Day 1

Personalised medicine and you Q&A

Queensland Genomics Health Alliance

- **Katrina Cutler, Communication and Engagement Manager**

Katrina: Did anyone have any questions of Aideen first up? Excellent.

Audience member: Thank you, that was very clear. In terms of things like mental illness, which people have been showing affect a number of genes possibly that express the illness, we don't know whether it's environmentally caused the protein or the changes, or whether that is... what's your thinking about this? Where are you going to go with this, with mental health?

Aideen: What's your name?

Audience member: Elsa.

Aideen: Elsa, that's an excellent question. I was trying to figure out how much to put into this presentation before I thought everybody's eyes would glaze over and they'd nod off to sleep. So that is what we call a multifactorial. Instead of a rare disorder like Huntington's disease or cystic fibrosis where the gene is the big strong effect and the environment has some role, but it's fairly minor. In instances like mental health you've got multiple variants in lots of different genes that are adding together to add what we call a cumulative genetic background risk and then there's a lot of other environmental factors, life experiences, viruses for all we know, various other things. This is where we do a lot of hand waving, because the truth is nobody really understands it completely at this point. But you're absolutely right, there is a genetic predisposition there, but it's usually from multiple different genes and it's a small additive effect for those. She's behind you!

Audience member: Hi, I'm Suzanne from Caboolture Hospital. How accessible are these tests? Is this something that anyone can do, or is that the plan,

or do you have to go through the hospital? Do you have to be unwell before they'll do these?

Aideen: Again, another excellent question. Thanks so much Suzanne. Pharmacogenomic isn't quite here yet. We're working on that and we're based at the PA Hospital and hoping to roll that out there in the next year or two for pharmacogenomic. For cancer genomic sequencing, that limited panel of the old-fashioned five or six genes is available to most cancer patients in Queensland where there is demonstrated evidence that that would benefit them. Whole exom e sequencing is only available to patients at the princess Alexandra Hospital and we're doing it as a pilot and what we're hoping to prove is it's more sensitive, better health outcomes and that the economics also makes sense and that it'll become standard of care over time.

Katrina: Just further to that Aideen, could you talk about what would be the referral pathway for a patient who okay I've got a cancer condition, I think I might benefit from being referred for genomic sequencing.

Aideen: Absolutely. So if you're found to have a mutilation that's just in your cancer, then that's something that usually your oncologist would talk to you about and talk through that with you. Sometimes there's a genetic counsellor or a clinical geneticist. If you're found to have a mutilation in a gene you were born with, you inherited from your mum and dad which increased your risk of getting that cancer in the first place, then your doctor would refer you to Genetic Health Queensland, based here in Brisbane, but they've got outreach clinics throughout Queensland and you would meet with a lovely genetic counsellor like Jenny who you heard from earlier, or a clinical geneticist and you would talk through what that finding meant for you in terms of what screening you should have now and in the future and what the risks might be to other members of your family and what testing is available to them, too.

Katrina: I think there's private services available, too, I guess.

Aideen: There are, this is entirely true, absolutely. There's a couple of private clinical geneticists in this area. Not so much in the rest of Queensland, but on the south-east corner there's a couple of private clinical geneticists, too. Depending on the waiting list and urgency Genetic Health Queensland try to triage people depending on the urgency of their condition and how much their clinician needs that information in order to make decisions about how to manage them. If not, you could always go privately, as well.

Katrina: Excellent. Any other questions? Lovely.

Audience member: Just wondering where the notion of gene switching fits into this conversation? Where they can be switched on and off.

Aideen: Switched on and off, yes okay. Another interesting really good question. So there's various gene therapies that people are looking at for various different conditions and switching it on and off idea has been going on for a while. Very often just popping it in if it's missing particularly in conditions caused by disorders that affect the bone marrow, because it's pretty easy to get at the bone marrow. The idea is that you destroy what remains of the bone marrow and do a bone marrow translate or you do a transplant with the gene working. So that's gene therapy in the original form. When they originally introduced it, it was in children that really had a very poor prognosis if they didn't have it and they generally responded pretty well, but sometimes there were downstream effects like the gene that was slotted into the wrong place and put them at increased risk for malignancies later in life, but that was back in the 90s and they've gotten better at doing it since then. There's a wide variety of conditions you can do it for, but the trick is getting access to the tissue that's affected. Getting access to the brain is more challenging than getting access to the bone marrow and that sort of thing.

Katrina: Last question.

Audience member: Mine's in relation to population health, where you have broad spectrum identified disorders or mutilations and I'm thinking of a couple of things, the impact of resistance to antibiotics, or big um Mutitjulu ological

information that's changing the nature of cancer treatments.

Aideen: Resistance in terms of antibiotics and cancer treatments?

Audience member: Yes.

Aideen: Exactly. Two different of kind origins. With the antibiotics obviously that's happened over decades and you get this cumulative effect whereby the bug that's resistant to penicillin is more likely to stick around and, therefore, becomes a little bit more prevalent over time. So it enjoys a kind of Darwinian advantage as it were, so it becomes more prevalent. On the cancer side of things with the resistance, what you get is- you remember I said you get mutilations in the cells that lead to the cancer? Well the cancer itself continues to accrue new mutations over time. You might do a diagnosis where you're saying right this person has this mutilation they're likely to respond this therapy. In the meantime you get a new mutilation that confers resistance to that and it happens randomly, but because that cell is now no longer sensitive to the treatment, it enjoys a Darwinian advantage and it grows more and you get more of a colonial expansion of that group of cells. How do we do that? So the population, so the resistance side of things, that's one of the things we're looking for when we do the sequencing of cancer patients is we're looking for mutilations that confer resistance and those are really important to find in that regard. That's in that individual person. It happened by chance, it doesn't have a population significance, it just happened randomly. But we do find that people who are given certain types of treatment are more likely to get mutilations in certain types of genes and so, therefore, you can see the same mutilation in lots of people with the same cancer just because very often the treatment you gave them in the first place can actually lead to some of the resistance issues later.

Audience member: So are you suggesting that there's an advantage because of the genetic testing to the system in terms of improving the likelihood of a good outcome- is that what you're saying?

Aideen: Exactly. If you can say ahead of time "that tumour's resistant, there's no point putting that person on that medication, they're only going to get sick and not respond".

Audience member: Similarly with antibiotics, it's an alternative strategy rather than just keep creating new antibiotics.

Aideen: Exactly. One of the grants that QGHA gave out this past year was looking at exactly that, sequencing the bugs to find out what the bugs were sensitive to, so you gave the person with the infection the right treatment to start off with instead of going through the usual suspects and then finding they were resistant to them anyway. So that's one of the goals of the QGHA, as well.

Katrina: Which segues nicely into the next thing. Thank you for your interest everybody, and thank you Aideen. (APPLAUSE)

Katrina: Okay, now it's my time for a quick overview of Queensland Genomics Health Alliance and the projects we found, but first up, my anecdote of my first memory of the health care system and how I felt about it- that was the thing I forgot to add in my question, but luckily Aideen added that part. My first memory of the health system was when I was 6 or 7 years old outside Sunlands shopping centre at Caloundra jostling with choir friends, fell over, flat on my face. Split chin open. The door parent of a friend that was looking after me had to take me to hospital and I got four stitches. I don't know how my mum felt about getting me back after choir practice with a couple of stitches. But how I remember feeling about that besides "in pain" was I felt kind of it was great to be looked after. I really loved being cared for by all these really efficient and effective seeming people and all these caring nurses, so that was my first memory of the health system. But now I'm going to talk to you briefly about Queensland Genomics Health Alliance and what we're trying to do. So yeah, the first thing to be clear about is we're not a service. We don't provide genomic or genetic services. There are genetic health services in Queensland, the public service is called Genetic Health Queensland and they are kindly with us today on the booth if you have any questions, feel free and talk to them. There are private services as well, but what Queensland Genomics

Health Alliance is, is really a collaboration facilitator. What we're about is marrying up research and clinical people to implement genomics into every day health care. We're a 5-year program. We started in 2016 and our aim is to integrate genomics into health care. We've got $25 million to do it and we're funded by Queensland Health to do so, so we do that through a series of research projects, which I'll talk a little bit about. Our research projects address a list of eight objectives, which really have been identified as the challenges to implementing genomics into health care in Queensland. The way we're designed is we have a series of clinical demonstration projects which are essentially based around disease areas and we've got a series of capability building work streams which are really about building the infrastructure that the Queensland Health system needs to be able to integrate genomics. The projects that we run are the ethics legal and social implications of genomics, which Nic will talk to you about later today. Genomic workforce development, which looks at how do we make sure that clinicians not just genetic health care professionals, but how do we make sure that your GP if you go and see them and you've got a question about genomics and genetics that they know how to answer you and how to refer you properly. It's not just your GP, it's your oncologist, all of the other health professionals that you might deal with. Many of them know far less about genomics than what some people who have received treatment for example. The evaluation of clinical genomics and that looks at how does the economic case stack up? Is it going to save the health system money to implement some of these tests and treatments. Genetic testing innovation is a project that looks at making sure the lab capability across Queensland is appropriately accredited and tests are going to the right place and being communicated safely. And then genomic information management looks at how do we build an information system that can deal with the huge amounts of data that we're looking at if we implement genomics into health care? As I said, those capability areas are kind of being tested through a series of clinical projects which really are about disease areas and the four disease areas we're focusing on in our first round are melanoma, lung cancer, a form of diabetes called MODY and our infectious disease project, which Aideen mentioned before which is about sequencing bacterial genomes, not human genomes to basically stop the spread of hospital-acquired infections so we can prevent outbreaks of hospital-acquired infections. So our projects are all collaborations across these different

organisations and they involve all of the different disciplines, as well. So that's kind of the Queensland Genomics Health Alliance in a nutshell. Does anyone have any questions about us as an organisation? I kind of feel like we're the less interesting part of it. I understand if you don't- oo, a question. Excellent.

>>: Hi there, it's Lauren from the Sunshine Coast. So it's excellent that this is available in Queensland. I'm just wondering where we sit in the spectrum of are we just in the infancy, ahead of the ball game with other States in Australia?

>>: That's a really good question and I think you can probably guess the answer. We're not coming last, but we're certainly seeing leadership shown in the southern States. The model for Queensland Genomics Health Alliance is based on the model for Melbourne Genomics Health Alliance. They established a couple of years before that and we've been lucky to stand on the shoulders of the learning they've had there. There's an Australian Genomics Health Alliance which is a collaboration that deals with the national issues, because a lot of it isn't about hospitals, a lot of it is about national issues. So we're doing really well. The Government's chosen to fund this to the tune of $25 million. That was a couple of years ago, I guess the question is what happens after that five years.

>>: Are there opportunities to promote your work?

>>: What we're looking at is how do we address those eight objectives identified as the main challenges? We need to make sure we uplift public knowledge, but at the same time it's a difficult thing to do when the services aren't necessarily available yet, but people need to learn about them. There's a private market around direct consumer testing 23 and Me, those organisations that are effective at marketing their services to the public. For us working in the public health space, how do we make sure that consumers are adequately educated about this so they can make informed decisions about their decision whether or not to get that 23 testing.

>>: Hi. The ethical dilemma. Who owns the genetic make-up of that person you've tested and what's the safeguards of that staying within that parameter?

>>: That's a really good question and I am going to leave that to Nic who's up next to answer. Nic is a co-lead on a social ethical project and she is grappling with a lot of those issues and she'll probably get a bit into it in her talk. If she doesn't answer it, feel free to ask that question again. One last question about Queensland Genomics Health Alliance.

>>: Taking you back to the last thing, do you have consumers in relation to your committee?

>>: Oh my gosh, I can't believe I forgot to mention. Okay, so the whole reason we're here, we've got a community representative advisory group and many of them are here today. The members that are here, can you please stand up so everyone can see you. Excellent. So our community group represents, has membership from the broad community involved in genomics. We've got a bunch of amazing consumers on there, but also people who work in health care delivery, but also research in this space. They were kind of a key part of what we had to get set up to get going. We've got things moving on that this year which has been great and Erin Evans our chair will talk to you about the community group later. I might hand over to Nic now to talk you through some of the ethicals legal and social implications of genomics and health care. I'm just going to skip through. Nic, I need to remind you your anecdote about your early experience of the health care system and how you felt.

>>: Thank you, Katrina. I'm going to stand over here, so I can see my slides as well, if that's all right. My first personal experience of the health system, I'm very fortunate, because it was actually only six years ago and that was when I had my first child who was born at RWH, just across the road in some direction. As Katrina mentioned I'm involved with QGHA. I'm part of the ethics, legal and social implications. I will touch on those throughout this talk. I hope to answer your question midway through. If I don't, then just remind me as well. By way of background, my day job is I'm a medical genomics researcher, so I work at QMR

Berghofer. I run a group of people with a medical genomics group and we analyse genome data all the time in a research perspective and mainly focus on whole genome analysis. That whole genome chromosome, cancer sequencing, that's my background. Over the past few years we've analysed thousands of research participants who've kindly taken part in our studies. I'm going to start with this slide. I don't expect you to read it at all, but I wanted to know who had heard of the Human Genome Project? A lot of people, that's good. It was a big project at the time. It started in 1990 and the first draft was released in about 2001 and deemed to be finished in 2003 and I put this up because this was a really defining project for anyone doing genome work or personalised genomic-type work. This is the project where it took 13 years to complete and the sequence of the human genome was done. They started each chromosome at a time and sequenced the chromosomes and put them back together and told us the sequence. 13 years it cost about $2.7 billion rumoured. Now we can sequence a person's genome for about $1700 and do it in a few days. This technology is being driven into the health system and so the genome project itself is really critical. For someone working in genomics for counsellors analysing genome data they would refer back to this human genome framework and template we've got so it was really important and it's what's allowing the personalised medicine to commence. But as part of this project, the people are really sophisticated and right at the beginning there was an ethical, legal and social program that was formed. This was first formed in the US by the NIH, because they realised this project could have ethical implication. You're sequencing someone's genome and finding out all there is about them, but not just about them. You're sequencing half of their mum and dad essentially as well and learning about their potential children as well. So the implications are not just for the patient, but also for their whole family potentially. And then this has been cut off at the bottom here, but in 1996 this is a really defining moment for this genome project and that's because there was a meeting that some principles came out of the meeting called the Bermuda principles and this was an agreement from all of the researchers in this big international effort that all the data coming out of this project will be shared instantly. For science, this was kind of a bit backwards. In science people used to hold on to all their data, publish it and then release it. So this was the first time where no, no, we have to release this information as it's done, because it will enable other scientists, this is important. In genomics, in

research we have a concept of data sharing that enables that data sharing and value of the data, it allows interpretation of future data. Then, in 2000, president Clinton and Prime Minister Blair both supported the free access of genomic information. So again, they were really supporting that everybody should be able to view that genomic data. This is really important and quickly afterwards the US were very smart and they came round and said well, actually for that to happen we're going to ban genetic discrimination and so they came out with that quickly and they have a thing called GENA in the US that stops health companies accessing people's genetic data. So it was really important and the same, it also stops workplaces sort of taking your genetic data and making choices based on that. It's very important. Genomics in health care I've put this slide up first, this will tell you about Genomics England. It is there, it is happening right now and in Genomics England they're targeting sequencing 100,000 people, this whole genome approach from a variety of diseases, people in the NHS, patients undergoing diagnosis and at the moment they're seeking 66,000 patients. Genomics England is ahead of the game and then so they've agreed for the data access that all individuals data will not be released. That means their name, date of birth, all the identifiable elements won't be released. They did say that all the deidentified data, this is where the actual genome sequence data and bits of deidentified information about it, so that could be the person had cancer, the person did very well on treatment, that sort of thing. Then so all of this will be available in a secure monitored environment and then so how it works is then the doctors and nurses that treat the patients have access to everything, but then researchers can apply to access this data in the secure monitored environment to ask research questions and some questions might be really important. It might be, we think this gene is important for this disease, can we go looking in all of your patient data to confirm that? Because that means in the future when you see a person with this disease it will help potentially diagnose them, because they might have the same variant. Really important questions. This is 100,000 people, it's in the health system in the UK, in the NHS and this is the approach they've taken so researchers can access the data and do analysis. It's very forward-thinking and very important. I just mention quick when we talk about the data, what does that actually mean? For a person who's had their genome sequenced it generates about 180gig of data. So if you think of a laptop today, you fit about 3 to 4 people's genomes on a laptop. The data is big. I use

computers and code and write scripts to access and mine data. The volume of this data being stored 100,000 people times 180gig the volume of data is huge so it has to be in a nice safe environment. For ethical considerations, right now genomics happening in the health care, all this patient data. Patients are going under diagnosis as part of this work, treatments based on genomic findings and all the sequencing in the health care system is accredited. In this country we have a body called NATA, they would accredit their test so you're operating to a certain level and standard. That's good. Then we have a lot of genomic research that's happening, too. For example, our research that we do, we don't get the patients undergoing sequencing in the health care, they undergo sequencing in research laboratories and so in that sort of situation we get participant data. We understand the questions we're trying to ask is we're trying to understand their disease and how it arose in the first place, find new treatment targets and improve therapy and diagnosis. But what we're seeing happening with genomics, research and health care are sometimes far apart. In genomics they've really come together and a lot of the research happening is very translational. For example, I was part of a big pancreatic study. We were sequencing patients recruited on to that study and we were sequencing the tumours just like Aideen was talking about. For one of those tumours we found a mutilation pattern that indicated that the tumour had been exposed to ultra violet light. In the pancreas, that's impossible. We were able to go back to clinicians and say this is weird we don't think it's a pancreas tumour. Two years prior, the patient had had a small lesion on their neck removed and the patient was rediagnosed. What's happened is they've had a cell, had it removed. Over the two years that metastasized and moved to their pancreas. That changed their outlook. Pancreatic is not good, this is a little bit better. Big implication. That's an example of how the research is coming together and there's a lot of interplay and lot of my collaborations are with clinicians. We've got Australian Genomics which Kat mentioned briefly and also Queensland Genomics Health Alliance. Understanding this is happening, but we need to do it carefully and ethically and safe to implement genetics from research into the health care, because you don't want researchers taking on genomics in the health care. It has to be done in a health care situation. So then, this brings lots of questions that people might ask. These are if people are having their genome tested as part of their health, or if they enrolled in a research study where we might do genomic

sequencing patients might ask questions like, where's all the data going? Who's looking after the data? What happens to my sample? Is it safe? Who owns the data? That was your question. Who has access to the data? What will I find out? All those sort of questions. Then so all these need answering and this is what the QGHA stream is trying to look at, the ethical, social and legal implications. In terms of ownership of data, I'll answer your question now, it's a really contested question in a lot of different countries and it comes down to legislation in those countries. So genomics England has taken the firm stance that the patient owns the data. It's their genome and so for their participation in the big 100,000 project, they have a thing where they have to opt out of their data being used for research, but they're completely entitled to do so, because it's their data and they can choose who has access to that data. So the question was, what about when they pass away, who has ownership of that data then? Again, very highly contested. People don't really know still and different countries will come down and interpret different rules very differently. In theory, the data is linked to the family and so there is questions where should the children then take ownership of that data and who can see that data, because it has implications to them, as well. But it's a great question. Then this brings me to, I'm going to touch on a few ethics things that are used in genomics research. One is informed consent. This is a process where if any of you as consumers have undergone genetic testing you would have gone through a consenting process and told about the purpose, the medical implications that you're being tested for. Possible risks and benefits that may be there and then the possible implications to family members and other things that may be discussed are the privacy rights, where the DNA will be stored and who will have access to that information. All really important things. You may be asked additional things, do you want to be part of research studies? This is what all the research studies are doing, would you like to take part as well? Informed consent is important. I've put informed in brackets. It's difficult to give informed consent, how informed can you be about the complex issues around that genetic testing. That's one of the things that QGHA is looking at potentially is getting patient material ready together to try to help with that informed consent process and give more information. There's a concept called dynamic consent, which moves away from the traditional paper-based system of doing consent and it might be an on-line portal where you can then log in and have a look and get information specific to you that's there.

Because it's dynamic at any time you can opt in or out of certain research projects that may be there or allow researchers to access bits of your data or not and then it's up to you. So an important ethical issue especially in Queensland, I've got the map of Queensland up there, is equity of access. When genomics is being rolled out in the health system we need to make sure people in the south-east corner have equal access as people in the far top of Queensland and in remote areas of Queensland. That's an area we're particularly passionate about and has to be addressed during the whole system. Things like telehealth services and that dynamic portal is better, but it has to make sure we do that right to make sure everybody has equity of access. Secondary findings, this is where you're undergoing testing for disease A, but your genome data finds something related to disease B. What do you do? There having a test for breast cancer and you've found out you have a high risk of getting cardiac disease later in life. Think about the ethical issues of that. Some people think "I don't care about heart disease later in life, I'm undergoing breast cancer at the moment and that's it". The ethical implications have to be carefully considered. If we were to sequence every one in this room we would find 3-7 pathogenic variants. A variant heavily implicated with a disease. If you have 3-7 on average in every person, when you find an incidental finding, is it really important? Everybody has 3-7 in their genome. How do we interpret that? We don't have enough information as researchers to really understand that. We know it modifies risk, doesn't mean to say you're going to get cancer. You're going to get heart disease. We need to understand that more to measure risk. For breast cancer it's perfect if you have a pathogenic variant you can take preventative measures or go on a screening program so your cancer is detected earlier. That's some of the things that can be done. There are things that can't be done, we're not sure of the implications. The last one is important. As well as implications for yourself it's also implications for your children and for their children and so on, and so whether they get tested to see the same things or not, it's very important. Some people don't want to know if their mother was a carrier of BRACA1 they may not want testing. It's up to the individual. We mentioned genetic testing at an insurance or workplace level, anybody using your genetic information in a negative context. This may deter people from taking part in genetic research. I mentioned that the US quickly brought in an Act to prevent genetic discrimination. In Australia, we don't have such an Act and it's something to

be mindful of. There is a very active working group with a long acronym Australian genetic nondiscrimination working group and they've been active in lobbying up reasons why we need to have a nondiscrimination act and this should be a thing particularly if it's going into the health system. We don't want have a situation in the future where suddenly private companies want to access it and change people's health premiums. That's not a good thing to do at all and so this working group have been very active. They've been lobbying Parliament, they've had a parliamentary committee, made releases and so this is a hot topic and hopefully we'll see things come out soon. There's been now some recommendations that are out there to Parliament and this is just one of those recommendations, 9.1, you can see there's a lot, which is basically putting a moratorium on life insurers using genetic information unless the consumer want to provide genetic information to counteract anything. So it's really important and this is going to move very quickly in this country. And then I think this might be the last ethical implication I'm going to talk about, it's patient expectations, we need to be careful in modifying patient expectations. Personalised medicine is getting a flavour of the month and it should be, because it's going to impact the health system and patient outcomes. It will help us prevent diseases or detect diseases earlier. So it's really important, but right now at the moment a lot of the tests come back with an uncertain result and so that means the patient isn't going to get the cancer they want sometimes. We need to be careful, we talk about personalised medicine a lot, I'm in favour, but we need to make sure for some people there will be uncertainty about what's driving their disease. Because we have all of this data and the more data we have and the more people we sequence, the more information we get out of that data, because we now can see rare diseases happen a little bit more and that helps us diagnose the next person with that rare disease. This is really important, but it also means we can go back and re-analyse people's data at any point in time and they may undergo rediagnosis really quickly and that's because the data is forming. People who didn't think they had a risk in breast cancer in the future they may find out they have a risk, so there is a rediagnosis that people undergo. Sometimes the test may give you a genetic diagnosis, but there is no treatment. It's important to remember that I think a diagnosis of a disease is sometimes what the patients are looking for and it's really important, because they can form a network and find other people with the same sort of diseases and just because there is no treatment,

that means now we know the underlying genetic cause researchers can look for a treatment. That's important to understand as well. From a patient being diagnosed sometimes there will be no treatment available. So this is questions that I've put up here for you to think about. Should consent for research be opt out as it is in Genetics England whereby people undergo genetic tests in the hospital and all of their data is used for research and they have to go on-line and opt out of that situation? Should patients and participants be made aware of incidental findings? This is very topical. Some people say you have to tell the patient everything. Some people say "No, no, we didn't do anything about it or if we don't know it's definitely going to be definitely associated with risk so we shouldn't be telling patients". Have a think about yourself, how would you feel about your genome data being shared? If you were sequenced would you want researchers like myself accessing that, companies accessing that or would you like other countries accessing that? Lots of questions there. Another very topical thing, should we sequence all new borns? This comes up a lot in the genomics community. Right now a child born in Queensland or in Australia they'd get a heel prick, a piece of blood taken from the heel and it gets rubbed on to a little card and that card is used to undergo some testing for a few conditions. But should that be flipped around now that genomic sequencing is getting so cheap and sequence every new born and the data we get out of that is self-informing and very big. In 50 years' time you can see what diseases they ended up with and form preventative measures in the future, because you can get a better estimation of how likely it is people are going to get diseases in the future for the next generation. It's a real topical question that a lot of people are talking about at the moment and that's it. If you have any other questions I'm happy to answer them. Microphone is coming.

>>: Thanks, I'm Janelle from Brisbane. I'm just wondering, with the insurance issue, let's say person A is completely healthy, their genes have come back "hey, you've got great genes", they let the health insurer know so they're getting a huge discount. Person B comes along with dodgy genes, they're paying the higher premium regardless of the fact that the insurance company doesn't have all the data.

>>: I didn't quite hear, but it's an insurance question.

>>: So it's person A is getting a discount, because they've got good genes and shared it with the insurance company, so the insurance company is offering a huge discount and person B isn't sharing, because they know they've got dodgy genes, so person B regardless is paying extra money, because they're not sharing information.

>>: No, I don't think that situation would happen. Right now if you've had a genetic test even for a research testing, then an insurance company may come along with say "I want to have access to that data" we don't know of any cases that's happened, but they may. The reason why that is there, that's one of the recommendations I pulled out. The reason why that's there is because if an insurance company thought hang on you've got a genetic test there must be something wrong that's an opportunity for person A to say "No, there was nothing wrong". So it's not a way of saying I want cheap insurance premiums because I have no variants. Everybody has 3-5 pathogenic variants. No one has the perfect genome, we're all a little bit odd and we don't know enough about the genome to understand risk. For some diseases we can get an accurate prediction of risk based on a few variants, but the entire genome, insurance companies don't have an idea, that's why it shouldn't be used in that context.

>>: Yeah, this is a bit out of the box, but it kind of pulling it all together that if in the future when virtually everybody around the world is having it done, going to the 6 degrees of separation you'll find out that certain countries and origins are prone to certain illnesses. Japanese have problems with kidneys, they have a lot of kidney disease... don't quote me, but that way you could sort of look at that and do a mass cure or research.

>>: That's all around thinking out of the box and looking at populations. You're right, certain populations are more predisposed to certain conditions. Some of that is environment and that's what we can't pull apart. In some countries they're getting a high-fat diet and getting things like oesophageal cancer and obesity that wasn't there before, so there is environment context, as well. You can

definitely find some genetic things causing some diseases and they're more enriched in certain populations than others. Yes, that's right. Thurrock while we're out of the box, my question is around the future implications of artificial intelligence and the mass data and both the storage and importantly, the other side of the question for me is what are the safeguards in terms of corruption prevention? There's obviously money somewhere tied up in the big picture, whereas in other parts of society we have red flags for critical areas which are available for organised and other crime to move in on. Is there thought... I imagine there must be thought in that direction, because you are already storing data?

>>: Yeah, so they're really good questions. In terms of artificial intelligence we're actually now looking at artificial intelligence to mine the genomic data and do that interpretation of the data to figure out risk and potential disease causing variants. As a researcher you can do it one by one but not en masse through the whole genome. We're looking at AI at the moment definitely. A lot of people are and it's a big area. In terms of storage of all the data, I think that was the second bit, so the storage of data is very problematic and so at our institute we have a great big data storage on our fifth floor and we keep that off-site and archived in a safe environment, but that's on local hardware our institute purchases and it's held a lot of the research institute's work, but it's not sustainable, because we can't afford to keep buying hardware every three years and we can't renew that. Cloud implementation using Amazon and Google that's where it will go in the future. I can say comfortably that's where the health system will go too and that's a safe environment. It's got a bad rap sometimes. People don't know how to use it properly. It's much more safer than having it on my local computer or in a building next to me. It's very safe if you use it correctly to the point Queensland Police Services now have all their information on the Cloud. The CIA all the information on the cloud, other health system, the UK, all their information is on the Cloud. You have to do it in a manner you're using it correctly and using the right recommendations. It's encrypted on disc, when it moves. You have to use it right. With it being on the Cloud that means you can have other people access the data if they're allowed to. Researchers could go and access the data. I know you've got another question.

>>: Just recently there was a cold case in the US solved by genetic testing. I think the police were able to get access to some data from say 23 and Me and found someone who had a genetic test done and was linked to the perpetrator who they were able to track down and there was a furore around "I didn't know that's how my data could be used". Do you think that could happen in Australia? Secondly, how far does the genetic link go? Is it just immediate family, or is it my second cousin's cousin? How far does that go?

>>: How far does genetic information go? Right now there's a company called Ancestry and Me, they're common and advertised on TV. You give them saliva and they tell you where your genetics are from. They can trace through journeys from Asia to Europe to Africa. How far it goes, all the way back, so very far and also, this might scare people a little bit, just think about this. If we had a genetic sequence of somebody, we would never do this, but just to show you how far it goes you could then take the variants in that sequence go looking in all the publicly available sequencing and find family members. It has potential to go very far back. The first bit of the question was?

>>: Just about services like, say, the police force or others having access to this information as well not just in health care?

>>: Having others access that information, that all comes down to the legalities and the legal things put in place to deny people to have access. In a situation where it was the police that needed access to that data that would be an extreme case. They would have had to get explicit permission of reasons why. There that would happen, some people will say it shouldn't have, we'll say it should because it came to a good outcome. So long as the ethical and the legal barriers are there to prevent anybody accessing it and you have to do it through a safe means, then yes. One more question.

>>: Hi, I'm Jill from Canberra. My question is about health literacy. There's a lot of consumers out there who are probably taking one look at this and going we're less interested in the magic of how the sequencing is done and how the magic of the science is done and more interested in the ethics, the legal and the

social implication. I guess my question is more about advice from you. How would you talk to a consumer and what are the big issues that consumers are having and how do you market it or make it comfortable for consumers to know, so they know, they can ask the right questions?

>>: It's a really good question and there's a lot of information out there if you Google, in this space. There are really good resources to use, though. The genetics England as do Australian Genomics, as well. It's more about just communication, just finding people to talk to, finding useful contacts and talking to your treating clinical geneticists and genetic counsellors, they're the people that know things, so yes, that would be my advice. Thank you. (APPLAUSE).

>>: Thanks so much, Nic. Now I would like to invite Gary to the stage. I've got a blush to read about him. Gary is based in Bundaberg and his interest in genomics is personal after son Dallas who's now 9 was diagnosed with a rare genetic condition. Dallas has the only known case of Van Maldergem Syndrome 2 and they have formed a support group for the handful of families dealing with this diagnosis. He is a member of the patient travel subsidy scheme steering committee within Health Consumers Queensland. Thank you, Gary. So Gary and I are going to have a chat. You have a quick anecdote about an early memory of the health system?

>>: My first sort of memory of the health system was when my wife was pregnant and our bub was due and in the heat of the moment sort of thing I left my wife at home 40 kilometres away from the hospital and in a panic to get her to there. Then saw my senses and went back to the hospital, back to the house, picked her up and went in at 3 o'clock in the morning and yeah, so, that was my first introduction to the health system. Now, we sort of live it every week.

>>: Excellent, I'm sure you're not the only father has driven off to hospital without the pregnant woman. Can you tell us about the first time that you and Charmaine saw evidence of Dallas's health condition?

>>: With Dallas, he was 14 months and 4 days and we went in to check on him early in the morning and he was completely paralysed. There was no life, no movement, no nothing. His eyes were flickering, but it was like he had a stroke. So natural reaction we went straight to the emergency department and panic stations and still got no answers. There was a lot of issues where we were just faced with "we don't know, it's out of our scope, we don't know". We'll send you here, we'll send you there and we ended up coming to Brisbane and still didn't know anything.

>>: How old was he when you ran out of testing options?

>>: Dallas was 6 when we ran out of testing options within Queensland Health. So that's when we saw the little story on 'Australian Story' about little Mossimo and that family enlightened us to pursue genetic testing and as soon as we started progressing with that, that's when we found a lot of barriers of no information. No one knew about genetic testing. The costings were astronomical, $7,000 per person we were quoted, $1800 per blood test. So it ran into the thousands and of course, we couldn't facilitate that so we had to look further for research grants.

>>: How old was he when he was eventually diagnosed?

>>: The diagnosis time it took two years, so he was 8 when we finally got a diagnosis. He's been diagnosed now for two years. He's still the only one documented in Australia. Our second youngest son has been tested through the luck of genomics and he has the exact same mutilation. It's not documented on the Queensland Health file yet, so storing of data has benefited us. It took us a week to get the results back instead of 12 months.

>>: What treatment does Dallas need for his condition?

>>: Dallas uses sort of wide spectrum treatment. He needs speech, he needs OT, he needs physio, gastro, neurology, spinal, orthopaediatrics. There's a whole raft of it that he needs and since diagnosis of getting the diagnosis, we've now instead of going down every 3-4 weeks from Bundaberg to Brisbane, we're back to

12-monthly now.

>>: A lot of people who are carers of children with rare diseases, there's this term used called diagnostic odyssey, so I guess your diagnostic odyssey was close to 7 years long?

>>: Yes. We were undiagnosed for nearly 7 years, so we were just living in limbo. We call it the guinea pig stage, because everyone wanted to poke and prod and have a chop at him to see if they could work a monarchist Kell cure. Since then now that we've got the label so to speak, it might be a diagnosis on medical terms, but it's just a label. It does nothing to cure him or anything, it's just a label.

>>: So on that, there's not currently a cure for Dallas's condition, but there obviously are benefits that you can identify from getting the test done. Can you talk a little bit about that?

>>: The benefits we found is like I said 12-monthly appointments instead of every couple of weeks and the relationship that we've built with our physicians. So now we no longer go to a doctor's appointment and sit there uneducated. We, in turn, are pretty much partners. So our paediatrician and us, we talk outside of business hours now. Our metabolic specialist in Lady Cilento we talk well and truly outside of business hours. A lot of it is done just by email or a telephone call. We actually don't have to present to the hospitals anymore. Then, they've actually partnered with us and we're now pursuing international research grants. So it's not just us learning, it's us learning together with our doctors which is really beneficial.

>>: And how do you think that spirit of partnership came about with your specialists?

>>: A lot of head butts and a lot of disagreements, but you've got to be open and honest. That's one thing that anyone going to a doctor has to be is very open, very honest. If you disagree, speak up and then also, do the research. Help your doctors help you. So if you've got a rare condition, go home, play with Google. Don't listen to what Google says, because otherwise you might have a sore toe and

you have to amputate your leg. But just go and do some research and then take those findings back to your physician and you'll find that you will work better as a partnership than just relying on your doctor to do all the work for you.

>>: Did that come naturally to you and Charmain, just that ability to be persistent and push your case? Was that something that you two, is that how you're like anyway?

>>: It took a little bit of learning. We were pretty easy-going at the beginning, just sort of going with the flow, not asking questions. But once you become sort of the advocate for a child, that gives you the power to pretty much say no and ask lots of questions and if you don't agree with something it's easy just to pick that child up and walk to another facility and there's not much people can do. But now we have the best relationship with all our specialists, so.

>>: I want you to talk a bit about how you've connected with other people who are in a similar situation with you. I know you've set up a few different ways to do that.

>>: Our first initial sort of reaching out was my wife started special needs kids Australia Facebook page. Now that's got, I think it's roughly 9,000 members now, so we communicate with lots of people daily. But then once we got the label of the syndrome we started searching for other people. There was no way of finding anyone that had been previously diagnosed, so we went to Facebook and started putting up posts on international sites and we were lucky enough to actually connect with the whole 18 that's been diagnosed in the world. So now we have a collaborate of people on a Facebook group that communicate daily, share medical history, share stories and photos and collaborate with each other's specialists. Now it's not just an Australian-based thing, this is worldwide.

>>: Excellent, you've been really proactive, Gary. So what's now? What's next for Dallas?

>>: Well, next for Dallas is just ride out the journey, but I want to know that when Dallas gets older we can look him square in the eyes and we can say we fought, we did every single thing in our power to make sure that everything was achieved for you and we fought to make sure that you were appropriately managed and if by some chance he may talk in the future, he may walk. No one knows, no one can predict what's going to happen. If by some miracle he does able to do independent living we can sit there and say we did everything in our power to make sure that you are the most comfortable in life.

>>: Thank you, Gary. Can you talk a bit about anything that you're particularly grateful for relating to your family and Dallas's journey within the health system?

>>: Oh, I'm just grateful for the humour and the partnership with our specialists. When you go to an appointment with us, there's none of this us and them mentality. It's two people, friends, laughing, joking. It's probably classified as unethical in some aspects. But I really don't care about the ethical atmosphere. I like to know that the doctor is on the same page as us and our paediatrician is fabulous. If she sees us up the main street and we're doing the wrong thing, she'll come up and she'll politely clip me under the ear and say "You shouldn't be doing that". That relationship is fabulous. We walk into a hospital now, we're not scared. We know what we want, we know how to help and we know who we're talking to. So it's been fabulous.

>>: And I think it's important to note, though, it wasn't something that anybody handed to you both. I'm looking at Charmain here and I think it was a funny story she was telling me earlier when she requested Dallas's medical records and there were all sorts of remarks in there about her behaviour and labels put on her really just for being such a strong advocate for your son. That's a lesson for all of us. I don't know that kind of ability to advocate for ourselves and our family comes easily to a lot of people and I'm sure it was difficult. But I think obviously the dividends have paid off.

>>: It was a long road and it's still going to be a long road. We've still got a hospital transition, so to speak. So from child to adult. I'm not looking forward to that. That's a whole different range of problems, but at this present point in time we're comfortable, Dallas is happy. He does everything he should do and we get to enjoy family life. So as long as he's comfortable.

>>: I'm sure with your experience you'll whip the adult clinicians into shape, as well. I'd really like to open to the floor to see if there's any questions at all of Gary.

>>: Gary, firstly thank you. I wanted to applaud multiple times during that, but didn't want to interrupt you. Obviously it's been a really long journey. Has there been any moments where the health system was particularly difficult? Things like "Hey, I'd never want that to happen again", things we can learn. Anything that we can take away?

>>: Our most difficult time would be when our son contracted scurvy. There was lack of information. Another experience of doctors not knowing. Our son was turned away from the ED multiple times, because they just said "Oh, it's his hip dysplacia, it's nothing". We as parents knew it was much more. It got to the stage where Dallas was, he was in the final stages of scurvy and paralysis had set in. He was bleeding from all over and no one would listen, so that was the hardest part. The fact that going to an emergency department, no one listening and then being treated as if you don't know what's going on and then having to fight while your son is still in that condition and still piece it together. That's the hardest. It's not hard for a nurse or a doctor to come in and say "My name is..." and sit there and talk to you. It's pretty simple, but sometimes that doesn't happen and I understand the stresses of nurses and doctors, but parents in that sort of aspect, they really don't care about the other thing. They're focused on their one child and that's it.

>>: Thank you.

>>: Any other questions from the floor? Here we go.

>>: I was just wondering, my heart just goes out to you and your wife. How do you manage? Do you get a lot of support in your community? Do you still live in Bundaberg?

>>: Yes, we're still in Bundaberg.

>>: Do you and your wife get out to go anywhere with your child or just the two of you? I think there's so much you've had to do and be in all these years, how do you survive?

>>: This is our first official time away from no kids, because we've got four kids.

>>: Party time!

>>: Dallas is our youngest child, so this is our first time in probably 14 years that we've actually had time away from our kids. We've always done it on our own, never asked for help, but yeah, so it's just something that comes natural.

>>: In relation to those clinical notes, I'm assuming some of the comments weren't on the happy side of things.

>>: Weren't the most complimentary- are you seeking details?

>>: I am not seeking details, but when we're thinking of person-centred care and advocacy, I come from a mental health background and recovery-orientated language and I'm a consumer, that had to be shit to read that they're saying that stuff about you guys, too. That's not cool.

>>: Obtaining the health records in the first place was hard, because you have to go through the Freedom of Information and then there was the whole other hurdle of us trying to get enough information to be able to send overseas and then we get the encrypted CDs that we can't print, we can't do anything. Then we start going through it and we were lucky enough to obtain the doctor's notes and to

find out some of the snide remarks that actually get written on to your file, especially pertaining to my bossy wife.

>>: That's a compliment.

>>: Yeah, it was hard to sort of read and you've got the name of the physician right there, so that next visit when you go back and you approach that doctor, it's very hard not to sort of really lose it and be unpolite, but we've managed to sometimes hold our tongue. It hasn't been a real smooth journey, but now they know where they stand and where we stand and now it's very open. Our doctor's appointments are nothing like what you'd see on TV. They're quite relaxed and sometimes we have them done out in the middle of a park. We are probably the pinnacle of hospital experiences now, because our son doesn't do waiting, and if he gets upset, we get upset, so.

>>: Thank you, and I think there was one more question just behind you.

>>: Hi, I didn't actually have a question, I just wanted to say thank you for coming to share your story with all of us here and it's been incredibly powerful and you have fought courageously for your son. You and your wife should be so proud. (APPLAUSE)

>>: Thank you.

>>: Thanks so much, Gary. So we were just extremely fortunate to have Gary involved in our community group. He and Charmain's contribution as genomics consumers, but also dealing with that rural and regional issue is really important for us to know how to deal with that. Now I'd like to invite David to the stage, the Queensland director of Queensland Genomics Health Alliance. He's just come from another meeting and has been giving us all heart palpatations he might not make it, but here he is. Just a reminder, we're starting by sharing a short anecdote, an early memory of the health care system and how you felt about it.

>>: Okay, so I had a few experiences as a kid, but I would say probably the birth of my first child, so my eldest is 23 this year and we were in Redcliffe Hospital and my wife had gone into labour and we went and they sent us back et cetera, et cetera. We finally got in and they admitted her and by this stage she'd been at it for days and she was exhausted and we had no real idea of what was going on. We were both in our early 20s so we barely had control of our own lives let alone bringing someone else into the world. We got into a situation where they asked us to sort of spend the night and to give Wendy some time to recoop and the next morning a new doctor comes on, a new shift comes on and they did a prick the skull, test the blood level and he was stuck with the umbilical cord around his throat. You go from days of waiting and not knowing what's going on and understanding anything and trying to support your wife and deal with the clinicians and then all of a sudden it's "Sign here, we're doing an emergency caesarian" and all of these sorts of things. It was not what we expected that was an early indication of an anecdote from me. Thanks very much for making time. I haven't been here today, I've been out and about doing things, but I would like to send my thanks to Health Consumers Queensland for the forum and to everyone who's participated today in the forum. We think it's really important and we're very pleased to have presented this session around genomics to not only talk about what our program's about, but to showcase some of the people, clinicians and researchers and consumers, parents and carers who are involved in the program and what this means. So I'm going to just do a few slides. You've hopefully heard already what the genomics program is about and you understand the investment the Queensland Government is making in its future and in the citizens of Queensland. So the $25 million program is there for the duration of the course, so I don't have to go backwards and forwards. It was an interesting journey getting the money in the first place. So I guess my job, I'm the executive director, so I'm basically responsible for seeing the mission of the organisation come to fruition, but really what I wanted to do today is talk a bit about what it really means to activate community engagement and people's engagement in the program. I guess I've been working for a long time now and I've worked in health for the last more than 10 years and I've been involved in programs of significance that have been about how you really contemplate patient-centric services and clinically-led programs and it's something I bring a fair bit of experience to, but at the same time

I don't want that idea of community engagement just to be a sort of tick the box. So we're very serious about how we activate the community voice and why that's important. How does the organisation go do its work? We invest in clinical projects in particular areas. We try to match all of this amazing translatable research in genomics that's going on around the world and focus that on what the needs in Queensland is, where can the progress make impact? Genomics is so pervasive across health care. Everything from the legals and the social implications to the cost of services to an emerging market for sequencing technology based on disruptive technologies and so you really get to address all parts of the health system. But at the same time it's about being able to show the success of how genomics can be applied and that's how our investment scenario works with the clinical projects. At the same time that you do that clinical activity you also have to be thinking about the infrastructure within the system. Infrastructure obviously means things like where is the data being stored and managed and who has access to the information, those kind of very hard aspects of infrastructure. But it's also about having the right policy and legislative protections in place and you will have heard from Nic and Aideen and others who have spoken about the work we're doing in that part of the genomics world. But it's really this last point that I really want to make sure that everyone hears today is about how we engage community. So we were tasked with creating a community representative advisory group. It was written into the business plan, the rules and approved by the Director-General and the Minister for Health. But how do you go about doing that? There's a low bar and a high bar. Low bar is get the thing done, but we always try and achieve the high bar. We understand what it would really mean to be successful. So how do you get a group together? There's not a lot of experts from a community point of view in genomics. It's a rapidly emerging area of science. There's not a lot of expertise in the clinicians either so we really looked at the sort of individuals that could help advocate for the various groups. So whether it's Aboriginal and Torres Strait Islanders, whether it's regional and remote, different groups from community ethnic backgrounds or different languages and we were lucky to work with Health Consumers Queensland to populate some of those roles on our community group. But it's the networks those people connect to. You've heard Gary talk about the Facebook and the community work that he and his wife have done in Bundaberg. That's really the power that we're trying to bring to bear here. It's that network of

network effect and we're trying to get that awareness out and about genomics, but at the same time make sure there's a voice for consumers and patients in the way we make the selection around the clinical projects and we contemplate the issues that come when you build digital infrastructure. That's really how we're going about it. The next thing to talk about... I've sort of covered this, but we have this idea of what we call a quadruple bottom line which is a business buzz word, which means everything we do, every investment we make we consider the core objectives for the genomics alliance, but we think about what is the benefit for patients, what's the benefit for clinicians, what's the benefit for the health system itself? We have an increasing population and an ever increasing cost to deliver health care the way we know it and think it should run in our country and also for research. Not as an afterthought, but actually we need to have the research community engaged in the process so we get that virtuous cycle between health delivery and research and they start informing each other. It is an unfortunate aspect of our system that they are different systems and they've got very different measures and ways of doing work. So the program's about a collaboration between the health and hospital systems and the research system. We try to make sure all of those factors are considered when we contemplate our investment. I'll go back. I wanted to make a point about how genomics fits into precision medicine. I started working in digital health it must be more than a decade ago and I would often tell my team when I'd go to E-Health conferences and there'd be a dozen people sitting around patting each other on the back and saying, " No one understands this stuff" and last week E-Health Queensland had their expo and they have nearly 2,000 people there and it's a strange thing to see that transition over time and I'm here to say really that genomics within this idea of precision medicine is on that journey and in years to come you'll reflect on this conversation and what you've heard today. But it will just be part of the way the health system works and that's core to what we're trying to achieve is to make it part of the way the health system works, not trying to do something as an aside. Personalised medicine, it's about a tailored approach to health care, it's not about the one size fits all that we have today and so this means if we understand, because we can access the data in a clinically safe and a privacy-aware way to understand the information about you and how other people who've got that issue or been given that treatment how that's working, but also about you as an individual and as Nic was saying about the

impact of the genome which is this kind of molecular ERA that we're living in and that we can get into very precise levels of care where we go it's not just a person who's got lung cancer, we can look at the difference genomically and genetically between you and that tumour which is basically something has gone wrong in the way cells are reproduced and it's created a tumour. We can look at the difference and think about what targeted interventions like drugs and different therapies would work as well. I'm not a scientist, but a little fact about things like codeine, most people know about panna dean and there's codeine, about 6 per cent of people in the population can't activate codeine, so it doesn't work. What does that mean? People who have chronic pain and they're being treated with things like codeine and they're saying "it's not working" and clinicians will sometimes go well that person's got a drug problem, or something along those lines- this is a really negative example- but that ideal that we know genetically that some people can't process panna dean and the codeine doesn't work. I have an allergic reaction to codeine if I take it. I've learnt that the hard way, but gee it would be nice if that information was stored in my adverse reactions in my medication history in my electronic medical record, but also that rather than having to find that out in a particularly painful and hospital-based way that you find that out and it means you're forewarned and clinicians are able to contemplate that when they're dealing with ewe yo. That's the precision medicine journey we're on. The last bit of this is something Katrina asked me to share and this isn't something that I normally do. I've been working for a number of decades now and I've worked in lots of different industries and I always say I could make more money making banks more money, but I really got into the health system working in the health system about 2016-7 and I'd had a particular scenario going on in my family that meant that the way the health system worked became sort of very much a problem for me and my family, so I really dedicated my time and energy professionally to working in health, because I could see the opportunity to improve it if we could connect the information together and now with genomics if we start treating people as individuals rather than with a herd mentality. So Katrina asked me to share a little bit about my story. My daughter Daelle is 19 and Daelle has cerebral palsy. The underlying genetic condition is her cerebral cortex for her brain didn't form properly and it's a rare genetic condition, a 1 in 10 million sort of thing. You guys have the jump on us there, but basically when Daelle was born everything was

normal. We got to 5 or 6 months and she wasn't developing properly and so she wasn't hitting milestones and we went through that diagnostics odyssey with everyone poking her and prodding her and trying to work out what was going on and we eventually got the diagnosis, but this is nearly 20 years ago. So it meant a lot to have that diagnosis, to understand the label. It didn't really make any difference in terms of the treatment. Although what we were going to get, it's a complete mystery. What's going to happen, and you sit down with the paediatric neurologist who tells you this is going to happen. I'm a tetchy by background so I go "If she's physically disabled augmentative technology and all this cool stuff will come and her life will be normal she'll just be in a chair", but yeah, no, we have severe mental disability, as well. So Daelle's like a ... it's hard to put a year on it, like a year and a half, 2-year-old if that, no words. She can't feed herself, medically incontinent, needs 100 per cent care. The most diabolical part is Daelle's epilepsy. Not treatable with medication, so she falls into that percentage of people with epilepsy who are drug-resistant. I've spent probably the last four or more years volunteering and working with Epilepsy Queensland. I'm the chair of the board, as well. I wear multiple hats is why I'm telling you this story, so I think in our program every day about how we can make the health care system better and I think about groups that advocate and try and raise awareness around different medical issues and conditions and we see that there's this opportunity to partner with community in our program to make sure that we can help those voices be accelerated and also be part of the journey you're all on. A bit like Gary's story, lots of medical intervention her right hip restructured, a couple of surgeons going at it for eight or nine hours. As the guy said to me, this is about the most you do to someone unless you're trying to save their life after a car accident and then six months in sort of this position here. So it's been a journey and the journey goes on. Daelle has reached the average life expectancy for someone with this condition and we shoulder on. She's doing it worse, because she's picked up the flu as well and that means her seizures get worse and the dramas in the family. She has a type of seizure where he schemes. She schemed for an hour and my wife rang me in tears. It's not a sob story. Everybody in health has a story. I've not met anyone in my journey in the health system working with people and professionals trying to improve it where someone didn't have a story about a child, or a parent or a sibling or a cousin and genomics is going to be one of those things over time where you'll know oh that

person has that condition and that's why it runs in the family. It's about that, being able to balance that idea of being a parent, because I'm Daelle's dad and that will never change. I'm her parent and project manager when I deal with the health system, so I'm an advocate. So you wear all of those hats and I'm lucky, I will say to you to be able to wear another hat where the government trusts my team and me with $25 million to try and bring about this evolution and revolution in health care and ease it into the system and we're very keen to have community's voice in there, because it's critical. Thank you. (APPLAUSE)

>>: Thanks so much for agreeing to share your personal story Dave. I really appreciate it. I'm sorry we're running short on time, so we don't have time for questions, but I would like to invite Erin to the stage to wrap it up and talk about work that the community advisory group is doing.

>>: Thanks for sticking around even though we're running a little bit late everybody. I heard a couple of questions about the advisory group and the importance of consumers so that's the main focus that I'd like to really talk to you about today. But apparently I need to tell my own story, and I know this story really well, but when Katrina asked it to me I suddenly remembered it this morning and it was just like a lightning bolt, because I realised some of the things that really drives me to do some of the work that I do with consumers. I'm also on the board of Health Consumers Queensland and that's an amazing opportunity. But I was about two years old and I'd broken my leg and I was getting my cast removed and I could feel that the doctor was cutting my leg and I told the doctor. I was quite good at talking by then. I'm still quite good at talking, I like talking. I was starting to cry and I was really raising my voice and the doctor started raising his voice telling my mother to shut me up and to control your child and "I'm not cutting her, she just can't understand" and then the cast came off and there was blood everywhere and I don't remember him comforting my mother or me or anything else. The way I felt was that I didn't really have a lot of trust for a doctor if they don't listen to the consumer and that's what I've continued to do and I think I'm in good company here with all the people here and it's great what you're all doing. I think it really is making a difference. You've heard a little bit about QGHA and the work that they're doing and I'd like to say that it really has been an amazing, it's a wonderful

partnership with the business team, with Dave, with Katrina and others at QGHA. We started as a community advisory group just at the end of last year so we're still relatively new in terms of being a community advisory group. We really tried to get our feet on the ground very quickly with doing things. Most of us who are from the consumer side of things we have all come through Health Consumers Queensland, and I'll show you a picture of us. There's a 5-year remit in terms of our community advisory group, a 1-year appointment and it's really the starting, putting down the tracks for this community advisory group within QGHA which is a new area for health. So we're meeting quarterly although we're quite enthusiastic, so we're doing things offline as well. One of the main things we focused on straight away the beginning of this year it was our second meeting. We jumped in and got into some strategic planning, because there is just so much that needs to be done in this area and we want to do as many of the right things that we can as possible and so we had a great opportunity of having a really sort of big planning day, of thinking about all different things. Thinking about all our different stakeholders, what's our focus? Because our focus at least in the beginning is to really lay down the foundations. We're the first community advisory group within genomics health advisory and we know that whatever we do regardless of who comes after us, we will be some kind of reference point for them. So we really wanted to be able to lay that down well and some of the things that really came forward for us was this idea as, not just a collaborator, but we wanted to put active collaborator at the beginning to really enforce the fact that we wanted to collaborate not just with QGHA, but the researchers, with people in the health system because this will be a long-term change and Dave mentioned just before that although it can personalise medicine can become a revolution for the health system, it's not all going to happen overnight. It's going to happen in a much more evolutionary kind of way and, therefore, we need to be really thinking through things. Thinking how we can influence and make sure that we keep the patient in the centre. Personalised medicine by its nature should have the person in the centre, however, we can't just rely on the fact that it's called "personalised medicine" and that people will be in the centre and that's really the focus of what we're trying to do, and you can see that just in the fact of hearing Gary's story or Dave's story. The long odyssey that people face when they're trying to get a diagnosis and a diagnosis that doesn't necessarily lead to a different treatment outcome. However, it does mean

something to the families. Before I get too far away, I just wanted to show our wonderful smiling faces. This was taken when we were doing some of our planning. As mentioned it's quite a diverse team and the diversity in the team is really great, because we have Nic and Aideen who come from a professional background within genetics and research and genetic counselling. We've got another genetic counsellor there who's come from Queensland Genetic Health Services and so in that sense as consumers or consumer advocates or carers we've got the resources there, or the networks there that we can tap into to really kind of expand the impact, expand the knowledge base that we're working from. Our vision there, that we're there as an active collaborator with the health system, with researchers, with clinicians and communities to facilitate equitable and accessible person-centred care for all Queenslanders. Because the thing around genomics is that it affects everyone. Everyone has a genetic code that makes them human and yet, most of us are not necessarily ever going to access, go in and get genetic testing for a condition right now at the moment and yet it has the potential to revolutionise the health system and impact all Queenslanders. So it's kind of this, yeah, a bit of a dichotomy at least at the moment. In terms of the focus of our objectives, we really wanted to see this idea of the nuanced, the long-term pathway that consumers often go on in terms of getting diagnosis and then the treatment and the way that we as an advisory group that we would be able to help to influence the health system in that way going right back to the researchers that might be so far away from the patients and the clinical health system that things kind of get a bit blurry for them. Just from mere time of where things are at and we came up with a number of themes of work. If you really love this strategy by the way and you want to do some more reading about it, there's copies of it out on the QGHA table. There's some nice glossy copies of it. Collaboration comes up again, because if we're going to be able to make change, we really need to be able to work with people. It's going to be the diversity of views and the fact that we're going to have to work together to change the health system. Equity and access, in a State like Queensland for something as specialised as genetics services and genomics, it even puts a greater focus on equity and access aspects because these specialised services are more likely to be focused down here in the south-east and yet, if we are going to make the transition in the health system we need to be thinking beyond what's our day-to-day. It's great to have members like Gary come in and

really help to refocus us. Just in terms of getting literature out there Gary saying there's no point just putting everything on a website, because people in remote areas may not be able to get proper access to Internet services to be doing lots of research there. We need to also think about what is not so much our day-to-day. The education approach, it's at all levels, where we're going to go through a process over the next years of quite a lot of differences in terms of who understands what, that the patient or the carer or the consumer may be more informed than the clinician, the GP that they go and see. So how the education of how we navigate the system, the navigation about the actual technical aspects is also really important. Educating researchers so that they understand the nature of the consumer as well and engage with them in the design of their projects, that they think with the end in mind rather than just think to the boundary of their own research. We like to think of ourselves as advocates, as well. I think advocacy really needed to stay there in the centre, because we need to be not just coming up with nice plans, but we need to know that we can take those somewhere, as well and that requires an advocacy and being able to prioritise as well. Laying down that foundation, but knowing where to prioritise and where to direct our action because we could soon see ourselves at the end of our term having gotten nowhere if we don't put a good priorisation in place. The last one was a real world context and that was to really bring it back to the lived experience. Make it practical. Come back to the power of patient stories and lived experience and so that it will actually make a difference to people's lives when we're working and that can be building up shared understandings in a whole lot of different ways. And so some of the areas that we're looking at, for example, were mentioned but areas such as dynamic consent, the insurance, the literacy and the education and one of the areas that we most saw as a sort of a central issue that we wanted to ourselves get involved with was having a patient registry that was patient-driven as well so that people could understand who's who around the place and that would really help with the diagnostic kinds of outcomes. As I said, we're aware that there's a lot of challenges in this area. There's challenges in terms of the different kinds of community that we can represent and how they will understand and relate to these areas and just the long-term nature of the change in the health system. Disruption is good. It means that things shake up a bit, people get uncertain and we're trying to provide some of that certainty and make sure that the consumer, the carer stays in the

centre of the conversation and is actively involved in the design and delivery of a personalised medicine and I would love if you are interested that you engage with us, as well. Just because we're a consumer advisory group from my point of view, we need to also be out ward-looking in terms of who we engage with. Thank you. (APPLAUSE)

>>: Thank you so much, Erin. I'm worried that we've run out of time, but Erin is going to be around at the forum for the next two days, so if you've got any questions about how our community group works, or advice on developing your own strategy for your community group, feel free to come and see Erin over the next two days. So that's that bit. Finally, just three things I want to finish up with briefly. Why are we telling you all this? This is just so you can be informed so that when you're engaging with your communities you understand that there are big changes to come within the health care system. The next thing I want to say is thank you to all members of the community that are here today and a special thanks to Heather Renton who's involved in the Melbourne Genomics Community group and she's come here for the next two days. That's really wonderful and finally, I realised I'm standing between you and drinks, thank you so much for your time and coming along and your attention. We really hope you got something out of attending today, so thank you.

>>: Thank you, we'd love your feedback. (APPLAUSE).