



AUSTRALIAN PATIENTS' AND FAMILIES' PERSPECTIVES ON GENOME SEQUENCING

REPORT AND PATIENT CHARTER



NOVEMBER 2016



Genetic Alliance Australia Ltd

Genetic Alliance Australia (GA), a tax-deductible registered charity, was formed in 1988 to provide peer support and information for individuals and families affected by genetic conditions. Genetic Alliance Australia is a peak body for rare genetic conditions for which there is no support group. GA has 181 members, 70 of which are support organisations, and has over 3500 individuals and families on a contact database. Genetic Alliance Australia works alongside medical services and in partnership with support groups, members, genetic services and allied health professionals.

Download a copy of this report here: <http://www.geneticalliance.org.au/genome.php?1>

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(Accessible here <https://www.geneticalliance.org.uk/our-work/diagnosis/taking-the-patient-and-family-perspective-to-the-centre-of-genomic-sequencing-policy-making/>)



Thank you to the **Icahn School of Medicine at Mount Sinai** for kindly allowing us to use their video, '*Whole Genome Sequencing and You*' as an accessible resource throughout our survey.

(Accessible here: <https://www.youtube.com/watch?v=IXamRS85hXU>)



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Executive Summary

Genetic Alliance Australia's *"Australian Patients' and Families' Perspectives on Genome Sequencing"* project commenced September 2015 out of an expressed need by our members to clarify perceptions of genome sequencing and to better understand the impact on families. Questions about financial cost, overall benefit, incidental findings, privacy and other aspects were addressed in the project. Following a detailed national survey (411 respondents) and a focus group, Genetic Alliance Australia (GA) is providing guidance on what should be deliberated when patients, carers, the general public and policymakers are considering genome sequencing.

The project showed that people who have a genetic condition, or their carers and relatives feel a need to be involved at all stages, have a strong desire to find a diagnosis, and want to receive as much information as possible. For 77% of respondents there were high expectations that genome sequencing would help provide a diagnosis, options for targeted treatments and deeper insight into their illness. There was the hope that a diagnosis could help with treatment. Seventy-three percent (73%) of respondents said that genome sequencing would have been useful to them in the past to find a diagnosis quicker instead of being referred to specialist after specialist, repeating the same story, struggling to find a diagnosis. Almost all (97%) agreed the survey was very worthwhile. Areas of concern included availability of genetic counselling services, who could access genome data, cost, and the accuracy and potential for misdiagnosis.

The need for medical genomic training is understood. However the need for genomic education of families and patients is paramount, *"Advances in medicines have led to unrealistic expectations on the part of patients"* (Anderson 2004).

This report lists twenty-one recommendations over six identified themes to be considered when incorporating genomics clinics and testing services into the healthcare system:

Understanding and Communication

1. Patients' understanding of the complexities of genome sequencing, its risks and benefits, needs to be guided by educators and healthcare professionals
2. Healthcare professionals should acknowledge the patient's search for information and assist them in determining accurate and appropriate information
3. All healthcare professionals should receive training in genomics, its' application and relevance, and patient communication
4. Healthcare professionals, research institutes and patient organisations should develop clear and easy-to-access resources for patients in different formats and languages
5. Adaptable resources should be accessible to people with varying disabilities

Consent

6. Ensure consent processes are informative and give patients enough time to process and understand the implications of genome sequencing
7. Dynamic Consent should be the standard model of consent used for clinical genome sequencing practice

Disclosure of Findings

8. Patients would like to be part of a shared decision-making process about what incidental findings they receive, when and how they receive them

9. Clinicians and genetics professionals should be trained in delivering genomic results to patients, and have a knowledge of referral pathways
10. Appropriate resources, information of patient organisations and options for further support should be offered to patients upon receiving their results
11. Obtaining a diagnosis should be a priority when genome sequencing is used in clinical practice and this should be reflected in the resources allocated to re-analysis

Genetic Counselling and Continuing Support

12. Genetic counselling services should be offered at all stages of the genome sequencing process, even in the case of research, and should be accessible by patients when requested
13. More support and resources should be given to expand the genetic counsellors' workforce

Research

14. Research aims, objectives, stakeholder involvement and access to data should be made clear to participants
15. Consent must be sought before a patient's genome is used in research projects
16. Guidelines should be developed to ensure an adaptable approach towards delivery of research findings to patients
17. Genomic research should be conducted by reputable organisations who will be held accountable for their actions
18. Researchers must ensure all patient data is securely stored

Cost

19. Cost and equity of access to genome sequencing through a Medicare subsidy must be a healthcare policy priority

Other Considerations: Life Insurance and Employment

20. Life insurance providers should be involved in an active discussion with stakeholders on how to provide for patients who have had their genome sequenced
21. Referral to information regarding patients' rights, and the implications of genome sequencing on employment and life insurance products should be provided as part of genetic counselling and consent processes

CONCLUSION

"As genome sequencing is integrated into clinical healthcare practice around Australia, this report documents the patient voice. Patients living with genetic conditions are calling to be a partner in research and treatment. Patients can provide insights into little-understood experiences and life impacts and in the rush to interpret genome sequencing, it is important that the patient is not forgotten" – Dianne Petrie OAM, Executive Director of Genetic Alliance Australia

Healthcare providers have a responsibility to ensure that the reality and expectation of genome sequencing is appropriately communicated and the risks and benefits clearly outlined. It must be

recognised that genomic technologies and medicine are still evolving and will change as technology and insights from data analysis improves. The potential for genome sequencing to provide a diagnosis, future treatment and the ability to reduce the cost of many diagnostic tests is an important point to be considered by policy and decision-makers in the healthcare system.

Family and patient education and collaboration are the keys to the successful integration of genomics in the Australian healthcare system. It was clear patients are very positive about genome sequencing but it is critical healthcare providers and service providers ensure that risks and benefits are clearly communicated and expectations are effectively managed by clinicians trained in genomics.

Patients are on the edge of a dynamic shift in medical diagnostic treatment. This comprehensive report and Patient Charter is the first of its kind in Australia. The report serves not only as an important patient voice for rare genetic conditions, but it is a starting point to build on to ensure Australians make well informed shared healthcare decisions. The Patient Charter outlines important points to be considered by patients and families when having their genome sequenced.

GENETIC ALLIANCE AUSTRALIA'S PATIENT CHARTER ON GENOME SEQUENCING

This Patient Charter serves as a guide for patients, carers and anyone considering genome sequencing in Australia. The aim is to provide questions that should be considered before consent for genome sequencing is given. You should ask your healthcare providers these questions to ensure that you really understand the process, risks and benefits

- **RELEVANT:** Will genome sequencing provide the information that I need?
- **CLINICALLY MEANINGFUL:** What will I be able to do with this information i.e. will my healthcare or decisions change as a result?
- **FAMILY CONSIDERATIONS:** When I receive the results, will I need to inform my family and relations? Will my results impact on them?
- **INCIDENTAL FINDINGS:** What happens if I find out about something unrelated to my current condition (incidental findings)? Do I want to know about these incidental findings? Can I choose when and how the incidental findings are reported to me?
- **IMPACT:** Will knowing this information impact on my daily life? Would it affect life insurance?
- **PRIVACY:** Will my data be kept anonymous?
- **DATA SECURITY:** Will my identified data be shared with anyone?
- **ACCURACY:** How accurate is genome sequencing?
- **CONSENT:** Can I change my consent for sequencing testing and receiving incidental findings at any time? Will I be given a copy of my signed consent form? Will I be given information to read at home?
- **RESEARCH:** If your sequenced genome is part of a research project – What other research will this information be used for? Can I choose which genome research projects I contribute to?

- **PROCESS and TIMELINES:** How long will the genome sequencing process take and when will results be available? How much will it cost? Who will give me my results?
- **RESULTS:** Will I be able to understand my results?
- **GENETIC COUNSELLING:** How many genetic counselling sessions will be available to me?
- **TECHNOLOGY CHANGES:** Will I need to have my genome sequenced again at a later date?
- **INTERPRETATION CHANGES:** Will I be contacted if the interpretation of my genome test results changes with increased knowledge?

HEALTHCARE PROVIDERS DUTY OF CARE

From the survey it was clear that patients wanted to be informed about genome sequencing if it would provide any benefit to their healthcare. To ensure patients get the best possible guidance and service the following should be available:

- **REFERRAL PATHWAY:** Clear referral pathway from general practitioners, allied healthcare and specialist medical disciplines is needed.
- **PLAIN LANGUAGE:** Information provided should be in plain language with appropriate glossary and references to help people put the information into context.
- **INFORMED CONSENT:** The information in the consent form needs to be explained in plain language so that the consent is informed, and a hard and soft copy of the form for long term reference given to the patient.
- **RESOURCES:** Patients should be provided with multi-media resources (e.g. Talking Glossary*) to ensure all patients including hearing or sight impaired can make fully informed decisions. Resources should also be provided in different languages upon request
- **TIME:** Patients should be given sufficient time to consider genome sequencing and research questions before final consent is given, they should also be given material to access and read at home
- **ACCURACY:** Accuracy of technology and results need to be clearly communicated by qualified clinicians trained in genomics
- **COMMUNICATION OF RESULTS:** The results should be presented in a clear and unambiguous way. Education and guidance should be available to ensure GP's, specialist and healthcare providers can explain the results accurately and consistently
- **SUPPORT:** Healthcare providers should ensure that patients are provided with appropriate genetic counselling and put in contact with relevant support groups to ensure psychosocial support
- **EDUCATION:** Healthcare providers should be given opportunities to take part in genomics education on its application, relevance and patient communication

*National Human Genome Research Institute *Talking Glossary of Genetic Terms* (<https://www.genome.gov/glossary/>)

Case Study: Leigh Atkins

My name is Lorraine and my husband is Ron. We have two children. Leigh (34), who is intellectually disabled, and Danielle (30).

When Leigh was born all seemed OK. It wasn't until he did not achieve the normal milestones that we feared what may lie ahead and decided to seek help. At that time the only help available was our GP, paediatrician and some small centres offering weekly therapy sessions.



The Atkins Family (L-R): Danielle, Lorraine, Ron and Leigh

By the time he was two years old, Leigh's physical abilities had developed to the stage where he could walk but his fine motor skills still lagged behind those of his peers. After countless physical therapy sessions there was a gradual improvement with his fine motor skills, but Leigh still has difficulty with simple tasks such as catching and throwing a ball.

Throughout his early childhood, we were told that he had a 'global delay' which gave us a false hope that he would 'catch up', but it soon became evident that he would need special education for his entire school life. During these years at a special school, Leigh had ongoing speech and occupational therapy.

Now at thirty-four years of age, Leigh has no intelligible speech, a basic form of signing only recognisable to those who know him, he can only just write his name when prompted and he can just manage to dress himself, provided there are no buttons involved. On the plus side he has an uncanny ability to read a street directory, he can operate a DVD/VCR recorder and watch movies on his PC after visual instruction. Leigh's inability to express himself has led to years of frustration for him and his family and we felt at a loss as to how we could help him without a reason for his disability.

Another big concern for us was about having more children due to the lack of a diagnosis. During the pregnancy and after Danielle was born we worried about her too, would she be OK, would she carry whatever Leigh had? After Danielle was born, we made the decision not to have any more children.

Like all parents of intellectually disabled children we sought a reason for Leigh's disability. After a few years of seeking help from the few geneticists accessible to us we gave up hope of ever finding an answer. However, as our daughter Danielle reached her early twenties and entered a long term relationship, we thought it was important for her to know if Leigh's condition was hereditary as we don't want her to go through the same anguish we have had to endure.

We, again, sought a diagnosis for Leigh's condition. Now, we hope that with genetic testing of ours and Leigh's blood through genome sequencing we may get an answer as to whether Leigh's disability is or is not a genetic condition.

Genetic counselling and a diagnosis would mean a lot of to our family after thirty-four years of wondering.

Case Study: Massimo Damiani

My name is Stephen and this is a story of our journey towards a diagnosis for our son, Massimo. Our journey, or mission if you will, really began in Paris almost fourteen years ago, where my wife and I had just enjoyed our European honeymoon. Our flight home was delayed so I whiled away the time reading magazines. One was an issue of Time Magazine titled “Cracking the Code – the inside story of how these bitter rivals mapped our DNA, the historic feat that changed medicine forever”. The article described their race to sequence the first human genome. Little did I know, six years later I would look back and realise its significance.



The Damiani Family (L-R): Marco, Leo, Stephen, Massimo and Sally

On the 22nd of July 2008 Massimo joined our world. The first four weeks were pretty much the norm – feed, sleep, poop, change nappy. On one occasion I noticed a small skin tag just above Massimo’s bottom and became mildly concerned. After an ultrasound, it turned out Massimo had Spina Bifida Occulta. Massimo underwent a brain and spine MRI to check for further involvement. The neurosurgeon advised to monitor Massimo for the next eleven months before making any decisions about surgery.

Massimo had a great first year hitting all the key milestones on or ahead of “schedule”. However, in the week preceding his first birthday we observed some concerning signs of regression. Massimo was struggling to pull to stand and was getting very frustrated. His legs and ankles were stiff. He started having trouble with balance and kept thrusting his head back. Something didn’t seem right and his follow-up spine MRI didn’t ring alarm bells, at first. The next day we visited his physio at The Royal Children’s Hospital. By now the MRI had been formally reported noting a possible abnormality in the region of his thoracic spine. We were ushered to emergency where we were met by his neurosurgeon and she suggested it was a possible transverse myelitis and ordered a brain and spine MRI.

I remember getting concerned after an hour as the procedure should have taken only forty minutes. Finally, the neurosurgery registrar entered the waiting room after two hours. He asked a series of odd questions mainly directed at my wife about her diet and even if we were related, but gave away little before leaving the room and saying Massimo was in recovery. Although uncertain of exactly what was going on, we somehow knew we’d just lost our son. There were no tears, just silence filled the room. When we saw the sombre faces of the medical staff in recovery it confirmed our worst fears. Needless to say we didn’t sleep a great deal that night.

The next day, Massimo’s neurologist gave us our first look at his brain MRI. A new language entered our vernacular, but the most frightening word we were to hear was Leukodystrophy: a condition where the insulation surrounding his nerves was breaking down and signals couldn’t effectively travel down the pathways. A barrage of tests were ordered and we were told we had to be patient, but at no stage did I think we wouldn’t have an answer within hours or at worst, days. Massimo was discharged and we returned home inconsolable. Leukodystrophies are a group of rare neurodegenerative disorders that can rob affected individuals of their sight, and eventually all senses. The prognosis for infant onset Leukodystrophies is particularly poor with a life expectancy in months, or at best only several years. There was no treatment, no cure and intervention was purely supportive of symptoms. In the weeks that followed every test frustratingly returned back normal. In our follow up consultation, I resigned myself to Massimo’s fate breaking down and saying “*There really*

is no hope is there?". I knew what the answer would be but Massimo's neurologist thought about it a moment and responded "*Never let go of hope – there is always hope*".

We were narrowing down the remaining possibilities and genetic tests were ordered from overseas laboratories in the United States and Italy. The time frames for these tests were many months, we needed to be patient. We were also told to be prepared because up to 50% of Leukodystrophies remain genetically unclassified. Meanwhile Massimo's condition continued to deteriorate rapidly. He started to choke on food and water, lost all his vocabulary and could no longer crawl or sit. I'm sure he knew something terrible was happening because he looked really scared. Things were happening too fast. Never had I found myself in such a hopeless situation. We couldn't see the end – weeks, months, years or decades? You start planning for every possible future outcome but can only live for the day.

The only way I could cope was to throw everything behind a diagnosis. I had to know why this was happening and if we were to one day access or develop a treatment we needed a diagnosis. I simply wasn't going to give up until I had one. This was going to be a moon-shot project. Clinicians and scientists needed to be inspired by imagination and let science drive the diagnosis. They needed to be free from all constraints financial, bureaucratic, or otherwise, but they also needed to have a fire in their bellies to make this happen. Enter Mission Massimo - its motto - *Scientia Est Potentia* - Knowledge Is Power. Because, the knowledge of a diagnosis would give us power to develop a treatment.

Three months passed since that initial MRI. We hadn't received the results from the previous overseas tests and they were already redundant. Without a confirmed genetic diagnosis the risk of having more children was too great. Therefore, in late 2009 we made the decision to proceed with a donor egg pregnancy.

It was at this point I raised whole genome sequencing (WGS). After all I'd read about it years ago and surely this '*historic feat that would change medicine forever*' was now a reality. After another long awaited MRI three months later, it was suggested that Massimo may have early stage Vanishing White Matter (VWM) disease although his clinical presentation didn't fit with the disease. Consequently, none of the limited public funding was made available as it was deemed there was no imminent risk, and private health insurance didn't cover genetic testing. At this point we were effectively on our own and if we were to fund the VWM tests privately the cost was around \$10,000 – coincidentally the new price of Illumina's whole genome sequencing (WGS) service. We decided to proceed with WGS. I was convinced Massimo had an unknown variant and we needed to go wider rather than focus on known conditions. Why not sequence all 30,000 genes instead of just a few at a time. The clock was ticking and I wasn't prepared to wait another twelve months.

I arranged to have the data analysed by the National Centre for Genomic Research (NCGR) in the US, who would align Massimo's genome to the Human Genome Project baseline. We hoped to identify only a few candidate genes, however the analysis identified 11,500 variations across 5,700 genes, a number clearly too great to be meaningful. The next logical step was to have our parental genomes sequenced to run a differential trio analysis and hopefully reduce the noise. We were introduced to a post-doctoral Fellow in Bioinformatics, Dr Ryan Taft who agreed to take our call. I think he was expecting a ten-minute conversation with a desperate father asking what a genome is. Instead Ryan was being asked to validate our strategy and two hours later we were still talking. Although not the focus of his research, Ryan was keen to perform the analysis himself and shared our story with Illumina who offered to provide reagents in-kind for the sequencing. Ryan completed the trio analysis in early December 2011. Incredibly he narrowed down our 11,500 variations to a single compound heterozygous mutation in the DARS gene. We hoped this was finally the answer to all our efforts, expecting congratulations. Instead, our efforts were met with a period of silence before an

inconvenient trust was raised. It wasn't possible to diagnose a single patient. Now we needed to prove it. We needed another Massimo.

Upon sharing the findings with the research leader of a project Massimo had been involved in at the Children's National Medical Centre in Washington DC, five families were identified with similar imaging and clinical presentation. All of these patients underwent whole genome sequencing for parallel analysis. A family of five with two affected children were identified as having mutations in the DARS gene validating the preliminary finding. The finding was further validated with seven patients identified in parallel from Europe and the Far East with mutations in the DARS gene. We had successfully diagnosed a previously genetically unclassified Leukodystrophy and provided the proof of concept for familial trio genome analysis as an effective clinical diagnostic tool for novel genetic disorders.

I feel truly privileged to have been part of an incredible international team of clinicians and scientists that worked tirelessly around the clock to help save a brave little boy from an uncertain fate and blessed to have had loyal family and friends provide strength and inspiration in troubled times. Beyond Massimo, I hope our collective efforts will pave the way to diagnose many other children and just maybe treat their rare genetic disorders. **What took us 1,161 days may soon take weeks, days or eventually hours, allowing early intervention and treatment.** Above all, looking past the incredible science and technology it offers **hope** to the many families in the terrible situation that we were once in ourselves.

Achieving a diagnosis was only the first step of our journey. Our next challenge is to develop a therapy. Our experience compelled us to establish the Mission Massimo Foundation to raise funds for genomic diagnostics and gene therapy and stem cell research (<http://www.missionmassimo.com/>).



Massimo Damiani

Background to survey

The project *“Australian patients’ and families’ perspectives on genome sequencing”* was conducted by Genetic Alliance Australia in order to develop a Genome Sequencing Patient Charter that outlines the views and opinions of what patients and families want from the accessibility of genome sequencing in the healthcare system. The Genome Sequencing Patient Charter is an important reference guideline to be used by policy and decision makers when considering the widespread incorporation of genome sequencing in the healthcare system.

Genome sequencing is a relatively new technology being introduced into clinical settings in Australia. With the advent of more affordable and publicly accessible genome sequencing services, there is a need to establish what the patients and families affected by diagnosed and undiagnosed genetic conditions perceive about genome sequencing. The requirements of patients and families to be able to use genome sequencing effectively within the Australian healthcare system also needs to be considered.

It is important policy and decision makers are made aware of the informed views of patients and families who will be directly affected by genome sequencing and who wish to be involved in shared decision-making. The experiences and opinions of patients and families with both diagnosed and undiagnosed conditions could greatly impact on healthcare decision-making on the use of genome sequencing, for example:

- What to do with potential incidental findings?
- How far confidentiality of findings extends within the family tree?
- Use of genomic sequencing information in research
- How patients see clinical genetic services operating

The Project

The project was designed and developed from November 2015 to February 2016 and it consists of two parts;

- **Survey**—30-45 minute questionnaire with multiple choice and free text questions that allowed participants to express their opinion and views on the experience the participant has had with a genetic condition(s), testing or genomic sequencing. Throughout the survey participants were allowed to access an educational video, case study text and glossary, which provide more information about genome sequencing.
- **Focus Group**—a forum for participants to think and talk about their views regarding genome sequencing and its uses in a clinical setting, including a discussion about personal situations/experiences, and an exploration of their opinion on future development in this area. The discussions were audio-recorded with permission.

Guidance was provided from our sister organisation Genetic Alliance UK who conducted a similar project in 2014-2015, and who kindly allowed us to adapt their survey and resources to the Australian setting. The project has been put together with detailed consideration of similar projects and work done by the National Institute of Health USA (Human Genome Project) as well as referring to the Centre for Genetics Education, National Health and Medical Research Council (NHMRC) Genome Sequencing Resources and Queensland Health Informed Consent Guidelines.

An ethics application was submitted to the South Eastern Sydney Local Health District Human Research Ethics Committee on 4th March 2016 to assess the appropriateness of the survey and focus group protocols. Feedback was received on 22nd March 2016. The Ethics Committee was concerned

about the wording of specific questions and appropriateness of some terms used pertaining to identification of data. These issues were addressed and a response was sent to the Ethics Committee on 24th March 2016, along with changes to the survey. Further feedback was received on 30th March 2016. Again the survey was reviewed and changes made and sent back to the Ethics Committee on 8th April 2016. The project was granted ethics approval on 18th April 2016.

The project was open to anyone aged eighteen years or older residing in Australia who was either;

- Living with a diagnosed genetic or rare condition, or
- Living with an undiagnosed genetic or rare condition, or
- A carrier of a gene fault (i.e. do not have a condition but are able to genetically pass on the gene fault to their children, or are at increased risk of developing a condition), or
- A parent of a child/adult who has a genetic condition (the child/adult may or may not be living at home), or
- A relative of a child/adult who has a genetic condition, or
- Caring for a child/person with a genetic condition

The online survey was launched on 6th May 2016 and ran for two months. It was closed on 8th July 2016. Hard copies were made available upon request (there were two requests). The survey was distributed to GA members, individuals and families through Genetic Alliance Australia's database, to other genetic and rare disease support groups and organisations. The project was advertised on GA's website and social media outlets (Facebook and Twitter). GA would like to acknowledge Syndromes Without a Name (SWAN) Australia, GA's support group members, families and individuals who completed and distributed the survey.

Participants for the focus group were recruited through the survey (at the end of the survey there was an option to take part, this was de-identified from the information provided in the survey), and through the advertisements placed on GA's social media and website. The focus group took place on 26th July 2016 with two participants in the GA office and five participants via teleconference. The focus group ran for ninety minutes. It was moderated and recorded by Nicholette Conway from GenomePlus. A transcript was written and coded. Participants were from different states and with different educational backgrounds. Each person's situation and their interest in genome sequencing was different. The focus group provided an excellent insight into patients' and families' needs and understanding, providing depth to some of the survey's key issues, such as informed consent, referral pathway and genetic counselling.

Data analysis methodology

The data was collected, collated and analysed using the online Survey Monkey platform. The following procedures were used:

- Survey Monkey methodology was used to collate and analyse the data. Graphs representing the basic count and averages and related percentages per question were reviewed and analysed for insights
- The Survey Monkey data was exported to Microsoft Excel as raw data.
 - Data was reviewed to determine any data deviations. Only two entries were found to be for non-eligible participants and were excluded from further data analysis.
 - The data was analysed with questions cross referenced to determine further insights

- All comments were read. Where relevant and of value to the question the comments were coded to ensure GA could quantify the scope of comments.

Survey response

- 413 respondents
- Respondents on average completed 74% of the survey
- 64% of respondents went through the survey and completed the final question

Data Deviations:

- Deviations were addressed and submitted to Genetic Alliance Australia for agreement. None were deleted.
- Names entered by respondents were deleted
- Two entries not from the target population, therefore 411 responses are used in this report

The response to the survey was very encouraging and showed that those who participated gave good consideration to all the questions. There was very little drop off.

In total there were 556 individual comments to questions, demonstrating participants' careful consideration in their responses, giving additional valuable insights. It also was an indication of a strong and growing interest in genome sequencing within the community.

Table 1. Percentage of respondents for each section in online survey

		Percentage of Respondents (%)				
		Section 1 – Informed consent	Section 2 – Impact of results	Section 3 – Interpretation of results	Section 4 – Use of genome sequencing in research	Section 5 – Information security and sharing
Participants responsiveness to questions	Highest Response	100	91	67	65	65
	Lowest Response	73	33	65	63	64
	Average Response	92	72	66	64	64

Demographics

- Cultural background: 93% Caucasian, 2% Aboriginal, 2% Asian, and the remaining 3% were of Maori, Pacific Islander, Middle Eastern and African descent
- English predominant language: 97%
- Age: 9% under thirty, and 15% over sixty-one years. Equal split over the other age groups
- Education: 85% grade ten or higher, 40% university educated

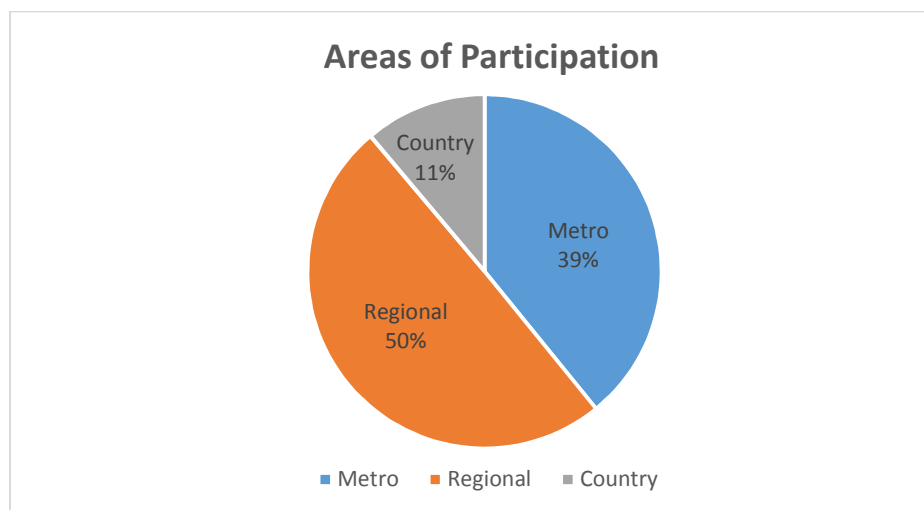


Figure (i) Areas of Participation

As per the Australia Bureau of Statistics (ABS) all the postcodes provided were classed under the following headings:

Metro – region consisting of a densely populated urban core and its less-populated surrounding territories, sharing industry, infrastructure, and housing.

Regional – Regional Australia includes all of the towns, small cities and areas that lie beyond the major capital cities (Sydney, Melbourne, Brisbane, Perth, Adelaide, Darwin, Hobart and Canberra).

Country – a rural area or countryside is a geographic area that is located outside towns and cities.

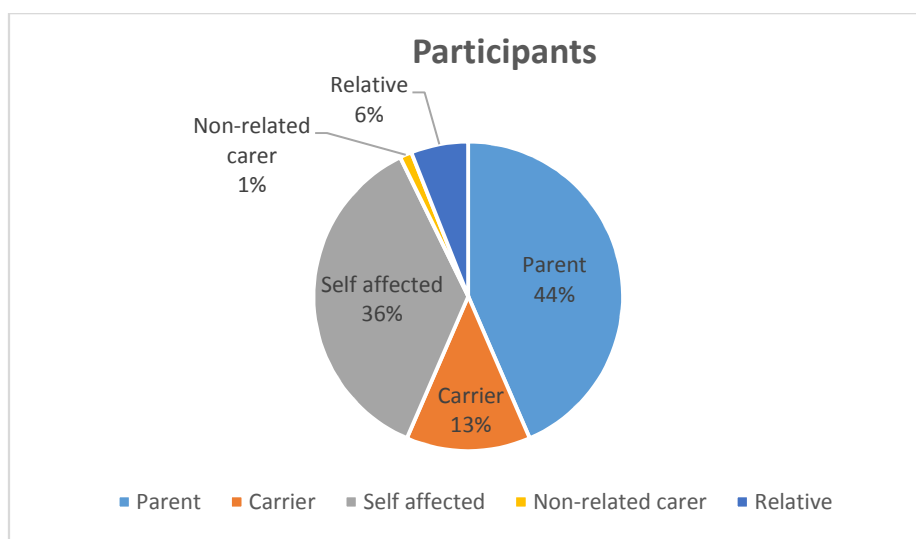


Figure (ii) Participants

There was a range of respondents completing the survey, with a majority of respondents being either a parent of a child with a genetic condition, or had a genetic condition themselves. Eighty-seven percent (87%) of respondents already had a diagnosis. This is important to note as the population GA surveyed is quite familiar with concepts of genetics having dealt with rare genetic conditions in their life. They are an interested and engaged population who may already have preconceived ideas and feelings of how this technology will impact on them. It is a population not just merely interested in their health but are looking for answers for the future/treatment.

Findings

The main themes emerging from the results.

- Understanding and Communication
- Consent
- Disclosure of Findings
- Genetic Counselling and Continuing Support
- Research
- Cost
- Other Considerations
 - Life insurance
 - Employment

Underlying all these themes was one key message:

Patients and families have high expectations about what genome sequencing can offer them.

The potential to find a diagnosis or to learn more about their condition and what lies in store in the future was a high priority

“Having a diagnosis can relieve uncertainty, anxiety, isolation and frustration. It leads to better treatment plans and allows you to plan for the future” – Heather Renton, Founder of Syndromes Without a Name (SWAN) Australia

For many patients and families, the potential for answers was so powerful they were willing to overlook the potential risks that come with this new technology.

UNDERSTANDING AND COMMUNICATION

Public engagement and education on genome sequencing and its implications is an important factor in developing the appropriate policies and infrastructure for its incorporation into the health system.

Perceived understanding vs. real understanding

Genome sequencing is a relatively new concept for most patients and families. From the survey it was clear that participants were aware of what genome sequencing could offer them in general, but were less informed about the complex details regarding genome sequencing. When asked about how easy it was to explain genome sequencing, 48% believed it was easy to explain. Comments from the questions suggested that their understanding of the complexity of genome sequencing depends on a patient’s current knowledge of the topic and their ability to take in information.

Participants demonstrated a basic understanding. Many patients have researched this topic at home and have come up with a concept of what genome sequencing can provide for them, which skews their real understanding of what genome sequencing can realistically provide for them. This is also impacted on by their personal experiences and the hope of what new technologies can do for them. Anecdotally, many families who have approached GA regarding genome sequencing have struggled to fully appreciate and understand the process. This is not surprising as there are numerous aspects to comprehend. Also with the respondent's personal involvement, the focus has been on immediate outcomes, where a wider group would be expected to flag and acknowledge how useful the results would be clinically.

Some respondents appeared to get confused when talking about genetic testing vs. genome sequencing. Seventy-five percent (75%) of participants had experience with genetic testing and 22% with genome sequencing.

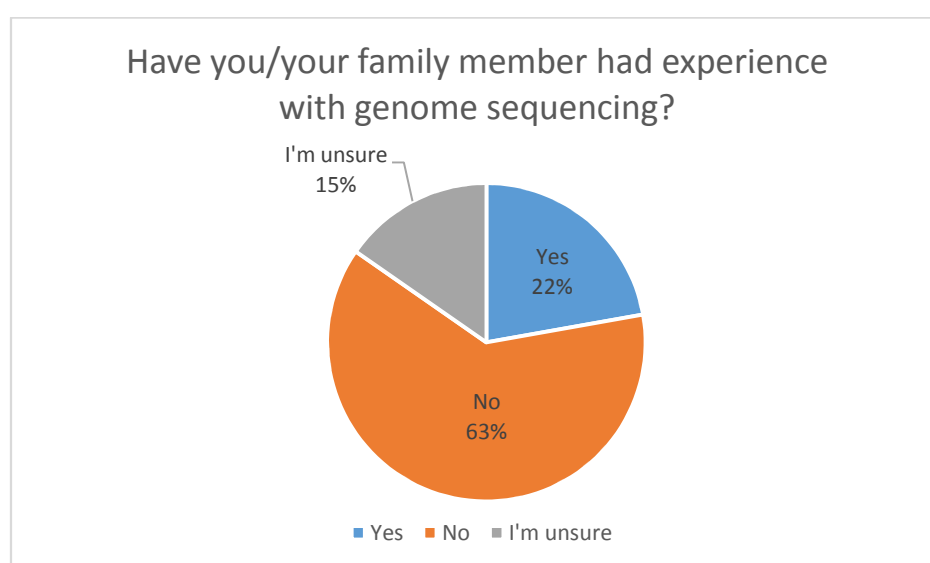


Figure (iii) Experiences with genome sequencing

Table 2. Of those who said they have had experience with genome sequencing, where did you get it done?

	Public system	Private testing	Research	Unsure
Genome Sequencing (n = 84)	60	8	2	14

With the limited availability of genome sequencing in the public healthcare system, with most being done through research, GA are cautious about the results in Table 2 (Appendix A). When these results were cross referenced it was noted that these responses did not match up well. Of the fifty-eight respondents who said they were unsure whether they had genome sequencing, they still answered questions about where they had it done and if they received genetic counselling. Eighty-four respondents said they or a family member has had experience with genome sequencing, however one hundred and thirty-six responded to the question about where it was done. These results should be interpreted with caution,

"Now I'm not sure if my whole genome has been sequenced or if they were only doing a genetic test to identify the [gene]"

however it indicates to Genetic Alliance that there is confusion over understanding of differences between genetic testing and genome sequencing.

As part of the feedback collected in the survey, Genetic Alliance asked whether participants believed that their understanding of genome sequencing had changed after completing the survey. Thirty five percent (35%) said “Yes” while 56% said “No”. Comments in this section suggested that participants felt more informed and that they had been made aware of issues that they had not previously considered.

“I am more sure about what information I would want to be given and what I would and would not want to happen to my information”

“My understanding has changed regarding the ethical and long term effects”

“I have more of an understanding of “incidental findings” and how my genetic information may be identified”

“I didn’t know about the different ways information can be identified”

“It made me think of scenarios I hadn’t considered previously”

“It got me thinking about an area of science/medicine I don’t know much about”

In the literature it is well documented that patients often have limited understanding of genetics and genomics, and what tests they can access (Kolor et al 2012, Gray et al 2012). A report released in the USA by the Secretary’s Advisory Committee on Genetics, Health and Society (2008) highlighted five gaps that can affect use of genetic tests such as genome sequencing, one of which was “*deficiencies in the genetic knowledge of practitioners, public health workers, patients and consumers*” (Ferreira-Gonzalez et al 2008). It is important for the healthcare industry to lead and direct this dialogue, rather than leaving it to the patient to find it all on their own and potentially developing an unrealistic view of what genome sequencing is and what it can provide for patients. It is also important for the healthcare industry to put itself forward as the authoritative body in providing reliable and accurate information about genome sequencing.

RECOMMENDATION: Patients’ understanding of the complexities of genome sequencing, its risks and benefits, needs to be guided by educators and healthcare professionals

Expectations of Genome Sequencing

Often for patients finding a diagnosis or an explanation of symptoms is much more important than any negative outcomes that can occur with genome sequencing. Many patients have been waiting for years for an answer to their questions. The benefits in many ways outweigh the negatives. Potential for diagnosis is so overwhelming, there is concern that the implications are not being considered enough.

“I think the benefits far outweigh all these concerns”

Patients’ over-expectations of what genome sequencing can provide is well documented in the literature (O’Rourke et al 2013, O’Daniel et al 2010). Research also shows that attitudes towards tests are influenced by what the test is and how the public understands it (Hall et al 2016). This can stem

from an incomplete understanding of what genome sequencing can provide “in response to input from a variety of sources such as health-care providers, researchers, insurers, industry, the media, and often dramatic experiences of friends, family, or others” (O’Rourke et al 2013).

“I would want to have the option, but with clear statements about the positives and negatives...I think there needs to be clear and specific guidelines on what should be communicated to patients”

It is important to maintain a level of enthusiasm and excitement about the potential of genome sequencing, while acknowledging that there are still limitations to its use and clinical utility. It is also important to identify all the various sources of input that can influence patient expectations to see how these can be used to convey the right message about genome sequencing to patients.

Sources of information

In order to ensure that patients are receiving the correct information about genome sequencing and how it will affect them, it is important to observe where they source information about genomics.

Participants were asked where they sourced information regarding genetic testing and genomics. As expected, genetics clinics and genetic counsellors were a valued source of information to participants. The internet, medical specialists and general practitioners were also a popular choice.

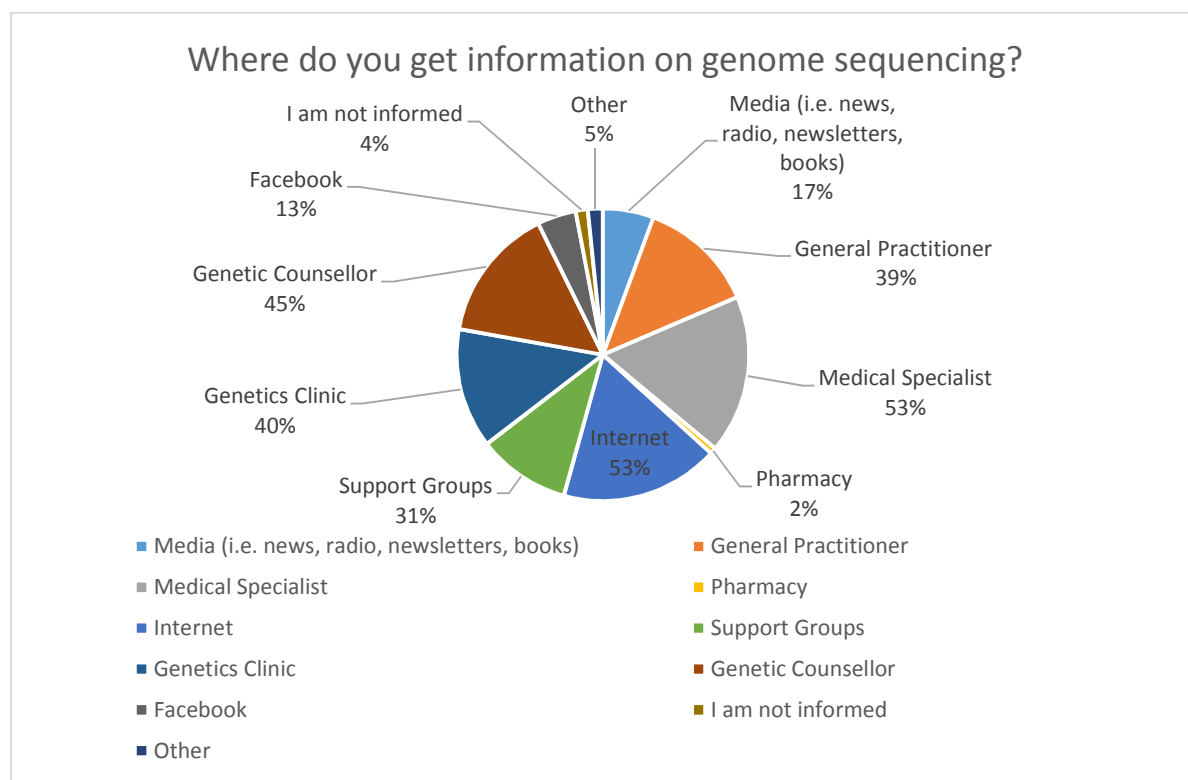


Figure (iv) Where respondents get their information from

Internet and Social Media

The internet is one of the most popular sources of information for patients with 53% of respondents reporting they use it as their main source of information on genome sequencing. It is an easily accessible platform that provides information almost instantaneously. Accuracy of this information is questionable and often provides biased and uninformed views. This can be extremely detrimental to

the development of effective genome sequencing services, and can provide high (or even low/skewed) expectations of what genome sequencing can provide for patients (O'Rourke et al 2013). Social media is another aspect of internet-based sources of information that should be treated with caution. Facebook was seen as a source of information for nearly 20% of respondents.

Table 3. Number of Facebook users in each participant age group

	31-40 years old	41-50 years old	51-60 years old
No. of Facebook users	17	21	11

Information is often posted and shared quickly on social media channels, about interesting or polarising topics. Content is shared in small bits or sound-bites, and can present various points of view, inciting comments and opinions from its audience. It has proven to be an influential platform that can sway and influence public opinion. Places like Facebook are also a space for people to connect with others in similar situations such as theirs, from all over the world. Formation of online support groups has revolutionised the way that patients find support and guidance. For many, these groups are also a space for patients to ask questions, express concern and share information. It is often treated as a place of trust for information relevant to them.

While patients do gather much of their information from the internet, this does not tend to displace the role of a healthcare professional (McMullan, 2006). Confirmation, or the need to find additional information is often what drives people to turn to the Internet. Patients have moved from being passive to active consumers, and healthcare professionals need to acknowledge this. Patients want to be fully informed and be part of the decision-making; a patient-centred relationship that focuses on a shared-decision making model.

THE SHARED DECISION MAKING MODEL (Elwyn et al, 2012)

Shared Decision Making has been defined as an approach where clinicians and patients share the best available evidence when faced with the task of making decisions, and where patients are supported to consider options, to achieve informed preferences

RECOMMENDATION: Healthcare professionals should acknowledge the patient's search for information and assist them in determining accurate and appropriate information

Medical Specialists/Physicians

Medical specialists are often seen by patients for specific issues. As they specialise in various areas of medicine, those with specific chronic issues will see specialists over a long period of time, developing relationships. Fifty-three percent (53%) of respondents from our survey have identified medical specialists as a source of information. When asked who they trust to talk to about genome sequencing, 46% of patients said they trust medical specialists.

"We think that physicians from different specialties can order this test if they're willing to take the time and commit to the effort to learn what the test is, what it isn't, how it works, what it tells you and doesn't tell you, and how to use the results" – Dr Leslie G. Biesecker, Chief of National Human Genome Research Institute's Medical Genomics and Metabolic Genetics Branch (NIH 2014)

Specialists are often familiar with a patient's medical history and background. During a survey of the healthcare needs of the rare disease population in Australia, it was observed that 81.4% of patients had seen a medical specialist in the twelve months prior to the study (Molster et al 2016). It is imperative that medical specialists are trained in at least the basics of genomics so they can advise patients appropriately when approached about genome sequencing. Clinicians with a non-genetic background need to be able to recognise if they are not able to advise their patient appropriately and seek further training. Currently non-genetics trained professionals can access the training opportunities provided by bodies such as the Centre for Genetics Education and Garvan Institute of Medical Research

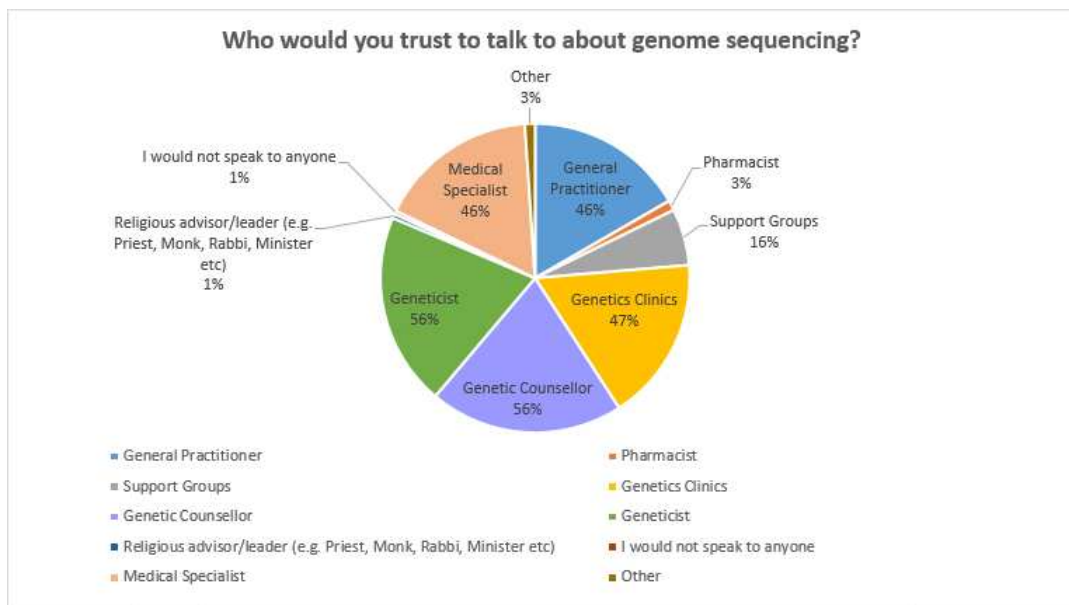


Figure (v) Who respondents trust to talk to about genome sequencing

Genetics Professionals

Genetic counsellors and genetics clinics were seen as trusted sources of information by 45% and 40% of respondents respectively. They are also the most trusted people that respondents would talk to regarding genome sequencing. Genetic counsellors and geneticists are now being faced with the prospect of expanding their skills set so they can adequately see patients undergoing genome sequencing. The workshops provided by the Centre for Genetics Education are also available for genetics professionals. There are discussions taking place around altering current university curriculums for genetic counselling to include a genomics component. It is important for genetics professionals to be kept up to date, as genomics can differ

“Along with the benefits engendered by genomic testing that has become cheaper and faster and more accessible has come challenges in terms of education and training for the genetics health workforce. Training programs for clinical geneticists and genetic counsellors are meeting these requirements and graduating genomics-prepared clinicians. To meet the needs of those in genetics practice, workshops have been held in many States and web-based education seminars are also promoted by professional societies” –

Associate Professor Kris Barlow-Stewart,
Director of Master of Genetic Counselling,
University of Sydney

greatly from genetic testing and requires a specific subset of skills to interpret results, assess clinical validity, how this will impact on patients and their families, and how best to communicate this to them.

General Practitioners (GPs)

General Practitioners (GPs) are often seen as the 'gate keepers' as they are the first point of contact for most patients. Patients trust them to provide information on appropriate services and to refer them on when necessary. Thirty nine percent (39%) of respondents said that they get their information from GPs and 46% would place their trust in a GP when talking about genome sequencing. The primary care setting has been flagged by patients as a vital source in obtaining at least preliminary information on how they can access genome sequencing and whether it is relevant to them. Rare disease patients in Australia in particular use GP services regularly (Molster et al 2016), most likely for management of their condition. GPs are in a unique position to provide advice and guidance to patients seeking genome sequencing (Blashki et al 2014). However, GP education on genomics has been relatively scarce to date (Blashki et al 2014, Walter et al 2012) and there has been little formal guidance for GPs in Australia on how to communicate to patients the process of having their genome sequenced.

With regards to the training of new GPs entering the workforce, The Royal Australian College of General Practitioners (RACGP) is the professional body for GPs in Australia. The RACGP is responsible for maintaining standards for quality clinical practice, education and training, and research in Australian general practice. It is important that the RACGP addresses the need to educate GPs on the increasing relevance of genomics in their clinical practice, and train them in the appropriate skills on interpreting and communicating results to patients.

Genetics in Family Medicine: The Australian Handbook for General Practitioners was developed to assist GPs on how to care for patients that present with conditions with a genetic component. The handbook was published in 2007 and has since been rescinded by the NHMRC. In May 2015 responsibility for the handbook was transferred to the RACGP. The handbook is still considered to be

Genetics in Family Medicine: The Australian Handbook for General Practitioners © 2007

GP's role

- Consult with, and refer, to Genetics Services for clarification of genetic issues, risk assessment, counselling, diagnosis, testing and support when needed.
- Be aware of relevant support groups that may be useful regarding specific conditions, promote their service, and promote patients to make contact with them.
- The GPs role in ordering specific genetic tests will vary according to the condition.
- Be aware that the patient's ethnicity or cultural background can guide the ordering of specific genetic tests.
- Inform the patient about the purpose, personal, and family implications, of a genetic test prior to obtaining consent.
- Be aware that patients who have had a predictive or pre-symptomatic genetic test have a duty to inform life insurers of the test result when applying for a new, or altering an existing, policy.
- A GP has no duty to inform the relatives of a patient about a positive genetic test result. Encourage and support the patient to share the information with their relatives.

a relevant document for GPs, requiring updating in some parts. Currently genomic testing is not addressed in this handbook.

Centre for Genetics Education

The Centre for Genetics Education is a New South Wales state-wide education service dedicated to preparing health professionals who are non-genetics trained with the skills and knowledge to manage the impact of genetic and genomic technologies on their practice. Based at Royal North Shore Hospital in Sydney, the Centre's educational activities aim to promote appropriate and equitable access to genetic services for the people of NSW.

“The Centre is involved in workshops for genetic health professionals around genome sequencing and is currently developing a case-based online module to update nurses, midwives and allied health professionals on genetics and genomics. The Centre also contributes to Program 4 of the Australian Genomics Health Alliance (AGHA) undertaking research into the genomic workforce, educational needs and ethics in Australia. The RACGP is currently updating Genetics in Family Medicine: The Australian Handbook for General Practitioners (2007) to provide an online resource on genetics and genomics in General Practice. The resource will be available in 2017” – Kate Dunlop, Director, Centre for Genetics Education

Genetics and genomics education for health professionals is varied across the states of Australia. In New South Wales there is a centralised body, Centre for Genetics Education, which provides genetics education to non-genetics trained health professionals. In Victoria the Murdoch Children’s Research Institute conducts Genetics Education and Health Research, which contributes towards the development of education programs. The Melbourne Genomics Health Alliance was established in 2014, of which one of their aims is to upskill health workers. In Western Australia the Genetics Services of Western Australia provides “educational and training programs relevant to the modern practice of medical genetics”. In Queensland, the Queensland Genomics Health Alliance was established in 2015 to oversee similar activities, a \$25m five-year commitment by the Queensland State Government to conduct research aimed at integrating genomics into everyday healthcare. It is not clear whether there is a service that provides further education opportunities to clinicians in South Australia, or if it is provided by the

“The Garvan Institute of Medical Research provides a suite of educational opportunities for health professionals, researchers and interested non-experts to find out more about genome sequencing and genomic medicine. It runs the Annual Australian Clinical Genomics Symposium (garvan.org.au/aacgs), a genomic events program and a Clinical Genomic Data Analysis course for physicians and genetic health professionals to explore and examine clinical genomic data in depth. Garvan is also involved in research to understand public expectations of personal genomic testing ([Genioz; genioz.net.au](http://Genioz.net.au)), and contributes to the genomic ethics, education and workforce research of the Australian Genomic Health Alliance (australiangenomics.org.au)” – Bronwyn Terrill, Coordinator, Public & Professional Education, Garvan Institute of Medical Research Division, Kinghorn Centre for Clinical Genomics

South Australian Clinical Genetics Services. Currently the Australian Genomics Health Alliance (AGHA) is mapping the education programs and resources available to health professionals. The World Health Organisation specifies that *“it [is] imperative that healthcare providers and genetic counsellors be carefully trained, in order that they can provide appropriate information, guidance and support to patients and their families”* (WHO, 2016).

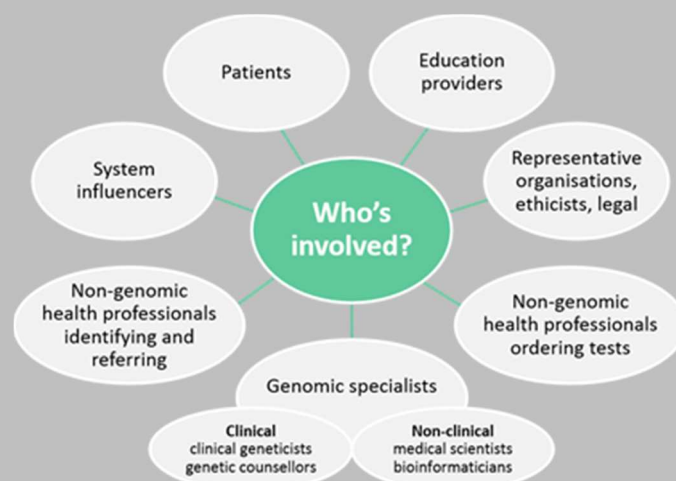
RECOMMENDATION: All healthcare professionals should receive training in genomics, its’ application and relevance, and patient communication

THE AUSTRALIAN GENOMICS HEALTH ALLIANCE

The Australian Genomics Health Alliance (Australian Genomics) is a national research collaboration committed to the integration of genomic medicine into Australian healthcare. Our goals are to shorten diagnosis times, enable early intervention, and to provide access to treatment for people with genetic disorders. Australian Genomics has four core research programs, one of which aims *to understand the education and training needs of professionals whose work will be impacted by clinical genomics*. This research program brings together experts in genetics and genomics education, clinical practice, evaluation, mixed-methods research, genetic counselling, social sciences, science communication and ethics.

Over the course of our five year grant (2016–2020), the research program is gathering perspectives on clinical genomics from several stakeholder groups (shown in diagram below). The research is being conducted through direct consultation, interviews, and surveys across several overlapping projects. The projects aim specifically to: 1) map current education and training activities available to Australian professionals working in genomics; 2) identify future education needs of health professionals; 3) develop an evaluation framework for future genomics education programs, nationally and internationally; 4) investigate the psychosocial implications of genomic testing with patients and families participating in Australian Genomics research, and their experiences of communication with health professionals; and, 5) conduct ethical analyses of clinical genomics, including data sharing, uncertainty, incidental findings and models of consent.

Find out more at: www.australiangenomics.org.au



Support Groups

Thirty-one percent (31%) of respondents believe that support groups are a trusted source of information on genome sequencing. Support groups need to ensure they can provide guidance on medically appropriate pathways and correct information. This is an opening role for patient and advocacy groups to take a more active role in educating and informing their individuals and families, as well as the general public. Support groups should provide balanced and consistent information for their members about genome sequencing, its relevance for their patient group and details about how they can access this information.

Resources for general public

Resources are a vital part of providing education and information to the public regarding genome sequencing. Resources from accredited and trusted sources are important in leading the conversation around genomics. As part of the survey GA provided participants with resources to assist them in understanding the background of the questions posed. The resources were a video, written case study for participants to read, and a glossary to assist participants in understanding some of the terms used throughout. Feedback on the use of these resources was collected. While 35-50% of participants used the resources varyingly, comments suggested that the resources were well received and useful in helping patients understand genome sequencing.

"I've printed out the glossary [from the survey] to keep and I did refer to it when doing the survey"

"The relevant video and case study have increased my understanding of genome sequencing by explaining both the process and how it might apply to an individual"

"I found the video easy to understand and follow"

"The YouTube video was really well done"

"Examples [case studies] also confirm if you think you know what you're talking about, if you think you've just mastered the understanding, an example actually confirms that or displaces it if it's true, that is why examples of case studies are good"

These resources are an example on how to engage patients, families and the general public on these new technologies. Easy to understand, to the point and relatively non-time consuming are important factors when considering resources. In Australia there are limited publicly available resources from reputable sources regarding genome sequencing. The Centre for Genetics Education provides much of this information, which is excellent with clear resources directed at patients and families (<http://www.genetics.edu.au/Genetic-conditions-support-groups/Understanding-Genetic-Testing>), as does the NHMRC (<https://www.nhmrc.gov.au/health-topics/genetics-genomics-and-human-health/genetics-and-genomics-resources-consumers>). This information also should be more readily available through patient support and advocacy groups who are in regular contact with patients, so they know how to access these resources. Support groups and genetics services also should provide a list of appropriate resources that can be accessed online.

"I think you've really got to cater for different mediums. People learn in different ways, some people are more visual, some people use more case studies, I think you've just got to present a lot of different examples"

RECOMMENDATION: Healthcare professionals, research institutes and patient organisations should develop clear and easy-to-access resources for patients in different formats and languages

Based on the findings of the survey and from discussion during the focus group, an appropriate combination of genome sequencing resources could be:

- Short take-home booklet – with pictorial descriptions, and including case studies
- Video – explaining technology(virtual walk through of the process), implications of results, and even consent process
- Ability to discuss issues One-to-One: this would be either over the phone or face-to-face but one-to-one is important

These can be supported by short learning courses or classes either face-to-face or online – seminar series, to build knowledge. There are various tools like these available such as Massive Online Open Courses (MOOCs) that aim to educate people on different topics at various learning paces and levels. Places such as University of New South Wales, Genomics England and St George’s University of London have developed courses focusing on genomics, however the effectiveness of these is yet to be determined.

The focus group highlighted the need for resources and information that are suitable for varying disabilities – visual, hearing, cognition and different learning styles. An example is the US Talking Glossary produced by the National Human Research Genome Institute (<https://www.genome.gov/glossary/>). The National Disability Insurance Agency (NDIA) has produced a useful document for the rollout of their National Disability Insurance Scheme (NDIS) that people have found to be easy to follow, especially for those of varying cognition levels (https://www.ndis.gov.au/html/sites/default/files/documents/Service_Agreement.pdf).

RECOMMENDATION: Adaptable resources should be accessible to people with varying disabilities

CONSENT

Informed Consent

In Australia, Informed Consent refers to *“consent to medical treatment and the requirement to warn of material risk prior to treatment”* as defined by the Equality, Capacity and Disability in Commonwealth Laws Discussion Paper 2014 (ALRC DP81, 10.48). It is the duty of care of health professionals to provide accurate information on the medical treatment, including all associated risks.

The literature investigating appropriate consent models for genome sequencing is vast and divided (Caulfield et al 2003, McGuire et al 2010, Appelbaum et al 2014), addressing issues of return of incidental findings, how much information to include and how the consent should be delivered. In Australia currently consent forms for genome sequencing vary according to whether the test is offered in a clinical or research setting; in the latter authorisation from an ethics body is required for the form to be used. There are general NHMRC guidelines that can be used to develop consent forms (<https://www.nhmrc.gov.au/book/chapter-2-2-general-requirements-consent>).

From our survey it was clear that patients want to be able to:-

- Choose when and how they receive information on Incidental findings
- Participate in research and also choose the type of research
- Have their genome securely stored once sequenced for future analysis

There was a lively discussion about consent processes in the focus group. Some important points that came out of the discussion were:

- Patient friendly language – participants found that some of the language used in consent forms and by health professionals was technical and medical, and often require further clarification. An accompanying glossary may help with this issue, or an information form.
- Suitable for varying disabilities – this issue was elaborated on in the *Resources* section, pg. 23
- A copy of the consent form and resources for themselves – whether hard or soft, to take home, read over and have on file
- Adequate time – to take in the information, especially before appointments with healthcare professionals, opportunity to have a few days to consider the information and follow up
- Opportunity to ask questions – this is important for patients when they would like to clarify details and to ensure they have all their concerns addressed
- Continuity – patients would like to be in contact with the same person throughout the whole process. A trust relationship is important and patients often feel comfortable talking to someone who knows the background of the patient and family (don't have to go over medical history every time with a new person; can lead to misinformation and personal trauma).

There was also some discussion about the possibility of consent processes on multiple platforms such as online. Consent forms can be viewed online, along with accompanying information, which can help with making sure patients have access to the forms when needed. It is important that consent processes clearly state the benefits and risks of genome sequencing, acknowledge the patients' "ownership" of their genomic data, clearly outline whether they will be included in research and what for.

"Sometimes people might feel pressured just to sign them [consent forms] just to get them out of the way and move on to the next thing. I think it's so important to go through it with a genetic counsellor and actually make sure people understand what they're signing cause it's not always easy to understand the scientific jargon that goes with them"

RECOMMENDATION: Ensure consent processes are informative and give patients enough time to process and understand the implications of genome sequencing

Dynamic Consent

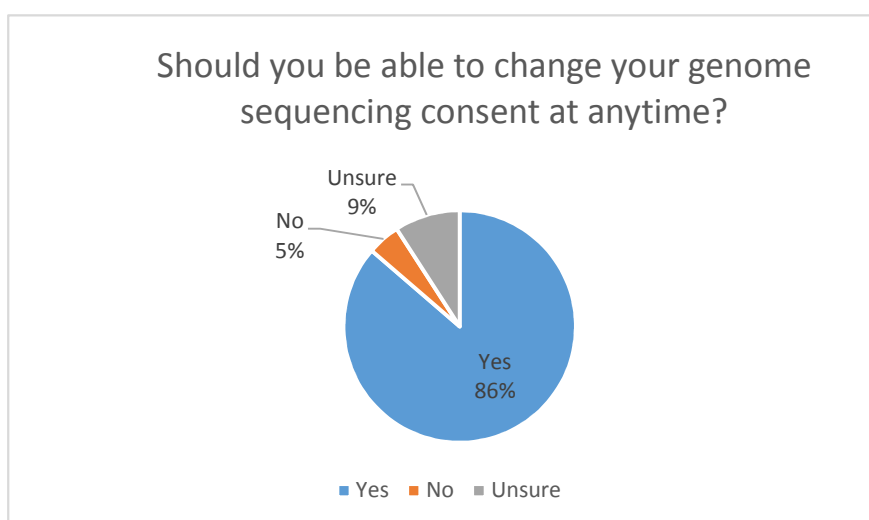


Figure (vi) Changing consent for genome sequencing

Dynamic Consent (Kaye et al 2015) is a new approach for engaging individuals about the use of their personal information. It is also an interactive personalised interface that allows participants to engage as much or as little as they choose and to alter their consent choices in real time.

- It allows the same samples/information to be (re)used with the knowledge and consent of the individual.
- It enables individuals to give and revoke consent to the use of their samples and information in response to their changing circumstances.
- It provides a record of all transactions and interactions in one place.
- It allows people to be approached for different kinds of consent or to obtain their opinions as new research projects are started and new ethical questions arise.
- Consent preferences can be modified over time.

“As it is my information I should always have the prerogative to change my mind about what information I would like to be given”

Patients would like the ability to change their consent concerning genome sequencing at any time. This means they would like to choose whether they continue to receive incidental findings, continue to have their genome analysed (regardless of whether a diagnosis has been found or not), and choose whether they would like to be included in research or not. Examples of when this may be relevant is when a patient decides they no longer want to receive incidental findings in future reanalysis of their genome. With

advances in science moving much quicker than ever before, it is anticipated that new variants will be found at an increasing rate, many of which may be associated with particular conditions. Those who opted to not receive any incidental findings may later decide that they would like to be tested for any new variants related to their suspected condition(s).

RECOMMENDATION: Dynamic Consent should be the standard model of consent used for clinical genome sequencing practice

DISCLOSURE OF FINDINGS

Incidental Findings

There are two types of results that can be obtained from analysing a person's genome:

- Primary findings – relevant to the reasons/symptoms for which the test was ordered
- Secondary findings – results that are not related to the reason for which the test was ordered, also known as Incidental Findings.

Patients/families expressed they would like to be told if incidental findings are present, with 83% of respondents saying if they took the test today they would like to know if there were an incidental findings. Some patients expressed they would like to be told if anything was found, and to be able to have the decision to find out what that is if they chose to do so. The varying answers to this question made it clear that this process is very much an individual decision. It is important that all aspects of genome sequencing are communicated to the patient and that they are included in discussions about what would be suitable for them and how the process will run.

Interestingly, in the UK a majority of patients perceived they would be happy to receive any incidental findings regardless of whether the condition is life threatening (Hazelton & Petchey 2015).

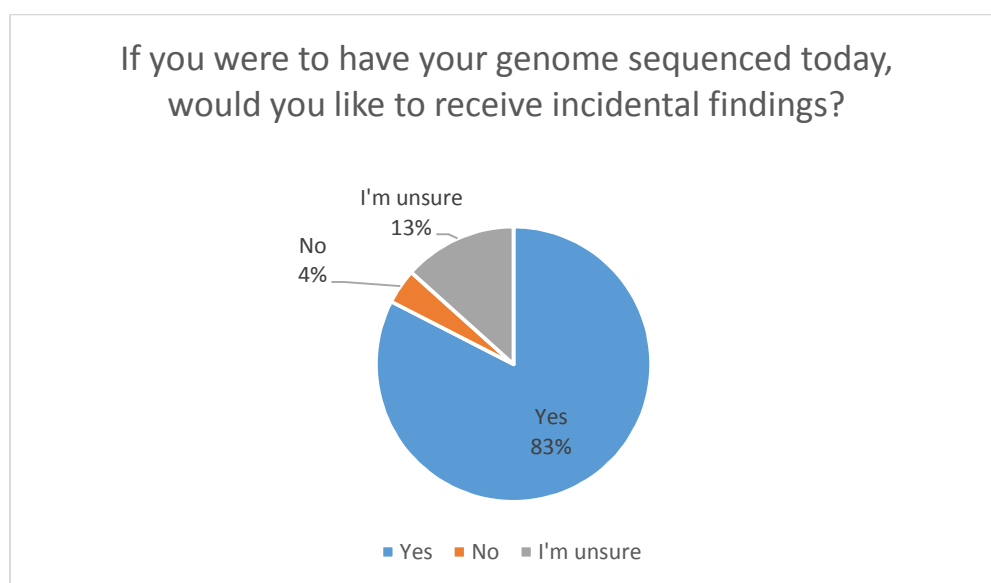


Figure (vii) Receiving incidental findings

Table 4. The percentage of respondents who would definitely like to receive incidental findings based on the condition

	Non-life threatening	Life threatening
Treatable	71%	69%
Not Treatable	55%	46%

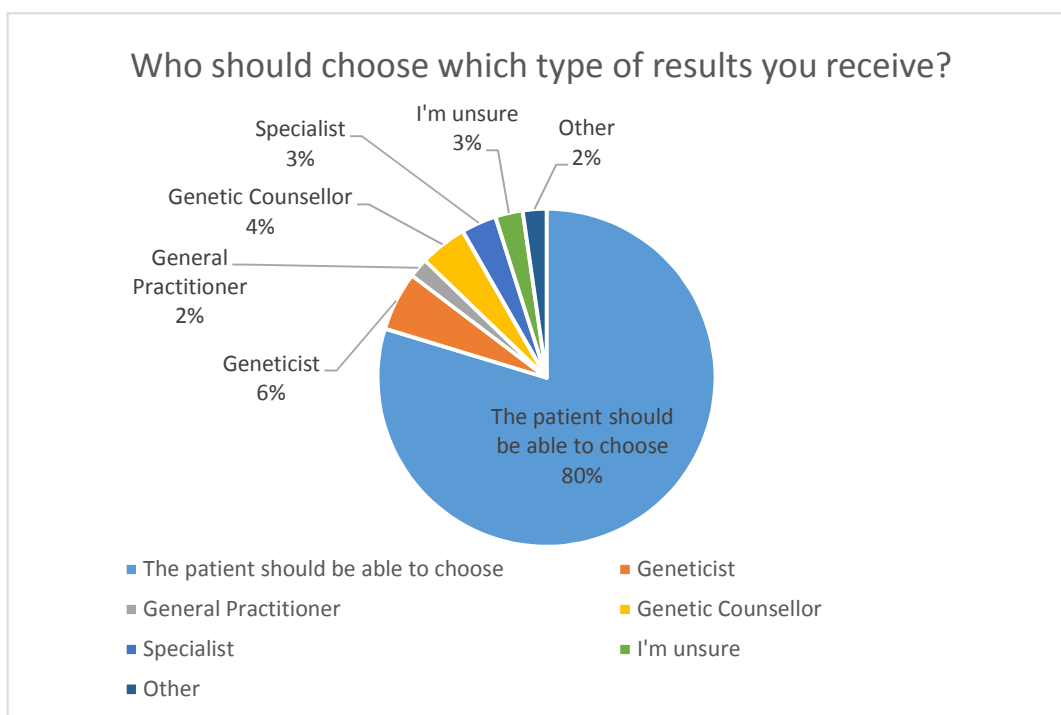


Figure (viii) Who decides the type of results received

"I would still want to know...new therapies are being formulated and tested all the time. If a therapeutic approach became available that could potentially alleviate my condition I would want to be able to access that treatment as quickly as possible. If there was no treatment or cure I am able to plan and prepare in what time I have left regardless of how long that may be"

"One has the right to know. This allows for life planning. It's my body. I want to know"

"My thinking would be around prognosis and being able to spend quality time with my family. Laying down significant and enjoyable times as I navigate through this phase of my life"

"It depends on each situation and the signs and the symptoms. I have recently found out my son has a neurodegenerative condition with no treatment and as painful as it is, I am glad we know. Sometimes not knowing is worse"

When asked about who should choose what type of results they should receive, 80% of patients believe it is up to the patient to decide. There were some comments that mentioned that it should be a team approach and healthcare professionals should give appropriate guidance enabling patients to choose. It was clear from the focus group patients wanted the choice, rather than clinicians making the choices for them without consultation. This shared decision-making process is extremely important in building a trustful working relationship between clinicians, researchers and patients. From the comments made it is important to stress that the return of results, and how they are returned should be a discussion involving the patient themselves. While clinicians are best placed in determining what information is relevant and clinically suitable to return, it is up to the patient to

decide what information from their genome they receive and when. Establishing a relationship of trust between clinician and patient is paramount. This is an area for further research.

RECOMMENDATION: Patients would like to be part of a shared decision-making process about what incidental findings they receive, when and how they receive them

Delivery of results

When considering the return of results to patients, it is expected that genetic counsellors, geneticists and any relevant staff in genetics clinics will be best placed to deliver these results.

Given that genome sequencing is anticipated to have an impact on many areas of health services, it is not unreasonable to expect that other health professionals may be in a position to deliver and interpret genetic and genomic information to patients and families. It is important that those presenting the findings to patients are properly trained to do so. GPs in particular will play an important role in communicating results to patients and families, and explaining how the result may impact on their management.

RECOMMENDATION: Clinicians and genetic professionals should be trained in delivering genomic results to patients, and have a knowledge of referral pathways

It has been reported that many patients feel they do not receive any, or enough information, at the time of diagnosis (Molster et al 2016). This could be information about the condition, what this means for them/their child, or continuing support services to contact. Clinicians need to provide and make available more information for patients and point them in the right direction. The patient's current health status could affect their ability to take in information. It can be a very stressful time for patients and this should be considered very carefully when explaining tests and results to patients. It is Genetic Alliance's experience in dealing with families over the past two decades that often patients cannot recall what tests they have and haven't had done in detail, and what the results yielded. Clinicians should be aware and perceptive when delivering information to patients, taking note of how the patient appears to be taking in the information and give them time to process.

RECOMMENDATION: Appropriate resources, information of patient organisations and options for further support should be offered to patients upon receiving their results

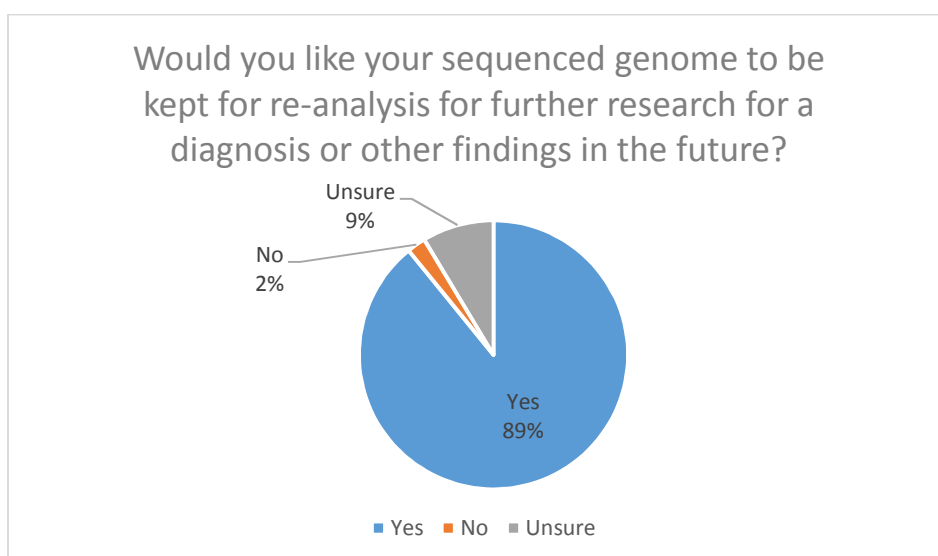


Figure (ix) Re-analysis of genome

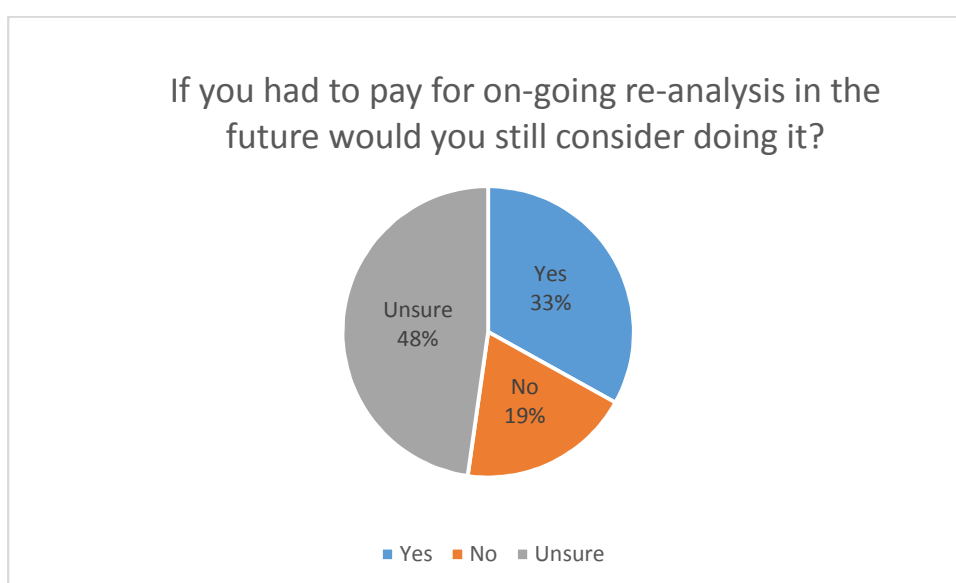


Figure (x) Paying for re-analysis

A majority of patients (89%) would like their sequenced genome to be kept for regular re-analysis. However this depends on whether there is a cost involved. Patients believe they would pay for re-analysis if they wanted to find out any additional information. Comments from the question suggested that some participants believe it should be free for those who had not yet been given a diagnosis. It is not clear if re-analysis is offered in current genome analysis in Australia, and if it is, how often a person's genome is re-analysed.

"I do understand by having your genome sequenced you may not get results at this time so I would want it to be re-analysed as science and technology advance to hopefully be able to re-interpret the results"

“Without a diagnosis I have limited information, support and funding. The future is unknown. I have children who are reaching adulthood where they may soon want to have families of their own. I believe they have a right to know all the information to be able to make an informed decision regarding genome sequencing”

Technology has surpassed what science can interpret from it, meaning that there is much more research to be done. Research on genomics is progressing fast, and new variants are being found. The clinical significance of these variants is also being investigated. For undiagnosed people, re-analysis of a person’s genomic information is paramount with the amount of new variants being found and information about how these variants will impact on clinical care.

RECOMMENDATION: Obtaining a diagnosis should be a priority when genome sequencing is used in clinical practice and this should be reflected in the resources allocated to re-analysis

GENETIC COUNSELLING AND CONTINUING SUPPORT

When GA posed the question of whether patients/families had received genetic counselling the results were split. It appears that many are unsure about what constitutes genetic counselling and who provides it. This is consistent with research done in this area (Maio et al 2013). Research also suggests that people are unsure what to expect from genetic counselling, and this impacts on the way they respond to the sessions (Maio et al 2013, Bernhardt et al 2000, Metcalfe et al 2007).

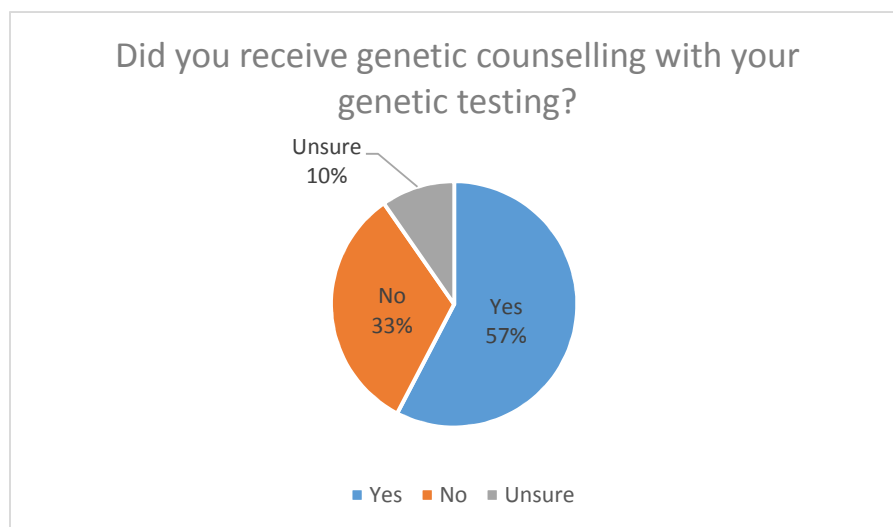


Figure (xi) Receiving genetic counselling with genetic testing

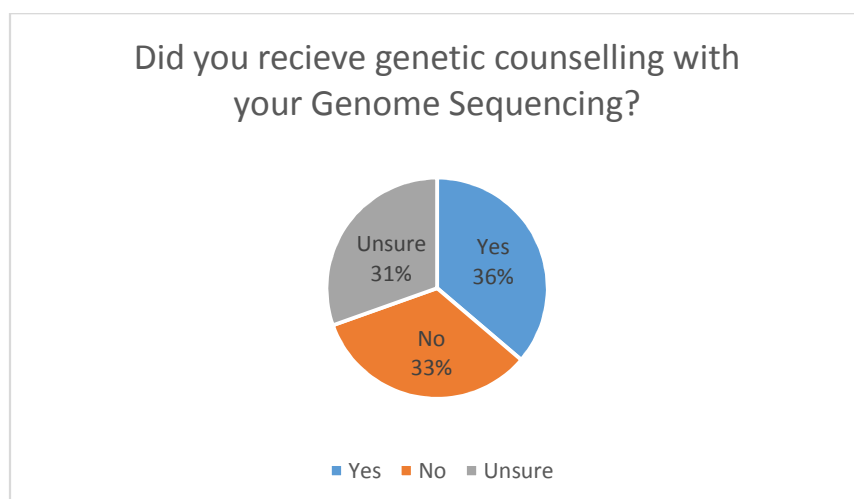


Figure (xii) Receiving genetic counselling with genome sequencing

Genetic counselling is provided by a team of health professionals who work together to provide an individual or family with current information and support regarding problems in growth, development and health that may have a genetic basis. This may include genetic counsellors, clinical geneticists and social workers.

A **genetic counsellor** can provide both verbal and written information about the condition and its impact on you and your family, to assist people in dealing with some of the issues that may arise from the diagnosis of a genetic condition. Genetic counselling is not primarily counselling in the psychological sense. Although the genetic counsellor will address emotional and psychological issues raised during a consultation.

Genetic Counsellors are graduate health professionals with specialist training overseen by the Human Genetics Society of Australasia (HGSA) (*Centre for Genetics Education 2016*)

According to the Australasian Society of Genetic Counsellors (ASGC) there are currently 311 active ASGC members in Australasia.

The Centre for Genetics Education provides information about genetic counselling and services. Support groups and clinicians should actively distribute this information. Some of the respondents reported that while they received a diagnosis or were taking part in research regarding genome sequencing, they had not seen a genetic counsellor or been offered genetic counselling. It is important for healthcare professionals to express to their patients the relevance of seeking genetic counselling. Patients and families need to be briefed on what genetic counselling is and why it is recommended when pursuing genome sequencing.

Table 5. Percentage of respondents receiving genetic counselling with their genetic testing or genome sequencing

	Seen a Genetic Counsellor	Not seen a Genetic Counsellor	Unsure
Had genetic testing	58%	32%	10%
Had genome sequencing	36%	33%	31%

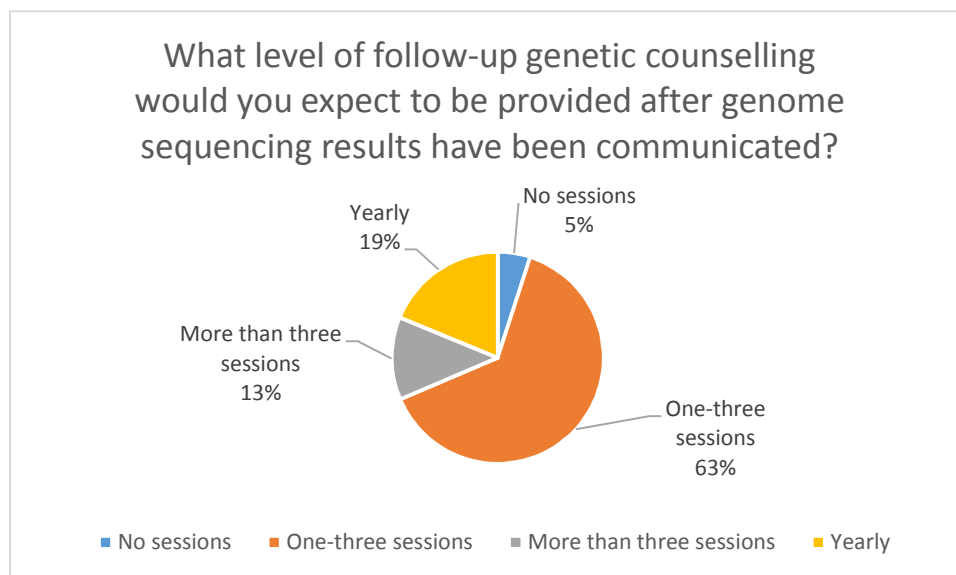


Figure (xiii) Level of follow-up genetic counselling

When asked about how often they would like to access genetic counselling services 63% of respondents said that one-three sessions would be sufficient. Patients stressed it should be up to them to decide how many are relevant and it should be easily accessible. Many were happy to use a service such as a call centre where they may be able to speak to a clinician/genetic counsellor when needed or an email services where they can contact their genetic counsellor when needed and who would reply in a timely manner. Given the complexity of genomic information and the types of results this technology can produce, genetic counselling services will play a very important role in communicating to patients any relevant information and how it will affect them.

"If patients felt that they needed more than this perhaps they could request it. I think one [genetic counselling session] is fine, up to three should be plenty."

RECOMMENDATION: Genetic counselling services should be offered at all stages of the genome sequencing process, even in the case of research, and should be accessible by patients when requested

Genetic Alliance Australia over twenty-six years of operation has observed a marked increase in waiting lists for genetic counselling consultations of over six months. Long consultation wait time places enormous strain on families who want answers. An increased awareness of genetic testing and a changing healthcare model resulting in more involvement from genetic counsellors has placed an increased demand on the workforce (Barlow-Stewart et al 2015). Mainstreaming of genetic and genomic testing as well as increased demand in speciality areas have also been identified as impacting on demand. The Sax Institute

"As genetic medicine and testing technology develop, there will be an inevitable increase in the need for genetic counselling services in Australia...strategies should be developed now to assess and respond to this increased need" – Essentially Yours: ALRC Report 96, 2003

Report on the NSW Genetic Counselling Workforce (2015) identified the following barriers to increasing the genetic counselling workforce:

- limited availability of places in the Masters programs
- limited availability of training supervisors
- employment requirements for HGSA Board Certification
- limited employment opportunities that meet requirements for HGSA Board Certification
- limited numbers of certified genetic counsellors who can provide supervision for training

RECOMMENDATION: More support and resources should be given to expand the genetic counsellors' workforce

RESEARCH

Participation in research

When GA surveyed respondents about participating in research, 87% said they would like their sequenced genome to be made available for research. Patients are very keen to contribute to scientific advancement, especially if it means that other families will have a better outcome or prognosis as a result of the research. It is clear patients would like to be involved in deciding what research they contribute to from a sense of ownership of their genome.

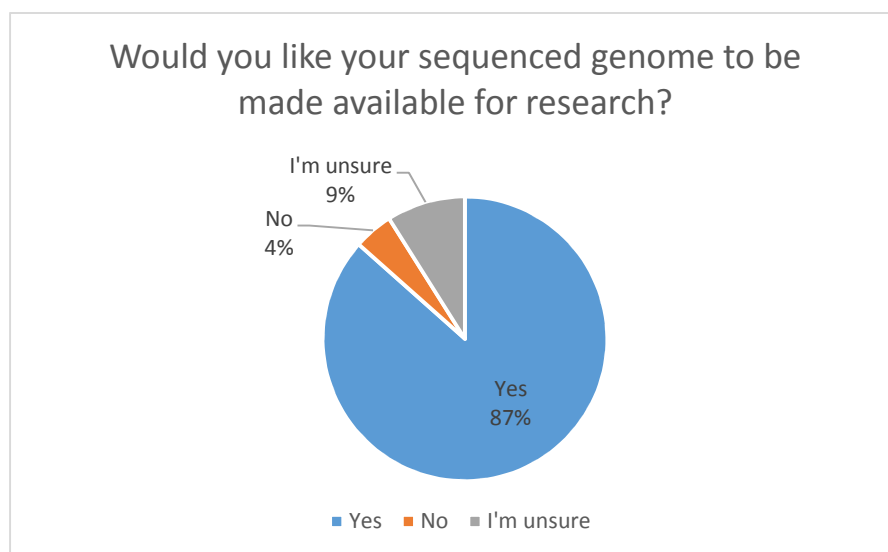


Figure (xiv) Making genome available for research

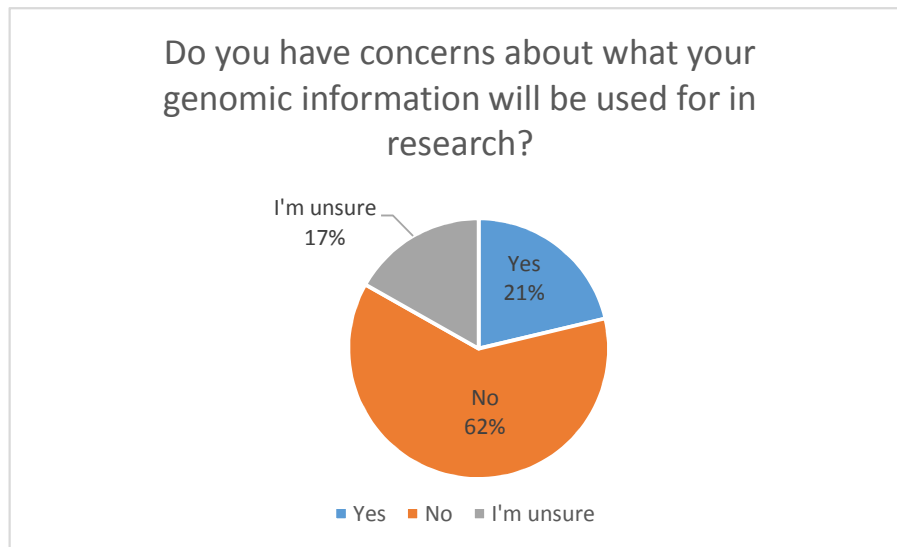


Figure (xv) Concerns about use of genome in research

Sixty-two percent (62%) of respondents said they had no concerns about what their information would be used for in research. Of the 21% who had concerns, the main concerns were that the patient should be contacted before having their genome analysed in a research project, and that their privacy might not be respected. This idea of privacy is associated with prevention of misappropriation of information. Many of the comments stipulated that they would have to be assured that their results would not be identifiable and that they would be stored securely and safely. For some respondents, their decision to contribute to research was dependent on the aims and ethics of the research.

"Depends on who is undertaking the research, what the research is for and whether the information is de-identified"

"I would not support all causes such as unethical ones"

"Have concerns about people making money from my genetic information"

In Australia research undertaken can also be contributed to consortiums, which can span many countries. Research findings can potentially be sent overseas to be aggregated into large research projects and under different protocols to those in Australia. This is a complex issue of being involved in offshore research and it should be clearly stated within the consent process. Patients should be informed if their research is part of a consortium, and if it will be governed by international laws regarding research.

RECOMMENDATION: Research aims, objectives, stakeholder involvement and access to data should be made clear to participants

"I feel you need to still be asked for consent each time your information is needed to be used"

When asked about whether they would like to choose what research projects they contributed to the results were split, with 46% saying yes and 46% saying they don't mind, and 8% were unsure. These reasons for having the ability to choose centred around making sure research was done

specifically for a condition they might have, or if the results might relate to them in any way. Again, the notion that patients should be contacted to obtain their consent before having their genome provided for any research project was prevalent.

As with clinical genome sequencing, the majority of respondents (84%) thought they should be able to opt in and out of scientific research. If patients are to be contacted every time a relevant research project appears, it would be counterproductive to give them the option to opt in and out of a general consent form to address research. Alternatively the initial consent form can address whether the patient would like to be notified of any relevant research projects they may like to contribute their genome to in the future, and they should have the ability to opt in and out of this.

"As long as information is de-identified or restricted to a few authorised by the patient who has the opportunity to provide consent on each occasion"

RECOMMENDATION: Consent must be sought before a patient's genome is used in research projects

Return of results

The return of results from research is not compulsory, but it is generally agreed as the ethical thing to do. The results disclosed to participants is summary data, once all the measurement data has been combined. The disclosure of research results is addressed in the consent form presented to the patient. In genomic research, there is a potential for incidental findings of individual participants to arise when analysing individual genomes. Currently there is no legal obligation for researchers to disclose these incidental findings to participants (Souzeau et al 2016). There is a growing body of literature that argues clinicians and researchers have an ethical obligation to disclose these findings to patients, after considering their clinical validity, utility and actionability, as well as future legal ramifications (Pike et al 2014, Johns et al 2014).

Eighty-six percent (86%) of respondents believe they should receive feedback from the research, whether it be research results, incidental findings or potential diagnosis for the primary indication. Overwhelmingly it was seen by participants as a patient's right and duty of the researcher to return this information to patients. This idea of patients being more involved in the research they contribute to is very prevalent, and should be recognised.

It is clear that patients would like to receive any information they can from the research. This should be addressed on the consent form and there should be a clear indication of what types of results will be disclosed (i.e. summary data, incidental findings etc.). This should be discussed with the patient.

In the Genomics England 100,000 Genomes Project, patients can request to receive their genome on are able to receive their genomes on CD's and USB's as long as they provide the storage devices; there is a strong belief that the data is theirs and so they would like a copy of it.

RECOMMENDATION: Guidelines should be developed to ensure an adaptable approach towards delivery of research findings to patients

Who conducts the research

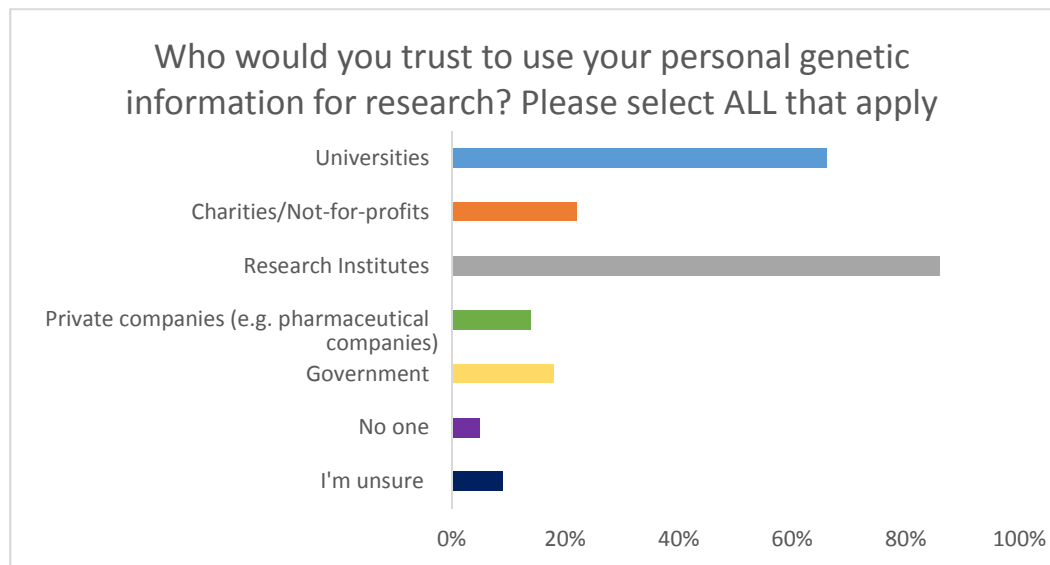


Figure (xvi) Who do respondents trust with using their genomic information?

Scientific research can be carried out in many different contexts by very different bodies. From the survey it was clear that research institutes (87%) were the most trusted bodies to carry out research, followed by universities (65%). Government and private companies such as pharmaceutical companies were the least trusted.

“Don’t think I would want private companies or governments or other organisations using it. Mainly because they may have their own agenda as to why they were researching certain things. Research institutes and universities would have less to personally gain or even worse, use it against me or my family in the future based on what they may find out”

RECOMMENDATION: Genomic research should be conducted by reputable organisations who will be held accountable for their actions

Data Storage, Privacy and Security

The main concern for these respondents when considering genome sequencing was regarding security of their personal genetic information (39%). When asked what was of importance to them before undergoing genome sequencing, 73% of respondents said that they wanted to know who would be able to access their information. In genomic research, a person’s genome will never be fully non-

identifiable as everyone's genome is unique to them. A genome will not be immediately identifiable if external indicators such as name, label or address are removed. There are three ways in which genomic information can be collected, stored and disclosed:

- Individually identifiable – this means the person's identity is connected to the genome data. This could include name, image, date of birth or address
- Re-identifiable – this means that identifying information has been removed from the sequenced genome and replaced with a code, but it remains possible to re-identify the genome by using this code
- Non-identifiable – this means that any identifying information has been permanently removed from the sequenced genome, which means no one can identify the person immediately by looking at the sequenced genome

Participants were amenable to have their genome either re-identifiable or non-identifiable.

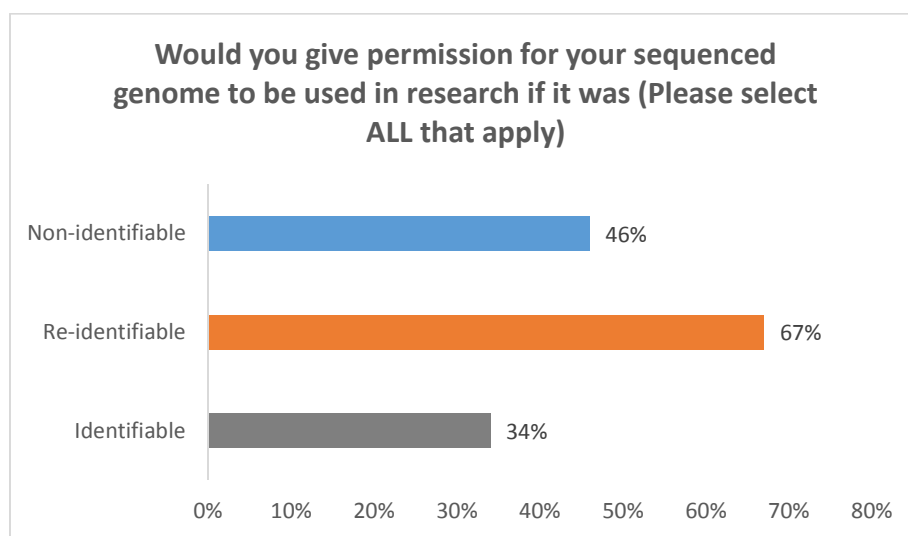


Figure (xvii) Level of identifiability for data storage

It has been shown there are certain ways that people's genomic information can be identified (Meller 2015). Various identification studies have recommended researchers explain that absolute privacy cannot be guaranteed in the future, and that patients may compromise their privacy if identifying information about themselves is easily accessible (e.g. use of social media platforms). If patients are informed of how their information will be used and given confidence in the privacy and security measures taken to store their genomic information, they will be happy to conform to scientific protocol. For various reasons research projects have varying levels of identifiability. The greater the transparency, the greater the participation, the greater the benefits. Rather than guarantee the privacy and anonymity of patients, it has been recommended that patients are better educated during the consent process on the potential for re-identification and how they can maintain their anonymity.

RECOMMENDATION: Researchers must ensure all patient data is securely stored

COST

Clinical genome sequencing is being rolled out in Australia, with the establishment of the Genome.One, a subsidiary of the Garvan Institute of Medical Research in July 2016, and the opening of the St Vincent's Clinical Genomics Unit, Sydney, in October 2016. Currently patients accessing Genome.One's services will have to pay approximately \$4,000 to get one genome sequenced, and approximately \$10,000 if they are to get themselves, their mother and their father sequenced. This is called a trio, and is required to see whether a mutation is de novo (new) or Mendelian (inherited from their bloodline). Hospitals who choose to offer this service to their patients, such as the Clinical Genomics Unit in St Vincent's Hospital, would have to make the decision as to whether the hospital pays for genome sequencing, or whether the cost is passed down to the patient. Currently, those taking part in research will receive genome sequencing free of charge, as the research grants pay for the sequencing.

It was clear from our survey not many people are aware of how much genome sequencing will cost them. This is a hard question to answer, as the cost will differ with each patient, depending on if they are going through a research project or if they are receiving it as part of their clinical care. Many patients believe they should not have to pay and others understand that with a new technology, it is mostly likely the case that it will cost an amount of money.

Many of the comments suggested the cost of genome sequencing be subsidised or covered by Medicare. The rationale behind this is, that it is more cost effective for the government to put money towards prevention to avoid ongoing costs of treatment. There was also a lot of support for a means tested system, where a person's demographic and social situation is taken into account.

"Although I know the costs need to be paid I unfortunately could not afford to pay for such testing. Yet my family carry this gene and half do not even know or are not willing to pay regular doctors to go and get the genetic test done as every state in Australia is different when it comes to costs. I would hope that the Medicare system would be able to help out and provide this for those of us who require it, especially since it runs in the family and there is a pattern"

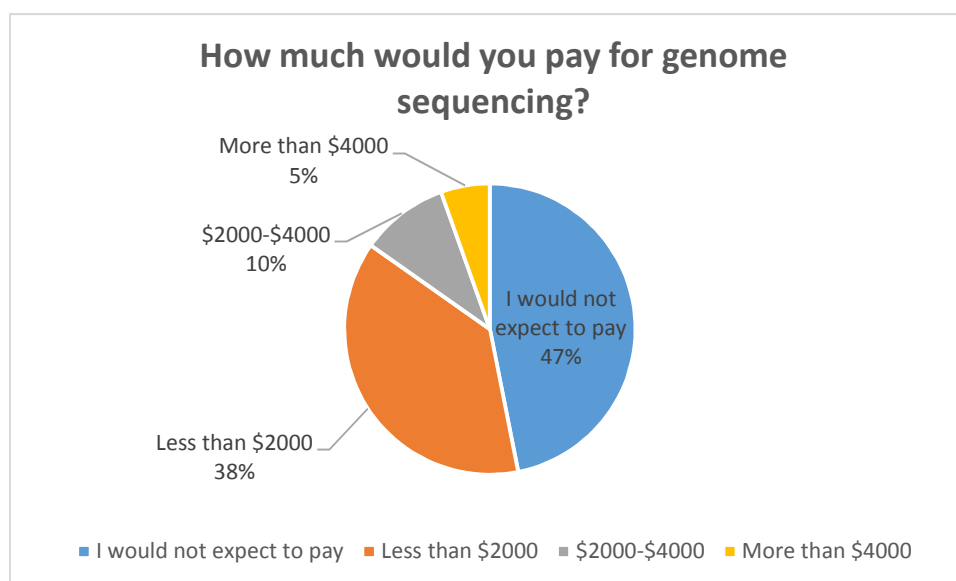


Figure (xviii) Payment for genome sequencing

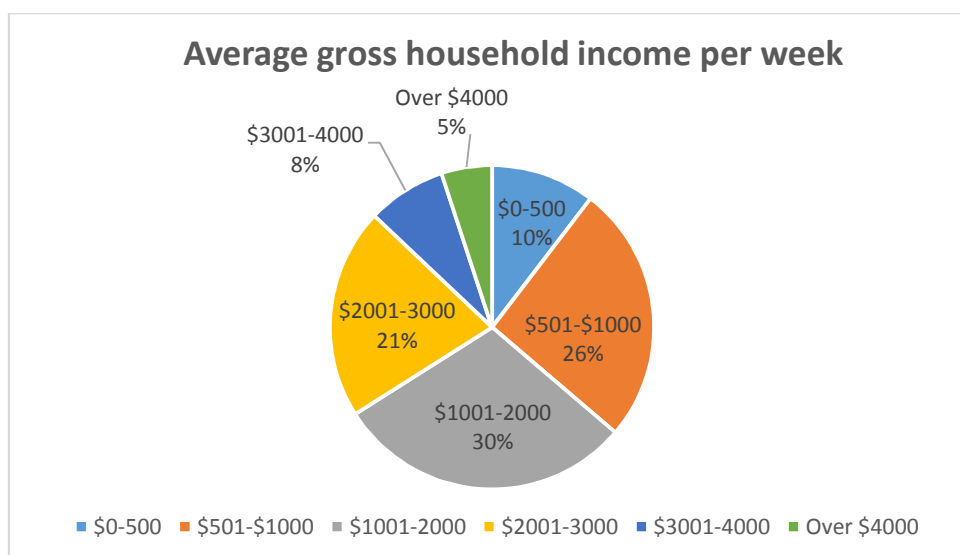


Figure (xix) Average gross household income per week

There was an even distribution on the income scale, demonstrating that the group was not disparate on any particular areas.

"If you know what you are dealing with it would be more cost effective to be preventative than to find yourself with these health problems that could be worse and more expensive"

RECOMMENDATION: Cost and equity of access to genome sequencing through a Medicare subsidy must be a healthcare policy priority

OTHER CONSIDERATIONS

Life Insurance

Genome sequencing is a fast moving area of genetics, and it is unclear what the implications will be for various levels of insurance for patients. How genome sequencing will affect life and health insurance was a high ranking concern for respondents, with 67% and 60% of respondents indicating health and life insurance respectively were important factors to them before making a decision on genome sequencing.

At the moment, private health insurance premiums are not impacted by genetic or genomic test results. This is because health insurance is community rated, meaning health insurance providers offer policies within given geographical areas at the same price to all people, without medical underwriting and regardless of their health status.

With regards to life insurance and genome sequencing, there is a grey area that is yet to be addressed by the life insurance industry in Australia. The Financial Services Council, (formerly the Investment and Financial Services Association Limited - (IFSA)) is the national not-for-profit organisation that represents life insurance companies in Australia. They have a Genetic Testing Standard that regulates the use of genetic tests by life insurance companies. As the standard covers DNA testing, genome sequencing will come under this standard as well.

Under **FSC Standard No. 11 Genetic Testing Policy (2005)**, Financial Services Council Insurance Providers:

- Will **not require** you to undergo a genetic test when you apply for insurance
- Will require that you make available the results of any **previously undertaken** genetic tests upon request
- Will **not** use your genetic test information to assess another family member's risk, for example genetic test information obtained from a parent will not be used to assess an insurance application made by the son or daughter
- Will take account of the benefits of special medical monitoring, early medical treatment, compliance with treatment and the likelihood of successful medical treatment when assessing overall risk
- Will ensure that genetic test results are only made available confidentially to the insurance underwriters and reinsurance companies
- Will provide, to you or your medical practitioner, reason for any adjustment to premiums or policy conditions after assessment of your application

With genome sequencing, there is potential for many variants to appear, however they may have little clinical significance. The worry is that insurers will misinterpret this information without considering clinical usefulness and rule against a potential client based on this information, leading to higher premiums or rejection of application. Research in Australia shows that the fear of discrimination when obtaining life insurance can impact on their readiness to undergo genetic testing (Keogh et al, 2009). There are also studies that have identified a few cases of incorrect risk-assessment judgements based on information from genetic tests (Keogh et al 2013, Barlow-Stewart et al 2009).

"I find it difficult to imagine these areas will not be impacted at some time in the future. Hence, confidentiality and protection of patients' rights are essential"

Another concern, where insurance may be involved is when people take part in genome sequencing research. It is important that they are fully aware of what results are delivered to them, if any, and if personal results are being provided. If this is the case, patients may be required to disclose this information to insurance companies when applying for life insurance. This is also important to consider if the researchers give participants the option to find out personal results, especially if they are looking to take out life insurance. The Centre for Genetics Education also provides some useful information on impact to life and health insurance (<http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/FactSheetInsurance>).

RECOMMENDATION: Life insurance providers should be involved in an active discussion with stakeholders on how to provide for patients who have had their genome sequenced

INSURANCE PROTECTION AROUND THE WORLD

(Durnin et al 2012, Association of British Insurers 2011)

In the United States a Law called Genetic Information Non-Discrimination Act (2008) prohibits genetic discrimination in health insurance and also in employment, but not in life insurance

In the UK, there is a voluntary agreement between the Government and the Association of British Insurers (*Concordat and Moratorium on Genetics*) where the results of a predictive genetic test will not affect a consumer's ability to take out any type of insurance other than life insurance over £500,000. Above this amount, insurers will not use adverse predictive genetic test results unless the test has been specifically approved by the Government. The only test that is approved is for Huntington's disease. This voluntary moratorium has been in place since 2001, and was recently extended to 2019.

In Austria, Article 67 of the Austrian Gene Technology Act prohibits explicitly employers and insurance companies to ask and access genetic information.

In Germany, Article 18 of the Act prohibits the use of genetic information in the insurance context, but for certain types of insurance contracts, e.g. life insurance with a significant premium, the insurer may demand and use results of a previously conducted genetic test.

In Portugal, Insurance companies are not allowed to seek genetic information, and employers only for justified health and safety purposes.

In Sweden, Genetic information can be used for risk evaluation in personal insurance in certain cases under the Genetic Integrity Act

In Switzerland Article 21-25 states that the employer is not allowed to demand a presymptomatic genetic analysis, to demand results of such an analysis if already performed, or to demand a genetic analysis to determine the characteristics of an employee not relevant to health. Exemptions include occupational diseases relevant to a certain position. Articles 26-28 states that an insurance company cannot require genetic analyses as a precondition for an insurance agreement. However, already performed genetic analyses can be taken into account in certain restricted cases.

In Brazil, for an insurance company to use a person's genetic information they will need the consent of that person

In South Africa the industry is self-regulated, and relies on the industry's assurance of fairness

In Asia and the Middle East, only South Korea and Israel have banned genetic testing for insurance purposes. No other countries have formulated stances on the issues of tests being used for insurance purposes.

Executive Summary – *Essentially Yours: The Protection of Human Genetic Information in Australia* (ALRC 96, 2003) represents the culmination of a major, two-year inquiry by the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee (AHEC) of the National Health and Medical Research Council (NHMRC). The Report, which contains 144 recommendations for reform, is the product of an extensive research and community consultation effort—the most comprehensive consideration of the ethical, legal and social implications of the ‘New Genetics’ ever undertaken.

Some of the key recommendations were:

- Discrimination laws should be amended to clearly prohibit unlawful discrimination based on a person’s real or perceived genetic status.
- Privacy laws should be harmonised and tailored to address the particular challenges of human genetic information
- As a matter of priority, Australian governments should develop strategies designed to assess and respond to the need for increased and adequately resourced genetic counselling services.
- Employers should not gather and use genetic information except in rare circumstances, for example, where this is necessary to protect the health and safety of workers or third parties, and the action complies with stringent standards developed by the Human Genetics Commission of Australia (HGCA) and the National Occupational Health and Safety Commission (NOHSC).
- A range of safeguards and improved policies and practices should be applied to the insurance industry’s use of genetic information (including family history) for underwriting purposes

Employment

According to the Australian Law Reform Commission, The Disability Discrimination Act 1992 (Cth) (DDA) and the Human Rights and Equal Opportunity Commission Act 1984 (Cth) (HREOC Act) *“are the most relevant pieces of legislation regulating discrimination in employment on the basis of genetic status”*. This means that there are particular acts that protect against the ability of employers to use genetic/genomic information in a way that affects equal employment opportunities.

Genetic testing may impact on an individual’s ability to obtain employment in certain professions. Australian employers are allowed to request genetic information for job applicants and employees, within the limits of relevant privacy and anti-discrimination laws. This information would be used to inform them on a person’s ability to perform the main requirements of the job, or if it relates to the employer’s common law or statutory occupational health and safety obligations. Testing may be in the form of diagnostic or predictive testing as part of a pre-employment examination, or as part of ongoing health surveillance program (these programs monitor whether the job is impacting on the genetic characteristics of the employee). Genetic tests that have already been undertaken by a job applicant or employee may also be requested to be handed over, even if they were for health reasons or through participation in research.

The following legislation has been in force since 5th August 2009:

The *Disability Discrimination and Other Human Rights Legislation Amendment Act 2009** (Cth):

1. Amends the definitions of 'disability' in DDA s 4(1)(j) & the *Workplace Relations Act* to clarify that the legislation applies to discrimination based on genetic status – including a genetic predisposition to a disability (per ALRC R9-3 & Productivity Commission)
2. Prohibits an employer from requesting or requiring information, including genetic information, from a job applicant or employee, except where this information is reasonably required for purposes that do not involve unlawful discrimination (per ALRC R31-3)

Again, there is potential for the results from genome sequencing tests to be misinterpreted by potential employers, especially when it comes to variants of unknown significance. If not properly informed, employers may use genomic test results to justify not hiring specific candidates (Taylor et al 2007). There would need to be specific guidelines regarding the use of genomic information by employers, as well as reinforcement of anti-discrimination principles, especially when considering what exactly do these variants mean and how will they affect someone from performing particular jobs. Some opinion posts suggest that employers may discriminate on the basis of finding certain diseases or personality traits.

It is important that implications for insurance and employment are communicated to patients. This may impact on their decision to undergo testing, or influence the type of results they receive.

RECOMMENDATION: Referral to information regarding patients' rights, and the implications of genome sequencing on employment and life insurance products should be provided as part of genetic counselling and consent processes

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Appendix

APPENDIX A Who has received Genetic and/or Genomic Counselling?

Q13 - Have you or your family member had any experience of genetic testing before? For example, this could be that either you personally have had a genetic test, or that you have consented to your family member having a genetic test?		Q14 - Was your genetic testing done within the Public or Private health system, or as part of a research study?		Q15 - Did you receive genetic counselling with your genetic testing?	
Yes	288	I am unsure	14	I am unsure	3
No	78			No	7
Unsure	17			Yes	4
Blank	3	Private	25	I am unsure	2
				No	9
				Yes	14
		Public	219	I am unsure	16
				No	64
				Yes	139
		Research	27	I am unsure	3
				No	8
				Yes	15
				Blank	1

75% Had experience of genetic testing
 9% Experience was Public Health
 76% Experience was Private Health
 9% Experience was through Research
 5% Are unsure of where their experience was

60% Received Genetic Counselling
 31% Did not receive any Genetic Counselling
 8% Are unsure if they received any Genetic Counselling

Public experience of genetic testing

63% Received Genetic Counselling
 29% Did not receive any Genetic Counselling
 7% Are unsure if they received any Genetic Counselling

Private experience of genetic testing

56% Received Genetic Counselling
 36% Did not receive any Genetic Counselling
 8% Are unsure if they received any Genetic Counselling

Research experience of genetic testing

56% Received Genetic Counselling
 32% Did not receive any Genetic Counselling
 8% Are unsure if they received any Genetic Counselling

Q16 - Have you or your family member had any experience of genome sequencing before? For example, this could be that either you personally have had your genome sequenced, or that you have consented to your family member having their genome sequenced?		Q17 - Was your genome sequencing done within the Public or Private health system or as part of a research study?		Q18 - Did you receive genetic counselling with your genome sequencing?	
Yes	84	I am unsure	14	I am unsure	3
No	236			No	9
Unsure	58			Yes	2
		Private	6	No	4
				Yes	2
		Public	60	I am unsure	4
				No	17
				Yes	39
		Research	2	Yes	2
		Blank	2	No	1
				Blank	1

22% Had experience of genomic testing
71% Experience was Public Health
7% Experience was Private Health
2% Experience was through Research
17% Are unsure of where their experience was

54% Received Genomic Counselling
37% Did not receive any Genomic Counselling
8% Are unsure if they received any Genomic Counselling

Public experience of genomic testing

65% Received Genomic Counselling
28% Did not receive any Genomic Counselling
7% Are unsure if they received any Genomic Counselling

Private experience of genomic testing

33% Received Genomic Counselling
36% Did not receive any Genomic Counselling

Research experience of genomic testing

100% Received Genomic Counselling



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