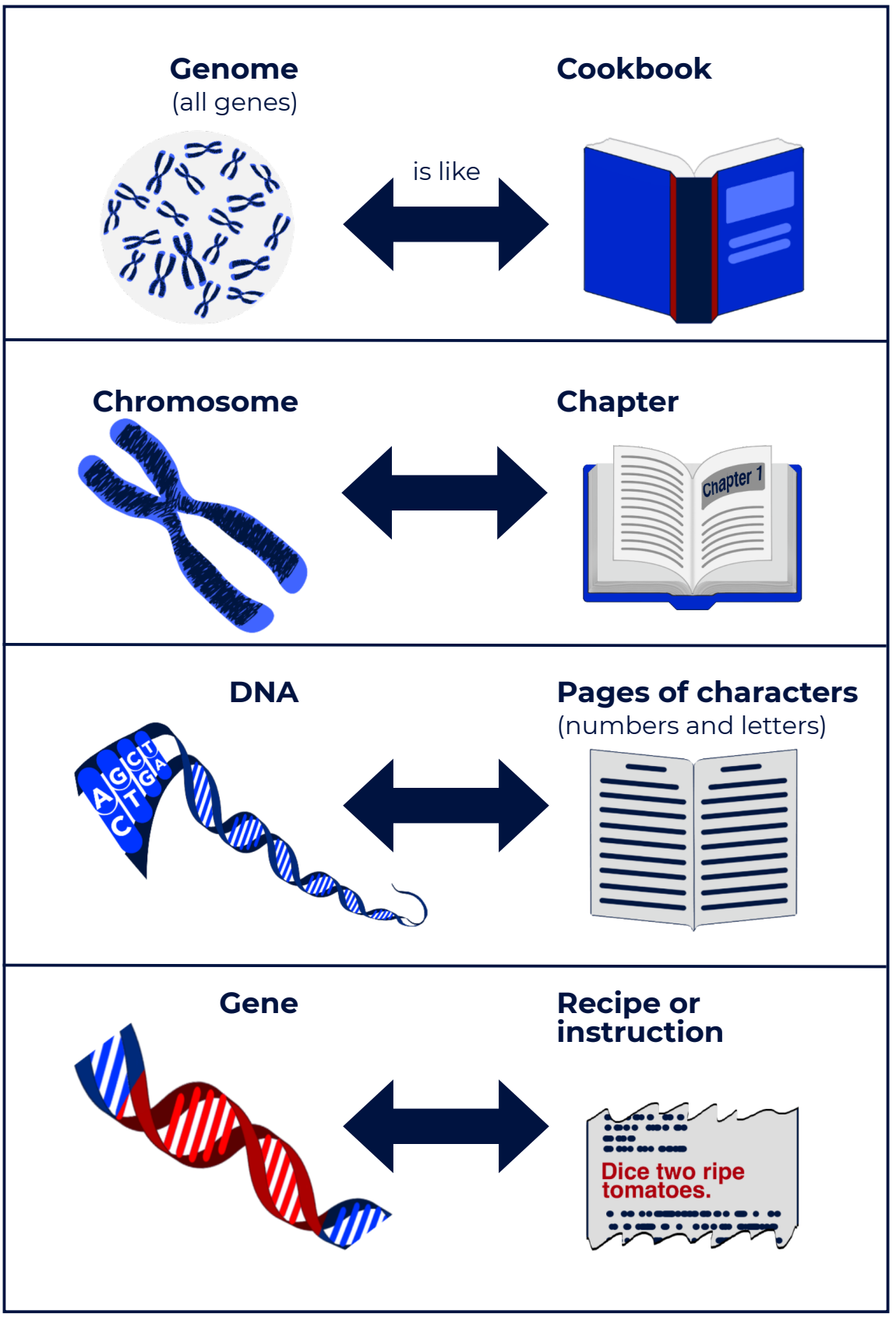
or

'Dice two pipe tomatoes.'

Sometimes the cell can cope with the typo. Other times the typo can stop a gene from working. This can mean a protein we need is not made, or not made properly. This can have serious health consequences. Diseases arising from DNA changes are referred to as *genetic disorders or genetic conditions*.

Some examples of genetic disorders are cystic fibrosis (where gene typos mean the mucus in a person's lungs becomes thick and sticky and makes it hard for them breathe), and spinal muscular atrophy (where gene typos means the nerves that control muscle movement do not work properly, resulting in muscle weakness and sometimes death).

Figure 2: The genome is like a cookbook for the body



5.5. WHAT ARE GENETIC AND GENOMIC TESTS?

Broadly speaking, when we run a *genetic test* we are studying a specific gene or group of genes, and when we run a genomic test we are studying all of a person's genes. Using the cookbook analogy, genetic tests look at a specific recipe or recipes and whether there are mistakes or typos in them, while genomic tests look at the whole cookbook.

Sequencing is the laboratory method that is used to work out the order of the nucleotides in a piece of DNA. Sequencing can be used on a single gene, multiple genes, or the whole genome.

There is a big difference between sequencing a handful of specific genes versus sequencing the whole genome. If you ask scientists where 'genetics' turns into 'genomics', they will give different answers, but generally, it is a difference of scale - genetic tests look at a small number of genes, while genomic tests look at most or all genes.

While scientists have discovered a lot about how genes work and how some gene typos cause a particular disorder, there is still a lot that we do not understand about how all of our DNA works.

There are thousands of conditions where we don't understand if or how genetics is involved. You will learn more about this *uncertainty* later on.

More information: https://www.genome.gov/about-genomics/fact-sheets/A-Brief-Guide-to-Genomics

5.6. TESTS THAT ARE CURRENTLY USED IN THE AUSTRALIAN NEWBORN BLOODSPOT SCREENING PROGRAM

Some genetic disorders lead to changes in biochemicals that show up in the blood and can be diagnosed using biochemical tests. See Table 2 for some examples of these conditions and how they impact children if left untreated.

Table 2: Examples of conditions tested in the NBS

Conditions	Type of screening test(s) used	Untreated outcome	Treatment plan
Maple syrup urine disease (MSUD)	Biochemical test	Children can die or have permanent brain damage within 2 weeks of birth	-Low protein diet -Protein equivalent supplement - Urgent treatment plan for hospital - some may have a liver transplant
Phenylketonuria (PKU)	Biochemical test	Seizures and severe developmental delay (Unable to self-care as adults)	-Low protein diet -Protein equivalent supplement.
Medium chain acyl Co A dehydrogenase deficiency (MCADD)	Biochemical test, followed by a genetic test	25% have sudden death in childhood; Others can get permanent brain damage with illness	Urgent treatment plan to provide emergency care in hospital using carbohydrates during times of acute illness from MCADD.
Cystic fibrosis (CF)	Biochemical test, followed by a genetic test	Affects pancreas and lungs; can lead to developmental delay and nutritional deficiencies	Treatment plan is directed at maintaining nutrition and ensuring organs, such as pancreas and lungs, are functioning.
Spinal Muscular Atrophy (SMA)	Genetic test	Affects the nerves in the spinal cord that control the muscles for head control, arm and leg movement, and breathing, coughing and swallowing	Specific medicines and a gene therapy are available for some types of SMA; Physiotherapy, occupational therapy, speech and language therapy.
Severe Combined Immunodeficiency (SCID)	Genetic test	Children have no T cells, a type of white blood cell that is needed to fight infections; at risk of severe infections in the lungs	Need to be kept away from settings where they might be infected; most children need a bone marrow transplant in their first year of life to stay alive.

The biochemicals in the blood are called biomarkers. Biochemical tests are used for most of the conditions currently screened for in the Australian NBS program. These tests are relatively easy to do and can be done quickly, which is important because treatments typically need to start as soon as possible in babies who are found to have one of the conditions. These biochemical tests are also low cost, which is important because around 300,000 babies a year need to be tested.

Sometimes the biochemical test picks up babies who look like they have a condition, but they do not. To reduce the chance of these 'false alarms', sometimes a biochemical test is followed up by a genetic test, but only in babies who have a positive result after the biochemical test.

However, many genetic disorders do not have biomarkers in the blood. This means the only way to know whether the baby has that disorder is by looking at the baby's DNA.

We already use a limited amount of genetic testing in the NBS program to find babies with SMA, the neuromuscular disorder described earlier, or Severe Combined Immunodeficiency (SCID; a condition where the gene typos stop the immune system working properly which means the baby has trouble fighting infections (see the two bottom rows of Table 2). Scientists know which genes to sequence to look for the most common typos that cause these disorders, so they can be pretty certain that if they find one of these typos the baby will have SMA or SCID.

5.7. HOW GENOMICS COULD BE USED IN THE AUSTRALIAN NBS

Genomic testing is not currently used in newborn screening in Australia or anywhere else in the world. Sequencing DNA takes more time and costs more money than biochemical tests.

But advances in genomics and reductions in the time and cost of sequencing mean it may now be possible to use genomics in the NBS program.

Image by Warren Umoh | Unsplash

Using genomics in the NBS program could identify up to 1,000 more health conditions than the existing program, which could mean earlier diagnosis and treatment for more babies and their families. It could also prompt relatives to be tested to see if they are also at risk or whether they might pass the condition on to future children.

Although research projects around the world and in Australia are exploring different approaches to using genomics to screen newborns, we do not know exactly how genomics might be used in the Australian NBS program. One approach might be to use genomics in addition to biochemical testing (similar to how we are currently using genetic testing in addition to biochemical testing).

However, in comparison to using genetic testing in the NBS program, using genomics raises significant economic, ethical, legal and equity issues. This is because sequencing all of a baby's DNA reveals much more about the baby and their relatives than biochemical and genetic testing. These concerns, and how to weigh them against the potential benefits of genomic sequencing, are what we will be focusing on in our discussions.

5.8. BALANCING BENEFITS AND RISKS

Making a decision about using genomics in NBS programs involves balancing benefits against potential risks. On the one hand, there are potential benefits for the less than 1% of Australian babies who receive an early diagnosis from newborn screening, and their families. On the other hand, there are potential risks for all screened newborns and their families. We also need to think about the implications for the health system of using genomics to screen for more conditions.

POTENTIAL REASONS IN FAVOUR OF USING GENOMICS IN THE NBS PROGRAM

BENEFITS TO THE CHILD AND FAMILY

Benefit 1: Using genomics for conditions that are already included in the NBS program could help doctors diagnose a baby's health condition faster and more accurately than current screening tests.

Using genomics in the NBS program could give doctors important information about a baby's DNA when they are still very young. Sometimes, it's hard to get the right diagnosis for a child's health condition, especially if it's rare. This can lead to a long, difficult journey for a family with many tests and doctor visits, sometimes lasting months or even years.

This process, known as the 'diagnostic odyssey', can be very stressful for families. Having genomic information in the newborn period could reduce the time families spend searching for answers, helping to reduce their distress, and avoiding unnecessary appointments.

Also, some treatments only work when the child has specific genetic changes. Having more detailed information about the genetic changes in a newborn would mean that treatments could be better targeted and families would avoid treatments that are unlikely to work.

Benefit 2: Using genomics in the NBS program could help find more rare health conditions in babies.

Using genomics could greatly increase the number of genetic conditions that are found through the NBS program.

Screening for more conditions could mean more families get the medical, psychological, social and practical support they need to care for their baby, as early as possible. Finding out about a child's health condition when the baby is a newborn can give a family time to plan and prepare for the right care and support.

Even if there is no treatment for a particular condition, knowing the child's diagnosis could help with getting financial support, accessing disability services, and making decisions about who will provide care at home. Families can also plan their finances and think about long-term care needs.

Benefit 3: Using genomics in the NBS program could help parents learn more about their own genes which could then inform their plans for having children in the future.

If genomics is used to analyse the DNA of the baby, this can also provide information for the parents about their own genetic make-up. Because most genetic disorders are inherited, if a baby is found to have a genetic condition, it is likely that one or both parents have a genetic change that could be passed on to other children.

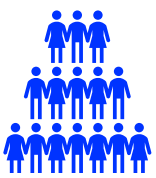
Families can use this knowledge to test children they already have (who might have the condition but not be showing any symptoms yet). They could also use the information to make decisions about having more children. For example they might choose to use reproductive technologies like in-vitro fertilisation (IVF) to test their embryos and lower the chance of having a baby with a serious genetic condition.

BENEFITS TO THE HEALTH SYSTEM

Benefit 4: Using genomics in the NBS program could help medical researchers improve the screening tests and treatments for rare conditions.

Most genetic conditions tested for in newborns are rare and doctors and researchers do not have much information about them. This can make it hard to diagnose them and to know how to treat them. With their parents' permission, a baby's DNA information could be used in research to help doctors and scientists better understand rare conditions.

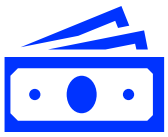
With more research, more information would become available to improve the accuracy of tests, reduce false alarms, and make sure the results are reliable. And once babies with a rare condition have been found, it could help medical researchers develop new treatments for other babies in the future who are found to have that condition.



Benefit 5: Using genomics in the NBS program could provide more information that is relevant to the different cultural groups in Australia.

Many of the research studies that doctors and researchers use to understand genetic health conditions and treatments have focused on people with European ancestry. Using genomics in the NBS program could allow researchers to collect information with parents' consent about people's DNA from a wider range of cultural backgrounds, including Indigenous people.

This information could improve scientists' understanding of conditions that affect different ancestry and cultural groups and help develop safe and effective treatments tailored to their needs. This information would need to be collected and stored in ways that are culturally safe for these Australians.



Benefit 6: Using genomics in the NBS program might be a good way to spend public health money.

Good value in spending public or government money in health means getting the best results for the money spent. While it's not yet clear if using genomics in NBS is good value for money, if it leads to earlier and more accurate diagnoses for some babies, this could be a good use of resources. It could prevent multiple rounds of testing and mean doctors can begin the right treatments sooner, preventing the baby's health from getting worse and reducing the need for more expensive care as the baby gets older.

Using genomics could also help with health service planning by providing more information on rare conditions, helping doctors and health services prepare for future needs and improve care. Although the costs to set it up might be high, these benefits might make it a good investment in the long run.

POTENTIAL REASONS AGAINST INTRODUCING GENOMICS INTO NBS

At the beginning of this section, we mentioned that making decisions about genomics in NBS requires balancing potential benefits against potential risks. We have considered the potential benefits: now we will consider some of the potential risks.

RISKS TO THE CHILD AND FAMILY

Risk 1: The use of genomics in the NBS program may not be equitable for all families.

Health equity means everyone having fair and equal opportunity to be healthy. To achieve equity, we need to provide the support each individual needs. To ensure that the use of genomics in the NBS program is fair for all families we would need to be sure that all families have access to the same screening tests. We also need to ensure that all families have

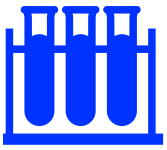
access to the health and support services needed to care for a child with one of the conditions found through the NBS program.

In the current NBS program, all families have access to the same tests and services. This is mostly because we screen for a limited number of conditions. But genomics would allow us to screen babies for hundreds of health conditions at once. Screening for so many more conditions would make it harder to ensure that all families have access to the same screening test and the same health and support services.

If genomics is used in the NBS program, it could be difficult to make sure families experience equity and have their individual needs met. This is because:

- Treatments are only available for 1 in 10 rare health conditions. Some babies could be diagnosed with conditions that don't have a treatment yet.
- Australians are unlikely to have the same access to the diverse medical care needed for such rare conditions. For example, it will be harder for a family from a remote town to get regular treatment at a specialist facility than a family living in a city.
- Genomic tests need data from an individual's ancestral group to work. Some genetic variants may be more common in one cultural group than another. Scientists do not have enough genomic data for every ancestral group, especially minority groups (groups that are smaller in a country's population). This means some families may not receive reliable diagnoses. Health conditions that mostly affect minority groups may not be tested for at all because there is still no reliable test for these health conditions.
- Genomic testing is complex. To understand how genomics is being used in the NBS program, families would need to be confident in communicating with health professionals, or have a good understanding of health science and the health system. This means some families will be able to understand information about genomic testing, while others may not.

- Different genomic tests have different levels of accuracy for different health conditions. This means some families may get clear and certain information, while other families may receive information about their child's health condition that is less clear and less certain.



Risk 2: Using genomics in the NBS program may give inaccurate screening test results for some conditions.

The accuracy of genomic tests is different for different health conditions and in people with different ancestries.

This means there is the potential that a test could tell us a baby has a health condition or is likely to develop a health condition, even when they do not have it and will not develop it (a 'false alarm'). If this happened, it could lead to unnecessary anxiety for parents during a time when they may already be under high stress. It could also affect parent-child bonding. It may also lead to the baby and its family getting more tests, seeing more doctors, or having extra treatments when they don't need to.

Inaccurate genomic tests could also fail to find a health condition that a baby has (a 'missed case'). This means a baby could miss out on early medical care which could impact their health.

Sometimes genomic sequencing can find genetic variants that science does not understand yet. This means it will not be clear if the variant will cause a health condition or how serious it might be. While these results are not wrong, if parents get uncertain results, there is potential for them to think that the child is going to get a disease.

Uncertainty and risk are very difficult concepts to explain and understand. For parents to understand these concepts, they will likely need meetings with a health professional before screening to discuss how accurate the test is, how the genetic condition might develop (or if it will at all), and how the test results could affect them emotionally or socially.

Risk 3: The use of genomics in the NBS program could lead to long-term uncertainty for families and create patients-in-waiting.

As a baby's genes can show what health problems the baby may develop in the future (although there is usually also a chance that the health problems won't develop at all).

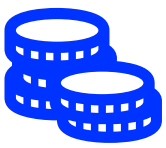
This information could cause emotional and psychological stress for families. Parents may experience unnecessary anxiety after learning that their child could develop a health problem later on.

There is also the potential for something known as overdiagnosis. Genomic screening might find signs of conditions that might never cause problems, but end up being treated all the same. This would mean the child has received unnecessary (and potentially costly or harmful) tests, interventions, or lifestyle changes. This can place financial, medical and emotional burdens on families.

Sometimes doctors are not sure if a particular gene is faulty or not, and so they will have regular check-ups with these babies and their families in case the baby becomes sick. As part of the check-ups the babies might have lots of tests and visits with different types of doctors and nurses. These check-ups could happen over many years. The costs of providing these check-ups in babies who might not become sick need to be thought about, as well as the costs of doing the genomic testing in the first place.

This long-term uncertainty can cause ongoing stress for families. Parents may become overprotective. They may see their child as especially vulnerable, even if the health condition has not yet developed. There is also the stress involved if the parents are told to come to a lot of medical appointments and tests. The long-term uncertainty may lead to babies becoming "patients-in-waiting". This is a term for those who have been

identified as being at risk for developing a condition, but who may or may not develop that condition later in life.



Risk 4: Using genomics in the NBS program could impact a child or family's ability to get life insurances.

Information about a baby's possible future health conditions could limit their access to certain insurance products throughout their lifetime. There is evidence that people will decline genomic testing if they believe insurance companies could use their genomic test results to stop them from buying insurance in the future.

In Australia, *health* insurance companies are already not allowed to use an individual's genomic data. However, *life* insurance companies can use some of an individual's genomic information when deciding to offer life insurance products to the individual.

The Australian government has recently committed to making a law to prevent life insurance companies from doing this, but it has not happened yet.

RISKS TO THE HEALTH SYSTEM

Risk 5: it might be challenging for the health sector and health workforce to deliver genomics in the NBS program.

Introducing genomics in the NBS program could be difficult for the health system to manage.

One problem is that there may not be enough people with the right training to explain genomics to families, especially since the information can be complicated. Health professionals would need special training to make sure they can explain things clearly and in a way that works for people from different backgrounds.

Another challenge is that using genomics in the NBS program might lead to more babies being diagnosed with genetic conditions, which

could create a higher demand for follow-up care and support. If there are not enough doctors, genetic counsellors, or services to help families after a diagnosis, it could be hard for the health system to keep up.

Risk 6: Using genomics in the NBS program will produce extra information – beyond the health information the program is looking for.

Using genomics in the NBS program will produce a lot more information than the current way of testing. This raises questions about what to do with this extra information, since screening results are usually only focused on specific conditions included in the program.

The extra information could include unexpected genetic information or information that scientists and doctors don't yet understand. There is also still a lot to learn about how changes in our genes cause different conditions, which means that genomic testing might pick up changes in genes that are not actually faulty.

Genomic testing might also uncover non-medical information, such as unexpected details about who is the biological father of the baby.

Risk 7: There is a risk of genomic data breaches.

The current NBS program has specific rules in place to keep the data it collects safe and secure. Because genomics would produce so much more information about the baby and its blood relatives, new rules would be needed to protect the genomic information.

A potential breach of this data – where information is accessed, changed, or used in ways it should not be (for example, by hacking) – is a significant risk.

Risk 8: Paying for genomics in the NBS program will mean funding is not available for other areas of health care.

The government does not have an endless supply of money to spend on healthcare. If government spends more money on genomic testing for all newborn babies, it might mean there is less money for things like genomic testing in people with cancer, care for young adults with mental health issues, or care for older people.

It's important to make sure that the way government spends its money is balanced and fair and that no-one misses out on health care because extra money has been spent on the NBS program.

Risk 9: It will take a lot of resources to store all the information that would be generated from using genomics in the NBS program.

It will take a lot of resources to do genomic testing and to store the genetic information for all newborns. There are questions about how much genetic information should be stored, and how long it is useful to keep the information. As testing methods get better it might make more sense to do new genomic testing in the future, rather than keep old genomic test results, but this is not clear right now.

Genetic information is stored in very large computers so there are questions about the cost of the computers, and the impact on the environment of running these computers all the time. The more genetic information is stored, the more it costs and the more energy is used.

5.9. WAYS OF THINKING: BIG ETHICAL AND LEGAL ISSUES TO CONSIDER

Thinking through all of these potential benefits and potential risks is challenging. In this section, we suggest some ways of thinking that might help. You will hear more about this in the final expert witness presentation, in our final online meeting. Some of this information is repeated from the earlier sections, but we are bringing it together here for your reference.

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ISSUE 1: THINKING ABOUT SCREENING POPULATIONS

As mentioned in Section 5.1, screening programs target populations (whole groups of people), not individuals.

As described earlier, making decisions about screening whole populations involves weighing up the potential benefits for a small number of people who will be found to have the condition through screening, against the potential risks for everyone who is screened.

If genomics is used in the NBS program every baby will have some or all of their genome sequenced. Most of these babies will be healthy. A small number will have a condition, but won't have symptoms yet. All of these babies need to be screened to find the small number with conditions.

Because of the focus on populations, several issues must be considered when thinking about the use of genomic sequencing as a screening tool. Some of these are:

1. **Screening effectiveness.** Just because genomic testing works well for diagnosing individual patients, it doesn't mean it will work as a screening test for everyone. For screening, we need to know if it helps improve health outcomes for large groups of people, and whether screening leads to better results compared with waiting until symptoms appear.
2. **Genetic diversity.** As we have mentioned, groups with different ancestry or cultural backgrounds may have different genetic changes. This means a test based on one group's genomic data may not work as well for other groups. This could lead to differences in how accurate the results are, potentially disadvantaging certain groups.
3. **Equity of access.** Factors like a person's income, healthcare infrastructure, and the availability of genetic counselling can vary for different groups of people. Ensuring that genomic sequencing is implemented so that all families, regardless of background, can benefit equally is essential.
4. **Cultural considerations.** Cultural factors can affect how people think about genomic testing. This can lead to differences in who chooses to participate in genomic newborn screening. It's important to understanding different communities' values and attitudes towards genetic information is to make sure genomic newborn screening is culturally safe for everyone.

ISSUE 2: THINKING ABOUT WHAT GENETIC INFORMATION REVEALS

Using genomics in the NBS program will produce information beyond the targeted health information the NBS programs are looking for. This creates uncertainties about what to do with information that is not reported back to the newborns' parents as part of the NBS program.

A baby's genomic information is not just about the baby in the here and now. It can reveal things about the baby's extended family, and about

the baby's future. We need to think about how to deal with additional information likely to be revealed by using genomics in the NBS program.

As you have heard, for example, genomic NBS could indicate conditions that are currently not able to be treated or may not affect the child until much later in life. A positive result for a particular condition could also indicate that other family members may also be affected by that condition or result in the need to sequence the DNA of both parents. These tests could reveal information about the health of the parents, and impact on their healthcare plans. These tests could reveal unwelcome information: for example, unexpected details about who is the biological father of the baby.

Thinking about how to deal with all of this information when we make decisions about using genomics in the NBS program means considering the potential impacts on family relationships and the baby's future self. There may be different expectations within families about what they should be told. For example, different family members may have different views on what to do with information about conditions that don't affect people until they are adults. Different family members may disagree on who within the family should be told about conditions that could affect other family members or future children.

ISSUE 3: THINKING ABOUT THE BABIES' RIGHTS AND INTERESTS

As discussed in the previous section, turning babies into patients-in-waiting can risk physical, financial, psychological and social harms to the baby and the family.

Another concern is the child's 'right to an open future'. Finding out genetic information about conditions that may only develop in adulthood could impose unnecessary labels or medical expectations on the child. It also removes the child's ability to decide for themselves whether they want to know this information in the future.

Also relevant here is the child's 'right to not know'. Providing information to parents in infancy about later-onset conditions removes the child's

choice to decide whether they want to know about these risks when they are older. For immediately threatening severe childhood conditions, this right is not relevant. But the case is more complex for conditions that do not affect people until they are older children, teenager or adults.

ISSUE 4: MAINTAINING PUBLIC TRUST

Around 99% of babies born in Australia undergo newborn screening as part of the existing program. Such high rates of participation indicate a high degree of public trust. Ideally, the use of genomic testing in the NBS program would not harm the reputation of the program.

However, the ability of genomics to detect a wider range of genetic variants could have an effect on the high levels of public trust that current newborn screening programs enjoy: for example, if a lot of people are told information that is uncertain, or that will not affect their baby until they are older.

False alarms arising from genomic testing could undermine confidence in traditional newborn screening methods (for example, the biochemical tests we talked about earlier). The difficulty of explaining genomic testing could raise concerns about a lack of transparency, leading to scepticism about the benefits of newborn screening in general. The questions we raised earlier about how this information will be stored, used, or potentially shared are especially relevant for genomic data, because it is so highly personal and sensitive. Public fear of data breaches could negatively impact the overall uptake of newborn screening.

ISSUE 5: CONSENT AND STORAGE FOR DATA AND BLOOD SPOTS

Consent is an ethical and legal requirement in the Australian NBS program. Consent is sought from parents or guardians for taking and analysing the blood sample, storing the sample and data, and for specified other uses, such as quality control and research on stored blood spot samples.

Consent matters because the information being collected, stored and used is highly personal. It is particularly important for samples or data

retained for other uses (such as medical research) because the newborn cannot consider future implications, or consent on their own behalf.

Introducing genomic NBS will make consent more complicated because of the greater complexity of the technology, and the greater volume and complexity of data. Retained biological samples or data could be re-analysed in the future to offer new interpretations.

In contrast to the clinical report currently generated in the NBS program, genomic sequence data can never be completely 'de-identified' – this is because your genomic information is unique to you. As discussed earlier, we need to think about how this sensitive data would be stored. We also need to think about how we would engage with families to help them make the right decision for them about having their baby's DNA sequenced.

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ISSUE 6: DIVERSITY, DIFFERENCE AND DISCRIMINATION

There is good evidence that people make ill-informed and often negative assumptions about what it is like to have a disability (including genetic conditions), or to have a child with a disability. People tend to see all disability prevention and cure as a good thing.

There is a risk that genomic sequencing could reinforce existing negative

attitudes, and possibly influence responses to people with disability in future. It could also change what is seen as “normal” and what is not.

There is already evidence that parents of children with genetic conditions (and the children themselves) face stigma and unfair treatment. In relation to parents, this includes negative views of them because they “chose” to have a child with disability, thereby creating a “burden” on society.

Image by Markus Spiske | Unsplash

This matters because using genomics in the NBS program will identify conditions the parents could then decide to avoid in future pregnancies. Some of the conditions that could be found using genomics in the NBS program will potentially be managed via medical or other interventions, such as diet, changes in the environment, or other health practices. Not all parents have the same ability to access these treatments. This means some children might be affected more than others, just because their parents were not able to access interventions. If the child’s condition is not prevented or treated, the child and their family may be seen as not having done enough or failing in some way.

Disability discrimination law sometimes makes such prejudice against the law. But the law cannot address all the stigma and harm potentially caused to the child and the family. It can deal only with specific cases of unfair treatment, one case at a time.

6. Key terms

Ancestry or ancestral group	The ethnic origin or cultural heritage to which a person identifies and/or to which a person's forebears are/were attached.
Biochemical test	The laboratory measurement of specimens, such as blood, to check for substances that could signal the presence of a medical condition.
Biomarker	A broad category of materials, such as biochemicals or genes, that signal a medical condition and can be revealed in tests.
Chromosome	Human DNA is arranged into 23 pairs of chromosomes, which means humans usually have 46 chromosomes altogether. We inherit one set of 23 from our biological mother, and the other set of 23 from our biological father.
De-identified	Making information about a person anonymous by removing identifying information such as name, address, or date of birth.
Diagnosis	The process of working out what condition a patient has, based on the patient's description of what is wrong, test results, and direct observation of the patient.
Diagnostic odyssey	A lengthy time and journey from when the suspected first symptoms appear, to making an accurate diagnosis.
DNA	Deoxyribonucleic acid: long strings that are made up of chemical units. Genes are small segments along the DNA strings.
Expressivity	The degree or severity of a trait or condition among those who do show it. The way the trait appears can vary widely.
Family planning	A set of actions, communications and decisions that allow individuals to decide if, when, and how to have children.
Gene	A segment of the DNA that contains instructions for building the body and making bodily functions work.
Genetic counsellors	Health professionals that help people make informed decisions about genetic testing, and understand the medical, psychological, and familial implications of their genetics.
Genetic disorder or condition	A medical condition caused in whole or in part by a change in the genes or DNA. Some conditions cause symptoms at birth, some cause symptoms later in life, while others may not cause symptoms at all.

Genetic variant	A gene that contains some form of change from the typical gene. Some genetic variants have no impact on health, while others lead to health problems or make it more likely that health problems will develop.
Genetics	The study of a specific gene or sets of genes.
Genetic testing	Tests that involve analysing one gene or a small number of genes at a time.
Genome	Refers to a person's entire genetic code or all of their DNA and chromosomes.
Genomic testing	Tests that involve analysing most or all of a person's genes/DNA.
Genotype	Refers to the genetic material responsible for unique traits or characteristics (or health conditions). For comparison, see "phenotype".
Late-onset or adult-onset condition	A health condition that arises later in life.
NBS	Newborn Bloodspot Screening, a program that offers a set of tests to identify babies at risk of illness from rare conditions.
Patient-in-waiting	A term for those who have been identified as being at risk for developing a condition, but who may or may not develop that condition later in life.
Penetrance	The proportion of individuals with a specific genetic variant (or mutation) who actually exhibit the associated trait or condition. In other words, it's about how often a genetic variant shows its effect in those who have it. For example, let's say 100 individuals have variant A but only 30 people exhibit the variant; and 100 individuals have variant B, but 60 people exhibit the variant. Variant A has higher penetrance (60%) than variant B (30%), which means variant A is more likely to manifest than variant B.
Phenotype	Refers to a person's observable traits or characteristics (or health conditions). A person's phenotype is influenced by genotype (or genetic material, see definition of "genotype") and environmental factors.
Pre-symptomatic	A state of health where a person is sick but the symptoms have not arisen.
Screening	Checking a large number of people who seem fine, to find out if they might have a health condition.
Sequencing	Tests that check a gene, many genes or all genes together as a single test.

7. Frequently asked questions

How are genomic tests already being used in children? Is this the same as using genomics in NBS?

Right now, genomics is mostly used for diagnosis when parents and doctors can see a child has symptoms but they are having trouble finding the cause. When genomics is used for diagnosis, it helps reduce the stress and anxiety for families. Very few children end up in the situation where genomics is needed to identify the health condition they have, but it can make a big difference for these children and their families.

Using genomics in NBS would be different to using genomics for diagnosis. This is because in the NBS program all the children appear to be healthy (they have no symptoms). Because the findings from genomics can be very complicated and uncertain, using genomics in the NBS program may cause stress and anxiety for some families. Because *every baby* is screened, this risk could affect a lot of families.

Can parents decide to screen for some rare diseases and not others?

Screening programs test many people – in the case of NBS, about 300,000 babies a year. This means it is not practically possible to provide different screening tests to different people. Population screening programs are only feasible at such a big scale because everyone receives the same test or set of tests and the laboratories can be set up to run these tests quickly and almost completely automatically.

Can we give parents a choice of using genomics in NBS for their child?

We don't think it will be possible for genomics to be optional in NBS, for the same reason as in the previous answer. Deciding to invest in genomics in NBS will require enormous resources and infrastructure. This investment will only make sense if a decision is made that genomic NBS is a good option, on balance, for the whole population.

Will all Australians have access to genomic NBS if it becomes available?

There is discussion about access and equity in earlier sections of this booklet: Sections 5.9 (Risk 1) and 5.10 (Issue 1).

If a decision is made to include a test or tests in the NBS, this is the same as deciding that this test or tests should be available to every baby born in Australia. If genomics was included in NBS, it would need to be available to everyone in Australia. This would need new investment and infrastructure, as discussed in sections 5.9 (Benefit 6 and Risk 9) .

What will happen to a baby's data after being screened? Could the data be stolen by cyberattacks?

As discussed in sections 5.9 (Risk 7) and 5.10 (Issue 4 and 5) data protection is a big issue for genomic NBS. Whenever data are stored, there is some risk of data breaches. The NBS is a public program, and governments would be responsible for the data from the program. The Australian Government and the state and territory governments already have big responsibilities to protect the large amounts of health data they hold about all of us (for example, all of the data in our hospital records, or our immunisation records). These responsibilities would also apply in genomic NBS.

Why don't we test parents for rare diseases instead?

People can choose to be tested before they have children to see if they might pass on a genetic condition. This is called reproductive carrier testing . Being a carrier means a person has a particular genetic change that is not enough to cause the condition in them, but might cause the condition in their child if their partner also has a similar genetic change. If a couple is at risk of passing on a health condition, they can choose to use assisted reproductive technologies (such as in vitro fertilisation or IVF) to reduce the chance their baby inherits that condition. However, people who are carriers do not always know they are, so will not always think they need carrier testing before they become pregnant. Also, sometimes neither parent is a carrier, and a genetic change happens for the first time in the baby. As you can see, this means some babies will

only have a condition detected if there is a screening program for all newborns.

Has any country implemented genomic NBS and if so, how well is it working?

In high income countries like Australia, there is a lot of interest in using genomics in newborn screening. So far, no country in the world is using genomics as part of routine newborn screening.

Research into the use of genomics in newborn screening is happening in countries including: Australia, England, Italy, Greece, and the United States of America.

In many of these countries, researchers can get permission to use the left-over dried blood on the card from the local NBS program. In some of these studies, that left-over blood is being used for genomics research. In other studies, a separate sample of blood or saliva is being collected for research.

Most studies are sequencing 500-600 genes thought to cause health conditions that are considered both serious and treatable. One study is sequencing 1,000 genes. But each study is testing a different selection of genes. Although there is some overlap in the genes being sequenced in these research projects, there is no agreement yet amongst researchers about which genes should be sequenced in an NBS program.

Are there alternative ways of achieving the benefits of genomics with fewer risks?

The main way to reduce the risks of using genomics in newborns would be to limit how much DNA gets sequenced.

As you will hear, using genomics in NBS reveals a lot more information about the baby and their blood relatives, but this information is complicated and uncertain and may cause anxiety and stress for families. A decision could be made to sequence only specific genes. This decision is a trade-off: it would mean that we do not pick up as many conditions as we might if we sequence the whole genome, but it would also limit the number of families experiencing anxiety and stress because of their baby's test results.

As you will also hear, if the NBS began sequencing the genome of all Australian babies, it may or may not report, and/or store all of the information in that genome (see Section 4.1).

Decisions about what to look for, what to report and what to store are decisions about the best balance of potential risks against potential benefits. This is the main focus of our jury deliberations: where should we draw those lines?

Will commercial companies be involved in genomic NBS?

As noted earlier, the NBS program in Australia is the shared responsibility of the Commonwealth, State and Territory governments. All of the dried bloodspots from screened newborns are analysed in government laboratories, and the cards and information from them are stored securely by government. If a newborn is found to have a condition via the NBS program they are immediately referred to the doctors and health professionals who will care for them in the public health system.

There are commercial companies that sell the equipment used for genomic sequencing machines to laboratories. If genomics was used in NBS, more of this equipment would need to be purchased by the newborn screening laboratories.

There are also commercial companies that sell treatments for the conditions that might be screened for if genomics is used in the NBS program. If genomics is used in the NBS program, governments would also need to consider if these treatments are safe and effective, and how much to pay for them.

It is possible that some privately owned laboratories might offer genomic testing to families, but this use of genomics would be outside the NBS program, and families would need to be careful about making sure their personal data is stored securely and not used for anything they have not agreed to. Also, because such testing would be done outside the public health system, families would have to find their own doctors to treat any condition that is picked up.

Is there governance in place for genomic NBS? Does it include Aboriginal and Torres Strait Islander leadership?

As described above, there is already strong governance in place for NBS but this would need to be strengthened further to cover the use of genomics in NBS. As part of this strengthening extra attention would need to be given to ensuring there is Indigenous leadership around how genomics data and information from Aboriginal and Torres Strait Islander peoples should be stored and used. This is because in the past governments in Australia have taken biological samples from Aboriginal and Torres Strait Islander people without their permission and also used this material for research and other purposes without their permission.

Is genomic NBS suited to the Australian population, given how diverse we are?

Not completely. At the moment, genetic and genomic tests are not always useful for all Australians because of limited data from some groups. Most international genomic research has been with people of European ancestry. So, for people from other ancestries, genomic testing may miss genetic changes that cause some medical conditions. But this is changing, and every year we are learning more and more about genetic changes in people with non-European ancestries. It will be important to consider this new knowledge as it becomes available.

Can we say yes to genomic NBS and opt out of research that could be conducted using our data?

The likely answer is yes. Currently, in the NBS program parents can say yes to screening and say no to research being done using the leftover blood. The same could apply if genomics was being used in the NBS program.

What are the most recent developments in Australia regarding newborn bloodspot screening and genomics?

The Australian government has promised to introduce new laws that would ban life insurance companies from asking for genetic or genomic data. There is more on this in Section 5.9 of this booklet (see Risk 4).

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For updates about the citizens' jury

https://uow.info/genomics4newbornsjury

or scan the QR code below

This information booklet for the **National Australian citizens' jury on using genomics in newborn bloodspot screening** was developed by the Australian Centre for Health Engagement, Evidence and Values.

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