Exploring SWAN Parents' Perceptions of Genomic Data

ACKNOWLEDGEMENT OF COUNTRY

SWAN Australia would like to acknowledge the Wurundjeri people of the Kulin Nations as the First Peoples, Traditional Owners and Custodians of the land and waters where we work and pay our respects to Indigenous Elders past, present and future. We recognise that sovereignty has never been ceded, and it always was and always will be Aboriginal land.

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ABOUT SWAN

SWAN Australia provides information, support, connection and systemic advocacy for families caring for a child with an undiagnosed or rare genetic condition. We are the national peak not-for-profit organisation representing approximately 2500 children born without a diagnosis yearly and around 1 in 12 children born with a rare genetic condition. This equates to about 350,000 children across Australia at any one time.

Of the children who present to a geneticist, 30 - 50% will receive a diagnosis. This is especially difficult if the child has a regressive or life-limiting condition.

SWAN increases community awareness and understanding of the impact and prevalence of undiagnosed and rare genetic conditions. We reduce the isolation and emotional strain of raising a child with health conditions and/or disability by helping SWAN families connect. We provide parent information sessions, peer support events and social networking opportunities where SWAN families can form lifelong bonds. SWAN advocates for improved disability supports free and equitable access to genetic and genomic testing, and increased research funding to ensure more children can be diagnosed.

We provide a public voice for our families, campaigning for better community education and improved resources and pathways so that SWAN children can thrive.

ABOUT THE STUDY

Exploring SWAN Parents' Perceptions of Genomic Data was an informal study conducted by SWAN Australia in October – November 2022. In response to data breaches in the media, we felt it was important to explore our community's perspectives about genomic data ownership and use.

Our informal study was based around the concept of data sovereignty - the need for consumers to have greater choice and control over who can access their genomic samples/data.

This study has not undergone ethical approval. SWAN CEO Heather Renton presented the preliminary findings of this study at the Human Genetics Society of Australasia conference in November 2022 to provide the genetics community with a snapshot of our community's perspective.

RESEARCH QUESTIONS

Our study seeks to understand the following questions:

- When consenting to genetic/genomic testing for their children, how much do SWAN parents understand about who can access their child's genomic data?
- What preferences do SWAN parents have with regards to their children's genomic data and DNA samples?

METHODS

Exploring SWAN Parents' Perceptions of Genomic Data is a quantitative study, distributed as an online survey through Google Forms. We also include a case study which describes one SWAN parent's experience tracking down their child's genomic data and DNA samples.

The study questions were developed by SWAN staff, the SWAN Consumer Advisory Group (members of the community) and eight professionals who work in the areas of genetics and research help contribute to and edit the survey. There were 15 questions in our survey (excluding questions about demographics), which were a combination of tick-box and yes or no questions.

We recruited a purposive sample of 70 SWAN parents (11% of members) by emailing the study link to our members and posting on our private social media groups. Recruitment did not involve consent, as this was an informal study without ethical approval. Entrants went into the draw for a $50 Uber Eats voucher.

Survey responses were collected through Google Forms over a two-month period (September and October 2022). For analysis, SWAN staff used statistics created automatically by Google Forms.

DEMOGRAPHICS

All participants were SWAN parents, 89% of respondents were mothers (n=62) and 11% were fathers (n=8). Most participants were 40-50 years old and only spoke English at home. About 14% of respondents (n=10) spoke a language other than English at home and one respondent identified as Aboriginal or Torres Strait Islander. The education level of our participants ranged from Year 11 or below to doctoral level degrees, but 86% had completed tertiary studies of some kind, indicating a highly educated group. One member indicated that they had religious beliefs that would influence their decisions about DNA sample collection and storage.

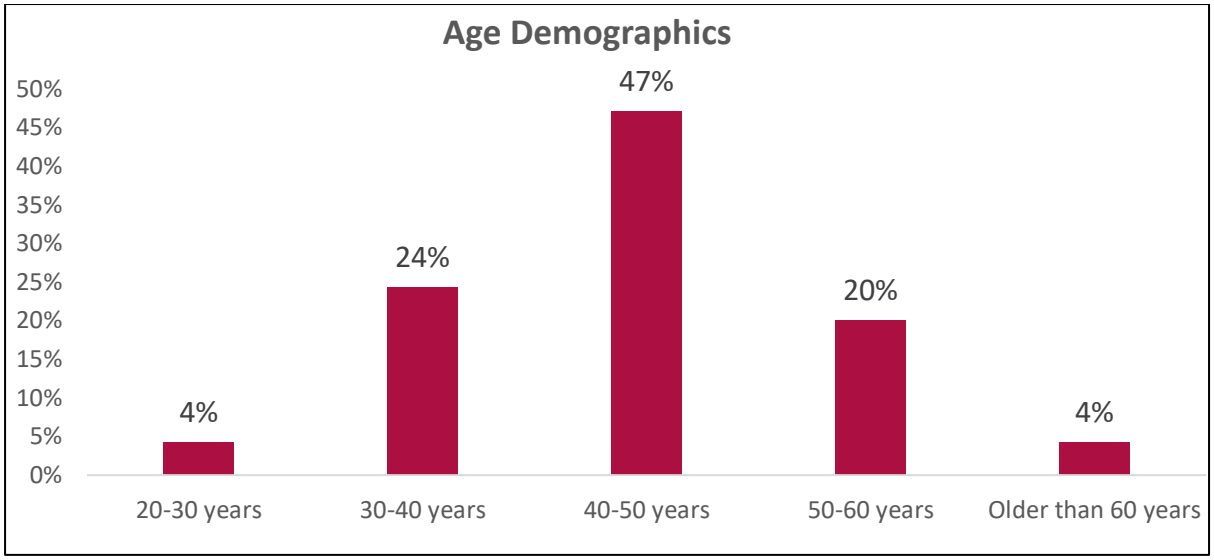

Figure 1: Age demographics of participants

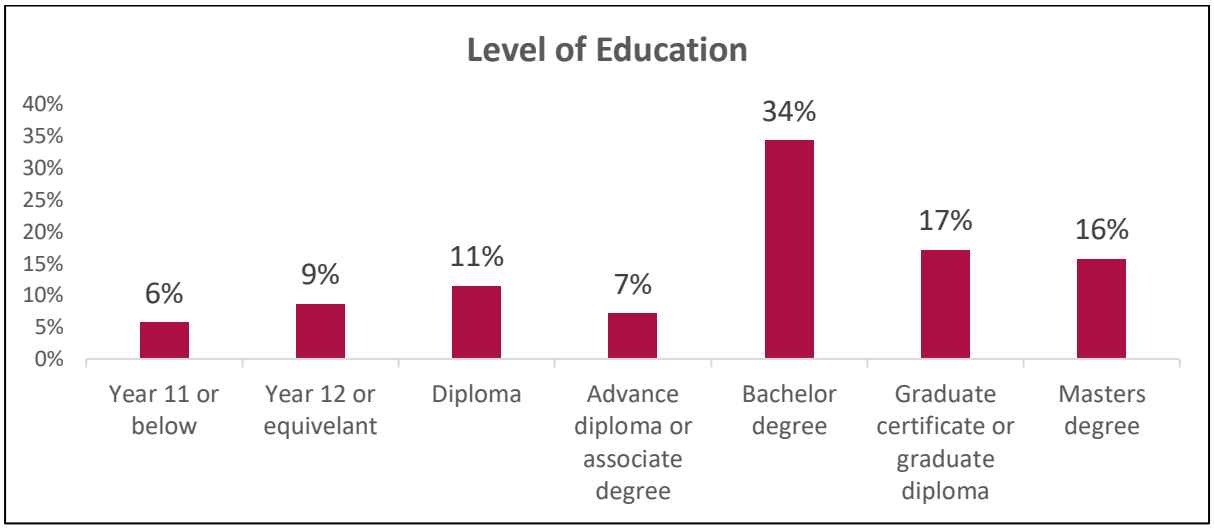

Figure 2: Education level of participants

CONSENTING TO GENETIC/GENOMIC TESTING

TYPES OF CONSENT

We asked SWAN parents whether they understood the difference between consenting to genetic testing in a research and clinical environment. Most respondents could identify the difference and only 3% were unsure about the setting in which they provided consent. We provided definitions about dynamic, specific and broad consent and asked participants about their preferred option.

- Dynamic consent: allows participants to change their consent overtime and facilitates ongoing communication between participants and researchers
- Specific consent: is obtained for each individual research project at the time a DNA sample is taken
- Broad consent: is consent to unspecified future research, either in relation to your child's condition or not related to your child's condition

Dynamic consent was the preferred method (47% of participants), followed by specific consent (30%) and then broad consent (23%). Many parents either did not remember (39%) or where unsure (37%) whether they were given the choice between different types of consent.

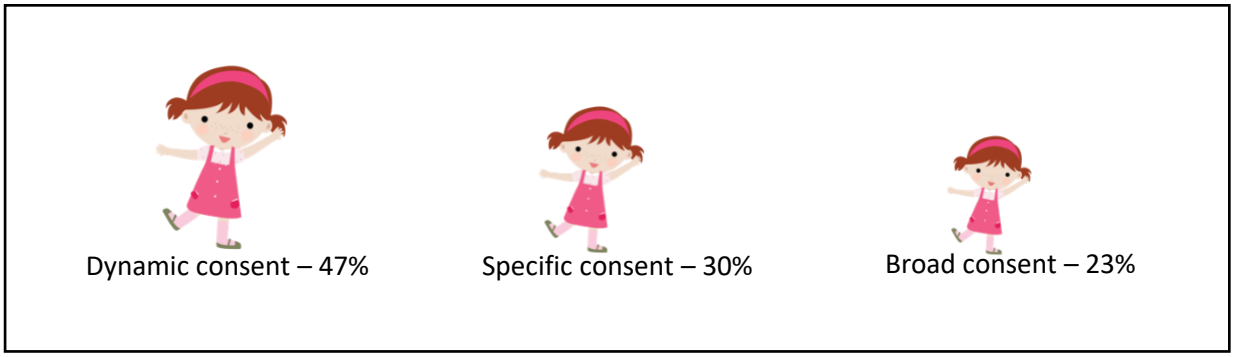

Figure 3: Preferences for different types of consent: dynamic, specific and broad

CONSENT – CIRCUMSTANCES AND FREQUENCY

After genomic data is obtained, parents may consent again for the same data to be used for other reasons, including research projects and clinical trials. We asked parents how often and under which circumstances they should be asked to provide consent again.

Parents generally indicated the need to re-consent when genomic data was being used for research and medical purposes. They felt it necessary to re-consent when:

- New research projects arose (77%)
- New clinical trials became available (76%)
- New information was found about the gene or symptoms (70%)
- New treatments became available (67%)
- When their child developed new symptoms (54%)

Parents felt less of a need to re-consent when their child turns 18 (41%). Some parents were more concerned about the frequency of consent than the purposes for consent. 31% preferred to consent yearly and 17% preferred to only consent once to their child's genomic data being accessed.

SHARING GENOMIC DATA WITH PROFESSIONALS AND ORGANISATIONS

Participants indicated the reasons they would share genomic data with various health professionals and organisations. The number of parents consenting are indicated in Table 1.

Out of the listed people and organisations, SWAN parents were more willing to share information with hospitals, research institutions and GPs than government, pharmaceutical companies and direct to consumer testing companies. In all situations, SWAN parents were most likely to share genomic data with hospitals.

The most popular reason for sharing genomic data was to determine a diagnosis, followed by finding out more information about the condition. Other reasons for sharing data were (in order of popularity): accessing treatments and therapy, connecting with other families and participating in

clinical trials. It's important to note that families did not wish to share information with certain companies and organisations. They had the greatest reservations about sharing genomic data with direct-to-consumer companies.

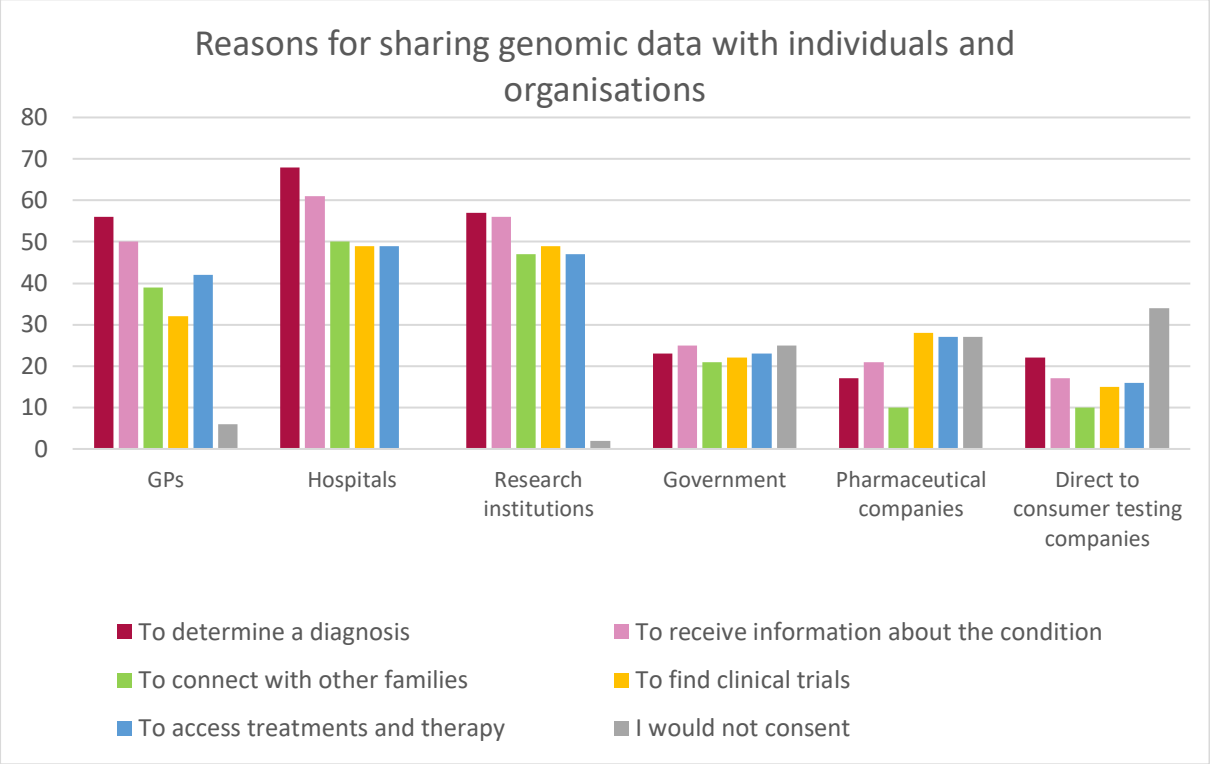

Figure 4: Participant preferences about who they would share genomic data with and for what reasons. The numbers indicate the number of participants.

GENOMIC DATA – OWNERSHIP AND USE

ACCESS TO GENOMIC DATA

We asked if parents understood who would access data when their child's DNA sample was taken. Out of 70 parents, 35 (50%) indicated that they did not understand who would access their child's data and 19 (27%) indicated that they were unsure. We combined these answers to infer that 77% of parents did not fully understand who would access their child's data.

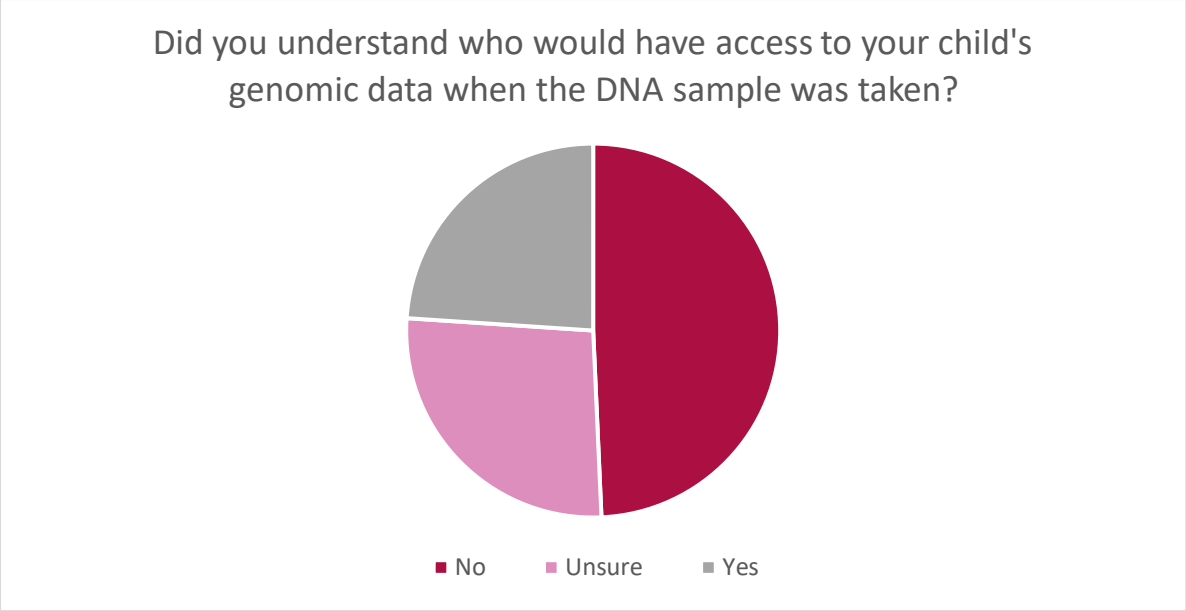

Figure 5: Participant understanding about who would have access to genomic data when the DNA sample was taken

We also asked parents who they think should have access to their child's genomic data. Out of the listed people and organisations. In order, parents believed the following should have access: parents/carers (n=69), children when they become adults (n=59), research centres (n=48), hospitals (n=40), laboratories (n=33), government (n=12), pharmaceutical companies (n=11) and direct to consumer testing companies (n=9). Parents were given the option to comment or add relevant people/organisations. Their answers included genetics doctors, anyone involved directly in the child's care and those involved in raising awareness about conditions.

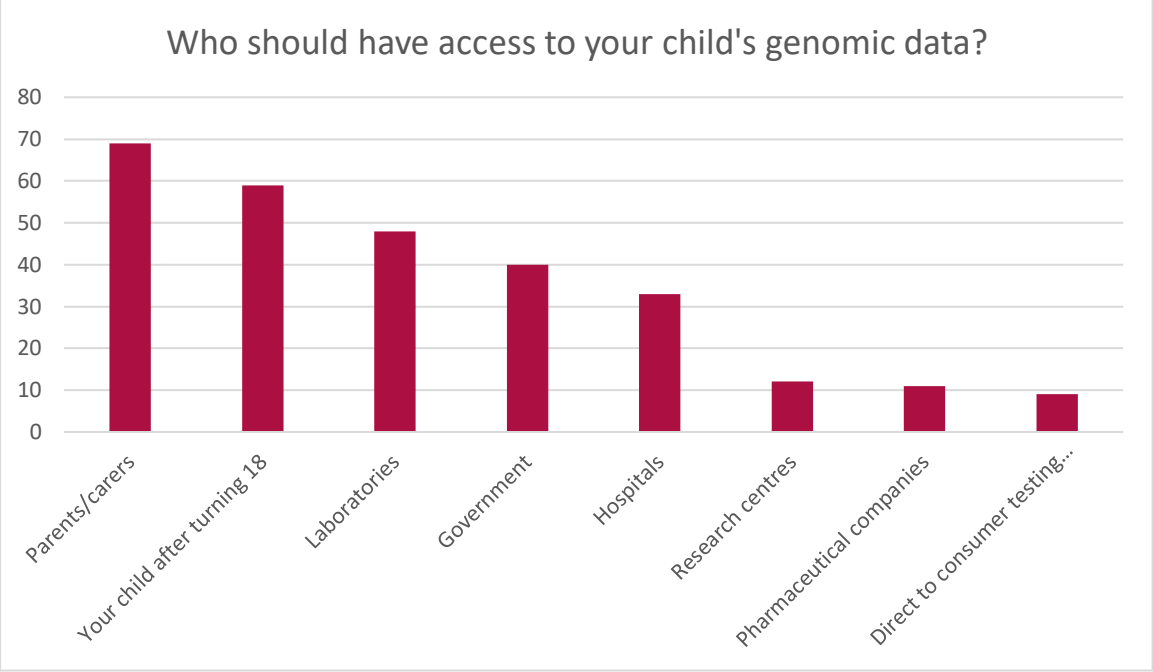

Figure 6: Participant views on who should access their child's genomic data. The number indicates the number of participants.

A significant proportion of parents believe their children should be able to access their own genomic data when they turn 18. A number of these parents (especially in the 40-50 age bracket) have children in their teenage years who are approaching the transition to adulthood. In order to support these families, there need to be efficient systems that transfer genomic data ownership to individuals when they turn 18.

GENOMIC DATA STORAGE AND SECURITY

There were two main questions we explored in relation to data storage and security.

1. What are parents' main priorities in relation to data storage?
2. What makes parents feel confident sharing their child's genomic data?

We allowed parents to select multiple priorities in relation to genomic data storage and security. Our parents indicated that data should be secure (86%), under the control of the parent (64%), private (60%) and easy to access and find (41%).

On a scale of 1-5, parents allocated importance to various factors affecting their confidence in sharing their child's genomic data. In order of most to least important, these factors were:

- Improved patient outcomes
- Reputation of the organisation storing the data
- Communication and transparency
- Privacy laws
- Existing relationships with those storing the data
- Ability to review and revise records
- The data would be shared only within Australia

THIRD PARTY ACCESS TO GENOMIC DATA

When consenting to genetic testing, parents are often desperate for answers, particularly those who have children with undiagnosed and rare conditions. We asked whether parents were concerned that a third party might access their child's genomic data at the time of consent. 47% answered no, 39% answered yes and 14% said they were unsure.

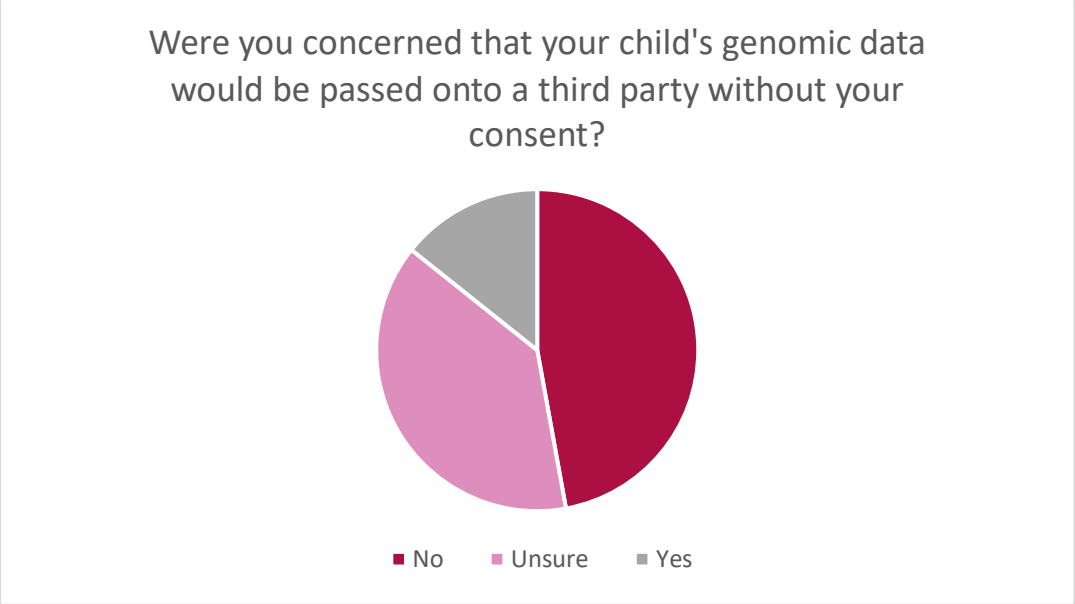

Figure 7: Participants indicated whether they were concerned about third party access at the time of consent.

RE-USE OF GENOMIC DATA

To understand parents' preferences about the re-use of genomic data, we asked whether this should occur in a clinical and/or research setting. Participants suggested that the re-use of genomic data was acceptable:

- For both clinical and research purposes (74%)
- For research purposes only (16%)
- For clinical purposes only (13%)
- Under no circumstances (3%)
- Unsure (24%)

DNA SAMPLES – SECURITY AND ACCESS

CONTROL OVER SAMPLES

What happens to DNA samples after they are analysed? We asked parents if they recalled being asked if they wanted their child's DNA sample destroyed after analysis. Most respondents (76%) did recall being given the option to destroy their child's DNA sample. Most respondents (74%) also indicated that they wanted greater control over their child's DNA.

CASE STUDY ABOUT ACCESSING SAMPLES

One SWAN parent tried to access their child's samples recently. She recently found out that her daughter's samples were sent to four different labs, and at the time she was not aware these labs' policies about DNA sample storage. She was surprised to find that a tissue culture lab at a medical/research institute had established a fibroblast cell from her daughter's skin biopsy. This was just described to her as "growing cells to analyse". She was also unaware that saliva and blood samples were still stored in freezers at the genetics clinic where testing took place. The consent form

was not available on the hospital portal to access and she had not scanned or hard copy of the record she could reference as it was probably destroyed.

Some cells were sent to overseas labs, which were impossible to track down. This parent was concerned about lacking ownership over DNA samples due to different laws overseas. During these independent investigations, the parent discovered that you could request for DNA samples to be destroyed any time after they are taken. Not all parents are aware of this.

This case study reveals issues around communication, transparency, accessibility of information and control over DNA samples. During the diagnosis journey, parents can be anxious and desperate for answers, and so they are vulnerable to uninformed consent. It is also worth noting how difficult it was for the parent in this case study to track down information, despite being well-educated and from an English-speaking background.

STUDY STRENGTHS AND LIMITATIONS

People who had an interest in genomic research were more inclined to be interested in the survey, which may have skewed the data. However, even with an educated audience, we still felt that families were unaware of the difference between raw genetic/genomic data, and their child's genetic result contained in a lab report. Furthermore, this was not an ethically approved study and SWAN staff are inexperienced researchers. We received feedback from our Consumer Advisory Group (CAG) about the readability and accessibility of the survey. However, we did not create a survey that was accessible for people with CALD backgrounds or who had intellectual disability. Strengths of the study were that we had professional input from healthcare professionals and researchers (on a voluntary basis) and we did provide the option for people with disability to call us as an alternative to survey.

CONCLUSION

Our study was inspired by the concept of Indigenous data sovereignty, a movement towards giving Indigenous people(s) and communities more control over what happens to their data, who accesses it, who benefits from it and how it is used.

The majority of SWAN parents indicated they wanted greater control over genomic data and indicated preferences for data to be used in a clinical and research environment. Trust in healthcare professionals and willingness to consent indicates a power imbalance that can be reduced by empowering parents with information. Often these parents are willing to share information for the greater good.

Many parents also believed their children should have access to their data when they turn 18. Future studies could also explore children's preferences as they transition to adulthood.

Parents need accessible information, so they are empowered to make informed decisions around their child's DNA and genomic data. This information should cover the types of consent, who has access to it, re-use purposes, data secuirty and storage and what happens to DNA samples.

Acknowledgment

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