# Expert Q&A Sessions

***Researchers' note:*** *This document contains materials generated during the Zoom session with the experts. The first part is the transcript of the live Q&A session. The transcript has been de-identified, and slightly edited for ease of reading. The second part contains the expert's response to additional questions that were not responded to during the Zoom session. After the Zoom session, these questions were collated and sent to the expert who was given a few days to provide a written response.*

**Date of Zoom session**: Sunday 23 March 2025

**Expert**: Professor Margaret Otlowski, Director, Centre for Law & Genetics, University of Tasmania; Professor Ainsley Newson, Professor of Bioethics, Sydney Health Ethics, University of Sydney

**Title of presentation**: Concepts and ideas to help you think through the information: Legal and ethical considerations

## PART 1: Q&A TRANSCRIPT

*Juror: There was a document on VisionsLive with regards to the LINEAGE studies and something, and I wasn't really sure if it was relevant to this, but like a word that really stood out to me in that document, which I'm not sure is or isn't relevant to this discussion, was the word stakeholders that sort of like made me stand up and like what? And are there any stakeholders in this particular field studying with the genomic newborn blood spot screening? And if so, who are they and who are they likely to be?*

Ainsley: Thanks for your question, Mark. So LINEAGE is another project funded by the same funder that's funding this project actually. But [in that project] we're looking very specifically at ethical and legal issues in how we control what happens to genomic data from tests, including potentially this one in the future. And Marg, who's here with us today, is also involved in that project. Your word 'stakeholder is, it's an interesting word. And actually, I think if you asked Yves or Stacey or any of the other people here who are working as academics in this project, we might all have slightly different ideas about what a stakeholder is. We might also have slightly different ideas about whether it's a good word to use or not. But for the purposes of this jury, what I would say is a stakeholder is an individual or a group of individuals who has an interest in what happens. And so to my mind at least, there are definitely stakeholders involved in this question of under what conditions we should use genomics in newborn screening in the future. And just off the top of my head, I'd say it's people like you here, members of the Australian public, it's parents, it's future parents, it's the government, it's people who run sequencing laboratories, it's the health professionals who might care for families in the future and so on. So it's [i.e., 'stakeholder'] basically just a way, a way that academics sometimes come up with of using a fancier word when you can probably use a simpler one just to describe all of the different groups whose opinions matter.

*Mark: And I think the automatic presumption that came to my mind was like with people that buy stocks, you know, that's a word using in regards to commercial aspects of products or processes or research or what are the commercial implications? Who are the stakeholders and they're going to profit from it. That that was the presumption that that, that I made when I heard that word, when I read the word in the document. Good, good to clear it up then we don't just mean commercial stakeholders. We would mean anyone that has a, has a stake or has an interest in, in what happens. So would that include commercial applications as well?*

Ainsley: It's a difficult question and I know I've read all the Q&A transcripts from the experts that you've talked to so far and I think this question has already come up. I think it really depends on how this [i.e., genomics in NBS] happens, if it happens at all in Australia in the future. And it might be that commercial entities are involved in the testing pathway in some way, for example by providing some of the equipment that gets used to generate the sequence information. But there are other ways that commercial entities might be involved as well. And I think the way that newborn screening is run at the moment in Australia is a pretty good indication of what it might look like going forward, in the sense that it's publicly funded and it's managed by States and territories. And so unless it was a stand-alone commercially obtainable test, then that would be a completely different thing because that wouldn't be 'screening' in the way that we use it for newborns in Australia at the moment.

*Juror: Mine was sort of along the lines of the whole consent issue that was raised when it comes to obviously the current blood spot screening programme that's in place. And like you said, like there's 99% of people quite comfortable and you're obviously worried about adding this one will make people like lose that trust. And you know, how do you do consent? Would it be an option to have it just as an added extra to the current scheme or would it have to be like either it's in or it's out? Does that make sense?*

Marg: Although I think Ainsley probably knows more about the some of those logistics. Look, anything is possible as we look at a future where these things need to be designed. The current screening has clear purpose and private and validity, but it doesn't deal with many of the genetic conditions. So without asking for permission to test for those and particularly through genomic sequencing, we're not going to get to that. So it is possible rather than necessarily replacing biochemical testing that we do it in addition and that people have the choice whether to go down this path and that might be appropriate because so many people may have misgivings and that may be a way that we can protect public trust. So it is a useful option, I think to keep in the mix and maybe one for, you know, early days while we need to be really, really cautious.

Ainsley: I'll, I'll just add to what Marg said. I'd say two things. One is we could divide it up in the way that I think you suggested Bronwyn, which is people just consenting to biochemical screening as it happens now and then consenting to the use of genomics as an extra add on to what is currently happening. I think, off the top of my head, I think that would probably be feasible, but obviously what the content of that consent for the genomic aspect of it would need to be carefully thought through. I think where things might get a little bit trickier is if we turn it into like a full 'menu' approach where we're asking people to pick out the certain kinds of genetic conditions that they might be interested in or not. I think that level of differentiation [of consent] is going to be quite resource intensive and quite challenging to do for 300,000 families a year. So, it's [i.e., consent] kind-of become that trade-off between allowing freedom to choose [what to screen in your baby] yet the complexity [more freedom will generate] for the programme as a whole. And the only other thing I'd say is some experts are using this complexity of consent as a reason to argue in favour of keeping the use of genomics in newborn screening quite tightly focused. They're using it [as a reason] to look for less things rather than more, just because we want to make sure that people really understand what it is that they are potentially going to find out. But I can provide a bit more about that in written form afterwards if you like.

*Juror: Yeah, my question, it also relates like the whole consent part of it. So do we have a way of reliably giving information about the genomics programme to parents like maybe before the babies born and they're in hospital, tired, not really receptive to information. You have a way of getting that information like before that time?*

Marg: You make a really good point about timing. And of course, consent to be valid needs to be informed consent. So the people need to know exactly what they're being asked to consent to, and you want to ensure that they do have the state of mind to be able to absorb the information, ask questions about anything they don't understand and so on. And genomics of its nature, because it is so involved and because we're not looking just for one thing, but potentially multiple gene variants that could create disease, there's a lot of information that would need to be imparted. So there's a very real concern that the amount of information is difficult to convey meaningfully in a way that people actually understand the implications in a very summary form. So you'll probably end up with something that is longer than a page if you set out all the issues. And I guess it really depends on whether are we just talking about the consent to the test, which is more contained or is it consent to then using the genomic information, the data, the sequencing and so on for future research uses, which is much, much more open ended. So if we're going to allow use beyond looking for a gene variation that will expand the volume of information. So you're quite right that it's possible we get more opportunity for meaningful consent if we think about the timing and giving information to people early, having perhaps podcasts sort of explaining a lot of the issues. There may be ways that this can be done to benefit the wider public and they're probably important things that need to be considered if we do go down the track of genomic sequencing.

Ainsley: I'll just say one super quick thing. In addition, we have a couple of examples from research projects. I think you met Professor Zornitza Stark and she talked to you a little bit about the project she was involved with. They used an online portal for engagement with parents and that was great. And we also have a technical thing that gets used sometimes called a 'decision aid' and it's like a structured instrument that can be used to help people make a decision about what to do. The only thing I would then ask you as our jurors is to think about how we could build [use of an instrument like] that into the existing pathways of antenatal care and care for people who are having a baby.

*Facilitator: So there's a couple of questions, Marg and Ainsley, in the chat around access to data and who might be able to get access to the data. So both in relation to potentially theft of the data and who might want to steal the data and for what purpose, and also access by insurers, by drug companies, by police. I wonder if you could both speak to that question for us. It's a big one.*

Ainsley: I'm also mindful Marg has in the past written a paper about the concept of theft of DNA. But I'd say this is one way we have to look at what current regulation might or might not give us reassurance about. But I think what we also want to make sure of is that people who decide to go into this, if we decide to add genomics to newborn screening and, and parents are weighing it up about what to do, what we need to know is that they feel confident deciding whether to engage in this or not. And part of that feeling confident is going to be around knowing that appropriate protections are in place. Now we have pretty complex privacy regulation in Australia at the moment and it is under review by the government and genetics was one of the things they were looking at when deciding how these laws should develop in the future. I think there possibly are some black spots at the moment where we're not really sure [about what uses of data are permitted or prohibited]. That said, this type of data that we're, we're generating genomic data every day in clinical practise in Australia. And that data is all already subject to pretty stringent privacy and security requirements. And there's no reason why this data would be treated any differently. And so it's not just carte blanche. Anyone can't just come in and take it if they want to. There's obviously all of the IT security around it, but there's also regulatory security around it governed by privacy regulation at both Commonwealth and state level. But it might be that we actually need to do some specific shoring up of that regulation for something of this scale so that families considering this feel confident.

Marg: I think one of the really significant things about creating this kind of resource and especially if it's all accumulated together is this idea of a honeypot that becomes very attractive also for potential misuse. And it's hard to kind of identify what would be the uses. Certainly genomic data is always

potentially identifiable. It can be kind of linked up with other information, people, people have weird motivation. So, I couldn't predict what, what the reasons would be, whether it's to then seek money from people for it to be returned or, you know, you just don't kind of know what the evil motivations might be. But coming back to the issue of confidence and having trust, we do just need to ensure that the method of storage and access, are secure so people can have the necessary assurances. I don't think we have too many problems with insurance. I mean, insurers aren't entitled to this kind of information. Sometimes when you apply for something, you have to share information, but that's a different thing. You know, even police wouldn't automatically have access, only with a court order. Might there be a reason that information would have to be disclosed? So the actual entitlement to access would be very, very limited indeed. It's information that would be for the health purpose of the baby and if permission is being given also for research purposes. But yeah, you can't pretend that there aren't risks And we should never, in inviting people to participate, make assurances that we can't keep. So we can never say information is 100% secure. We have to be really transparent about risk and potential breach and, you know, misuse and so on. So we do need to define those risks even if they are small. So that's very helpful.

*Facilitator: So [juror], I'm going to come to you in just a sec, but I just wanted to go to a comment from [another juror] around the importance of thinking about potential impact on other family members of the results for the baby and the need for the jury to consider that in their deliberations. Ainsley and Marg, I wondered if you wanted to add anything on that particular issue for the jury to keep in mind.*

Ainsley: So I might start, Marg, if that's OK. And I'm just having that moment where, oh my goodness, my mind's just going completely blank. So I hope I do this question justice. Because genetic information is shared within families, it is important to consider the potential implications for family members when we're thinking about providing genetic information at this point in time to this group of people, namely newborns. And in the current policy framework for newborn screening in Australia, there are a couple of mentions of possible impact beyond the newborn. One of them I think is reproductive decision making in the future. So if a diagnosis is made in a child, then usually, not always, but usually, that information has come down from the parents. And so actually that gives them information that they may wish to bear in mind in a future pregnancy or at least to have counselling about. But also if the condition is X-linked or dominantly inherited, for example, then I mean all of these conditions actually have implications for other relatives beyond just the parents as well. But where I think the interesting question arises in that [i.e., family implications] is what weight should that possible information have in deciding whether to look for something or not in a newborn? So what if the only benefit in in that information was the benefit to the family members? What if there wasn't actually any benefit to the newborn or at least not a benefit in the newborn. So I think that's when you have to start trading off some of these risks and benefits. And who is this screening test going to be for? So some of the experts are starting to think about this in the literature and the place that the possible value of this information for family members other than the newborn is being actively considered.

Marg: Just to add the importance of managing expectations, making clear what the limitations on what can be looked for and what can be reasonably useful and found. And as he said, the real importance of keeping the focus tight to manage the risks, because the more information you get, the more complex it is to decide how it's used.

*Juror: The, the segue is very well into the question I was thinking about which is also related to a family implications. And, and as you mentioned, I was wondering in the current consent process or in the consent process envisioned, is there any consideration in the consent process to the impact of one of the family members? As in when, when parents are asked about the consent, is that made clear in the in the consent documents, the informed consent documents that let's say screening done on let's say a male child may have an impact on let's say an unscreened female child who's already grown up or something. And is it and further on that to uncles to grandparents, etcetera, etcetera. Is that within the already you considered in the form consecutive documents? Or is that something that sort of still in the works?*

Marg: I can start, but Ainsley will probably know better but I think it would need tolook to the future. So regardless of what is even done currently, but I'm sure with other genomic studies that the nature of the information is a critical aspect, the fact that we're dealing with what we describe as genomic information. So clearly it's personal information, which the Privacy Act protects, it's health information and the genomic information of itself is also treated as sensitive information requiring the highest level of protection. So people would need to be told that it has, you know, part of the reason it's sensitive is that it's not just about the individual, that it is familial, that it can tell you not just about the person you're testing, but by its nature, about other family members. So I think there's avery basic piece of information that's got to be in an information sheet about anything to do with genomic testing. And, and as I'm sure we'll confirm that this is the indeed already the case.

Ainsley: I think so Jeffrey, your question, I think you were asking about the way that things happen at the moment and Marg's answer bought out this nice distinction, which is what happens now isn't genomic screening in newborn screening, but we can look at best practice in the programme at the moment to help us decide what we should do going forward. Newborn screening at the moment is managed by States and territories, as I think you've probably discussed in listening to other presentations that have come before ours. And each state or territory provides its own information to families. So I've read some of the parent information sheets in the course of my career, but I haven't read them all in the past week. So I can't give you kind of an exact answer to your question.. The consent form is pretty streamlined, but there will be information leaflets that will cover off quite a lot of the aspects of the programme. And if by chance it's not explicitly covered in the way that you described it in your question, in the event that a newborn is found to screen positive, that is they return a positive result on the initial test and at the moment in the programme they would come back in for subsequent testing, then that it is at that point that implications for family members are discussed. So if a diagnosis is made, that is the point at which the potential implications would arise and that's where it would be discussed. But if we move to genomics, the way that we present that aspect, the potential implications for family members might be something that needs to change to say more uniformly form part of pre-test information and thinking.

Marg: Can I just make clear when I was talking about, you know, what happens in in existing genetic testing and sequencing? I'm talking about not in the newborn common sense. Yeah, there is some you know, obviously there's genetic testing, there's for research purposes, there can be sequencing. And I'm just talking about, you know, expectations of what people would be told there could guide us to the future for newborn screening.

## PART 2: RESPONSE TO ADDITIONAL QUESTIONS

***Q**: Will the data be kept on the big hard drives or in I cloud ?*

**Answer**: This is not yet known. An evaluation of storage options and their governance would have to be part of any setup. You may wish to consider what difference this would make, and/or whether you may want to make any stipulations about this as part of any conditions for the use of genomics in NBS that you would want to stipulate. It is likely that data would be stored on a secure cloud-based server.

***Q:*** *Another one is duty of care if a severe issue is detected but not disclosed, could there be implications?*

**Answer**: It is very unlikely, unless there was a genuine laboratory error (which would be very rare in itself) that a severe issue would be detected but not disclosed. This is because the severity of a condition is a key consideration in criteria for newborn screening, and has also been a key aspect for the design of the gene lists in the existing research studies to gather evidence about genomic newborn screening. Any decision about what to screen for would be made under a multi-disciplinary consensus process, likely including input from citizens/patients. If a decision was made on reasonable grounds to exclude a condition and a child was then born and went on to develop that condition, a court would look to how the decision to exclude that condition was made.

The answer to this depends on whether the condition/gene variant falls within the scope of what is being looked for and is reportable back to parents (ie serious health condition in newborns which can be treated through early intervention.) If it falls within the scope of one of the conditions on the list for genomic NBS a failure to disclose that due to oversight/negligence would likely breach the duty of care owed to that patient/family. However, if it is not in scope for the genomic NBS, for example on the grounds that it is not a treatable condition and therefore not reportable, it would likely not be a breach of duty of care which requires a departure from accepted professional standards.