Supplementary File 2. Parent/Guardian Information Statement/Consent Form

chILDRANZ Flagship
Sydney Children's Hospital Network

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Coordinating Principal Investigator	*A/Prof Meredith Wilson (CHW), Dr David Mowat (SCH)*
Associate Investigator(s)	*Prof Adam Jaffe*
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This information statement and consent form is divided into three sections.

1. Information about the research project and genomic testing.
2. Consent to participate in the research.
3. Consent to participate in diagnostic genomic testing.

SECTION 1 - INFORMATION ABOUT THE RESEARCH PROJECT

1 Introduction

Thank you for taking the time to read this **Participant Information Statement and Consent Form**. We would like to invite you and your child to participate in a research project that is explained below.

This form tells you about the research project. It explains all the steps and procedures involved in the project. Knowing what is involved will help you decide if you want you and your child to take part in the research. Please read this Information Statement carefully.

Before you decide if you want you and your child to take part or not, you can ask us any questions you have about the project. You may want to talk about the project with your family, friends or health care worker.

Participation in this research is voluntary. If you don't wish you or your child to take part, you don't have to. Your child will receive the best possible care whether or not you take part.

If you decide you want you and your child to take part in the research project, you will be asked to sign the consent section. By signing it you are telling us that you:

- Understand what you have read.
- Consent for you and your child to take part in the research project.

- Consent to the use of your and your child's personal and health information as described.
- Have had a chance to ask questions and received satisfactory answers.

You will be given a copy of this Participant Information Statement and Consent Form to keep.

2 What is the research project about?

Genes are the instructions inside you that tell your body what to look like and how to work. Genes get passed down in families from parents to children.

Genes are made up of DNA and appear on thread-like structures called chromosomes. Genes are inside almost every cell of your body. There are over 20,000 genes in each of your cells, and together these genes make up your "genome". Each gene has a specific function, however the function of all genes is not yet known. Variations in genetic information in different people can occur, which is why we are all unique. Some variations are harmless, but other variations can make a gene not work, resulting in a health condition. Identifying variations may help to find the cause of your child's genetic condition.

Previously, testing to look for variations in genes involved looking at only one gene at a time. There are now new types of tests that can look at many or all of our genes at once. This testing is known as "genomic" sequencing.

This research is trying to determine the impact of "genomic sequencing" on patient care. The test being provided is a clinical test carried out in a diagnostic laboratory, and the performance of the test itself is not being evaluated.

This type of testing may or may not provide your child with a diagnosis for their condition. Your child's doctor or genetic counsellor will describe the test and research to you.

The aims of this research study are:

a) to look at how providing this test to certain populations of patients with suspected genetic conditions can impact on their clinical management,
b) to determine the cost effectiveness of offering this test, and
c) to evaluate if providing further research as part of this study for undiagnosed patients will provide more diagnoses and further our understanding of your disease.

We will collect a blood and/or saliva sample from your child, which will be used for the clinical test being provided. We will also ask you as a parent for a sample of your blood and/or saliva, as this can assist in determining the cause of your child's condition. If no cause is found in a known gene and you agree, your and your child's samples will be used in research to further understand how the condition occurs by looking at new genes. In some rare cases, we may also collect a skin sample, hair follicles or bone marrow from your child to provide cells that can be used for research as part of this study. We will use the blood and/or saliva samples to extract DNA and look at the genes and how they function. We can use the blood cells, skin cells and hair follicles to provide a constant source of material so recollecting samples should not be necessary. We plan to collect excess tissue if your child is required to have surgery for their condition to look for possible genetic causes.

Advances in technology means we can use new tests to give us more information to better understand the structure of genes and how they work. We may use the skin samples provided to:

- perform functional cellular studies: where we can look at skin fibroblasts (a type of cell that is responsible for making connective tissue) so we can find out more about the structure of the cells and how they work.

- generate an Induced pluripotent stem cell (iPS) line: where we take skin cells and use them to make stem cells which can be re-programmed and turned into another type of cell. This means that we can study the specific type of cell that is not working properly in the affected

individual. For example, if the heart is not working properly we can generate heart cells to try and understand what is wrong. These cells will give us more information about how a genetic change affects the formation of different cell types in the body.

In addition to this, the potential outcomes of genomic testing on families will be assessed. As part of this we would like you to participate in questionnaires on the health, financial, social and emotional impacts of living with a child with a rare genetic disorder, along with questionnaires about the patient/parent/guardian experience of having genomic testing.

We would also like to store any remaining samples and data that we have collected for the purpose of the research outlined in this document. If you consent to this, there is no limit on the length of time that we can store your and your child's sample/s and information.

Together this research will allow us to gather the information needed to determine the most appropriate and cost effective way of providing genomic testing to patients in the future.

3 What is the purpose of this research?

The purpose of this research study is to find out if information obtained from genomic sequencing is useful for the diagnosis and/or management of your child's condition compared to the routine care that is usually offered to people with your child's condition.

The research also seeks to understand your experiences and obtain your views of your child having the test and what you think is important for doctors to consider when offering the genomic sequencing test to people. The cost effectiveness of this type of test for people with your child's condition will also be examined.

This study will also examine if making your and your child's samples and data available for further laboratory research (beyond the first genomic test) within this project, helps us to provide more patients with a diagnosis and understand more about their disease.

Genomic sequencing is not usually funded for your condition and is therefore not widely available. However, the test will be provided to you as part of your participation in this research so the researchers can answer the questions above. It will be provided as part of your care and will be ordered by your doctor and performed in an accredited laboratory.

4 Why am I being asked to participate in this research project?

We are collecting evidence on the usefulness of genomic sequencing for a select group of health conditions. Your child has been asked to take part in the study because they have one of the conditions for which we are trying to determine if genomic sequencing is useful.

5 What does participation in this research involve?

Participation in this study requires your child to provide a blood and/or saliva sample (as advised by the clinician). In some rare cases, we may also collect a skin sample or hair follicles to provide cells that can be used in the study.

We would also like to collect a blood and/or saliva sample from you as a parent, as having your information from your DNA can help to determine the cause of your child's condition and may provide you with information about the possibility of future children having the same condition.

Participation in this study involves having a genomic sequencing test so the impact of this test on your child's healthcare can be investigated. In addition we will collect information about your understanding, views and preferences regarding the test, along with health, social and financial information. Specifically, we would like to:

- collect and store you and your child's DNA (at a diagnostic laboratory) from you and your child's blood and/or saliva sample and carry out a clinical grade genomic test on your child's sample.
- access the stored DNA and you and your child's identified or re-identifiable data, including clinical photos, for research if a diagnosis is unable to be made by the initial genomic test, or where further understanding of how your child's disorder occurs is required. This research may include more in depth genomic sequencing and/or other laboratory studies on your and your child's samples, either in Australia or overseas, to see if a cause can be found for your child's condition. [This research may also require access to other tissues and samples that may be collected as part of your child's standard clinical investigations as listed below. If these samples are required for this research and they are available and are no longer needed for clinical purposes we will contact the laboratory storing them directly. We will provide the clinical laboratory with your consent for your child to participate to this study to access them and you will not be required to fill out another consent form. These tissues include:

  Lung tissue and lung washings (lavage) for lung diseases]

- collect information about your child's condition, the tests your child has had, treatments your child has received, any other health problems and history of related health problems in your family. We will collect this information from your child's medical records, but the researchers may also ask you for more detail if required. This information will help the study doctors interpret your child's results and understand how genomic sequencing may change how care is provided for your child's condition in the future. We may also continue collecting medical information on your child, for up to ten years, from your child's doctors after genomic sequencing is completed to help us determine the long-term implications of our research.

- ask you about your views on genomic sequencing and experiences related to this testing, to help us decide how this testing should be offered in the future. We will ask you to complete two surveys to gather this information; one around the time the genomic sequencing test is ordered and the other no sooner than one month after genomic sequencing is completed.

- offer you the opportunity to participate in focus groups or workshops so that we can ask you questions about your experience of genomic testing.

- ask you about social and financial factors, that will be used help us understand the costs of living with a child affected by a genetic disorder and to determine if the genomic sequencing test is cost effective for people with your child's condition. We will ask you to complete surveys to gather this information; before you receive your child's genomic testing results and the other no sooner than one month after you have received your child's genomic testing results. Examples of the types questions asked may be your ability to work, quality of life, family planning and impact on relationships. This includes personal questions about your relationships and family planning.

- link your child's identifying information (e.g name, date of birth, address) to gather your child's data from hospital and emergency datasets. Throughout our lives, information is collected about our health and health care. Hospitals, health departments and other groups or organisations that provide health services collect this information. The collection of this information is usually required by law and is securely stored by the service or agency that collects it. Health research is very important as it looks at how health care is managed and how services are delivered and used. This study will collect your child's hospital and emergency records that are related to your child's conditions through data linkage. Data linkage is a technique for creating links between data sources (in this case hospital and emergency datasets) so that information that is related to the same person can be brought together. In brief, we will supply the data linkage agency with key fields for linkage, including but not limited to name, date of birth, address. The data linkage agency will then

create a unique ID for your child and send it to hospital and emergency data custodian. Your child's hospital and emergency records will be merged with the unique ID, and be provided to us. We will access information from 2007 (or the date of your child's birth) through to the end of the study, which is up to 14 years. We will then know about the healthcare resources your child has used. This will help us to study the potential economic impact of genomic sequencing in comparison to current standard care. We will collect information about your child from the following databases through third party data linkage agencies:

- hospital datasets
  - Victorian Admitted Episodes Dataset
  - NSW Admitted Patient Data Collection
  - ACT Admitted Patient Care
  - QLD Hospital Admitted Patient Data Collection
  - WA Hospital Morbidity Data System
  - South Australia Inpatient Hospital Separations
  - Northern Territory Inpatient Activity
  - TAS Public Hospital Admitted Patient Collection
- emergency department datasets
  - Victorian Emergency Minimum Dataset
  - NSW Emergency Department Data Collection
  - ACT Emergency Department Data Collection
  - QLD Emergency Department Information System
  - WA Emergency Department Data Collection
  - SA Emergency Department Data Collection
  - NT Emergency Department Activity Collection
  - Tasmanian Public Hospital Emergency Department Presentations

- collect information about your child's Medicare Benefits Schedule (MBS) and Pharmaceutical Benefits Scheme (PBS) data. You will be asked to fill out a consent form authorising the study access to your complete MBS and PBS data as outlined here. Medicare collects information on your child's medical visits and procedures, and the associated costs, while the PBS collects information on the prescription medications you have filled at pharmacies. The consent form is sent securely to the Department of Human Services who holds this information confidentially. This information will only be used by the current study and will not be shared. *Please note that if your child is on both parents' Medicare cards, i.e. not family Medicare card, it is important to obtain signed consent from both parents for us to obtain the complete Medicare record of your child.

- share your child's anonymised data with other researchers. To do more powerful research, it is helpful for researchers to share information they get from studying your and your child's samples. They do this by putting it into one or more restricted access databases, where it is stored along with information from other studies. Your and your child's data and sample/s may also be shared in an anonymised way with other laboratories involved in research for your child's condition, under appropriate agreements with the Australian Genomics Health Alliance.
- ask you to use a website to manage your choices about being in the study and to allow you to engage with the study on an ongoing basis. This website, that can also be used like an App on your phone, is called CTRL. The person who you provide consent to for being in the study will tell you how to register. We are introducing this website as a small pilot study which will tell us whether having more consent choices and ongoing contact with the study is better for you.

Optional Consent:

a) You will also be asked if you agree to share your and your child's data (health related, genomic and self-reported information) and sample/s for use in ethically approved research

outside this study. If you do agree to this you will not be contacted for further consent, and your and your child's data and sample/s will be shared in a de-identified way.

b) We may like to contact you in the future about the possibility of you or your child participating in other ethically approved research projects. This is optional and you can indicate your willingness to be involved in such research on the consent form.

Optional Parental Sample Consent:

As mentioned we would like to collect parental (biological) blood and/or saliva samples at the time of collecting your child's samples if possible. Having these samples can allow us to confirm a finding in your child, determine the possibility of having further children that may be affected by the same condition, or even find a new previously unknown cause for your child's condition. This is optional and you can indicate your willingness to provide your samples individually on the consent form.

There are no additional costs associated with participating in this research project, nor will you be paid.

A summary table of your and your child's direct involvement in this study is provided below:

Visit 1 – Regular appointment Consent	You will have the study purpose and procedures explained to you by your child's treating clinician and will be asked if you are interested in you and your child taking part in the study. You may either sign a consent form on the day of your child's visit or be given the option to provide your consent with a genetic counsellor over the phone or at another appointment. If possible, arrangements can be made for your and your child's samples to be taken at this visit too, or at a later date if it is more convenient.
Patient Experience Questionnaire(s) (pre-test)	You may be sent a letter or email requesting your participation in a survey to allow the researchers to understand your experience of genomic testing and how it may best be provided to patients with your child's condition in the future, to determine the lifetime cost of your child's condition and if offering genomic testing for patients with your child's condition is going to be cost-effective. The survey may ask questions about the social and financial situation of you and your family. You may complete it online or on a paper copy mailed to you.
Visit 2 – Regular appointment Return of results	At a regular appointment with your child's treating clinician the results of the genomic testing performed in a diagnostic laboratory will be provided to you. This may or may not result in a diagnosis. If there is no initial diagnosis found your and your child's samples may be accessed for further research related to your condition to try to find a diagnosis. You will not be required to sign another consent form.
Patient Experience Questionnaire(s) (post-test > 1 month after return of results)	You will be sent a letter or email requesting your participation in a follow-up survey regarding your experience of genomic testing. It may ask whether genomic testing has resulted in any changes in the social and financial situation of you and your family. Again, you may complete it online or on a paper copy mailed to you.

6 Who is funding this research project?

This research is being led by the Australian Genomics Health Alliance (AGHA), a collaboration of major research institutions, universities and clinical services across Australia, and is funded by the National Health and Medical Research Council. The study doctors and researchers are employees of the major collaborating institutions, universities and clinical services.

7 Do I have to take part in this research project?

Participation in a research project is voluntary. Please talk to your clinician about alternative testing options available to you and your child if you decide not to participate.

It is your choice to take part in this research. You do not have to agree if you do not want to. If you give your consent and change your mind, you can withdraw from the project. You do not need to tell us the reason why you want to stop being in the project. If you leave the project we will use any information already collected unless you tell us not to. To opt out, please contact the clinical contact person listed in section 14. If however you have consented for you data to be shared, it may not be possible to retrieve and remove all your data should sharing have occurred prior to withdrawal of consent. If you withdraw from the research before the genomic sequencing test is started, the test may not be performed and will not be funded by the research project You can speak with your doctor about other options for obtaining this test if you decide not to participate in the research.

If you withdraw your child from the research before data linkage to hospital and emergency datasets, the data linkage will not be performed. If you withdraw your child from the research before MBS/PBS information is collected, we will not collect your child's MBS/PBS information. Hospital and emergency datasets, as well as MBS/PBS datasets collected prior to your child's withdrawal will be retained unless you tell us not to.

8 What are the possible benefits of taking part?

The genomic sequencing test is not currently routinely funded for people with your condition. By taking part in this study you will be able to access this new test. The results may confirm or provide a diagnosis and may influence your healthcare. Expert advice and counselling will be available at your hospital to provide you with more information about the possible outcomes of genomic sequencing.

However, there may be no direct benefit to you or your child from taking part in this research, although the outcomes may provide valuable information about whether this test should be part of routine care for patients with your child's condition in the future.

9 What are the possible risks and disadvantages of taking part?

Blood Test
There are no major risks associated with a blood test. It is possible you or your child may feel some discomfort during the blood test. It is possible there may be some bruising, swelling or bleeding where the needle enters the skin. Some people can feel a little light-headed when blood is taken.

Skin Biopsy
A skin biopsy is generally a simple and safe procedure but your child may experience bleeding and/or bruising where the skin is taken. The procedure will leave a small scar measuring 3mm. In some cases, infection may occur. If your child is having another procedure under anaesthetic, we may ask if we could collect a skin biopsy at the same time to minimise the discomfort for your child.

Hair Follicle
A minimum of 20 hair follicles is required for collection. The follicle should be attached to the hair shaft on removal, to enable DNA extraction. Hairs may be taken from your child's head, face or body. Your child may suffer slight discomfort from the collection of these specimens, including a small amount of pain when the hair is removed and some redness in the surrounding skin after removal of the hair.

Tissue collection
If your child is having tissue removed as part of a planned surgery, we would like to access this tissue for this project. As no extra tissue would be removed for this study, there is no extra risk involved in the collection of tissue.

Genomic Test

Genomic testing can raise important issues. Although we do not expect many issues to arise, you should be aware of the potential risks and think carefully before agreeing for you and your child to participate in this research project.

We are only searching for genes that are related to your child's condition but it is possible that we may find genes responsible for other genetic conditions that you do not know about. If we find that you or your child have any genetic condition that you do not know about, your child's doctor will contact you to discuss the findings and refer you to a genetic counsellor.

On rare occasions, we may find a genetic change unrelated to the research that could have implications for your health. If changes are found in you or your child's genes, your child's doctor will discuss this with you. If we do find an unusual gene change and tell you about it, you and/or your child's health can be managed in the most appropriate way.

It is possible that we will find a change in your or your child's genes but not know for certain whether it is important or how it relates to your child's condition. If we find a change in you or your child's genes that we are certain about, your child's doctor will contact you to talk about it.

The genetic tests we perform may tell us something about you or your wider family. Learning about the results from genetic research may affect you and your family emotionally and could interfere with family relationships. You may need to decide about making your family aware of the existence of genetic information. Family members may or may not wish to know this information.

Some people in your family might want to know about your child's results and whether the result has implications for them. Your child's results will only be provided with your permission.

You may be required to inform insurance companies or employers in the future of any genetic information that you learn about yourself or your child through this project.

Because this research involves whole families, by chance, we may discover that parents and children or siblings may not be biologically related. Information regarding paternity or maternity will not be made available through this project.

Some people may find it distressing to receive information about their or their child's genetic make-up and future health. There is also the possibility that genetic information may be important in understanding the risks of having another child with your family's condition in the future.

Health Economic and Patient Experience Surveys

Being asked your experience of genomic testing, or the impact of your child having a genetic disorder on your social and financial situation may be upsetting for some people, but your child's genetic health care provider or specialist is available to discuss any concerns with you.

Data Sharing into national and international scientific databases

Your and your child's name and other information that could directly identify you (such as address or hospital number) will never be placed into these scientific databases. However, because your genetic information is unique to you, there is a small chance that someone could trace it back to you or your child. The risk of this happening is very small, but may grow in the future. As these databases are all access restricted and have traceable access the risk is minimised, and researchers will always have a duty to protect your and your child's privacy and to keep your and your child's information confidential.

If you require extra support as a result of participating in this study, your child's genetic health care provider can refer you to an independent counsellor if you wish.

10 How will the results be provided to me?

Your child's individual genomic test result will be given to you by your child's treating specialist/genetic healthcare professional.

Research results beyond the diagnostic report may be provided if available. This could include a summary of the impact of the genomic sequencing test for the conditions that are being studied as part of this research.

11 What will happen to information about me and my child?

By signing the consent form you consent to the study doctor and relevant research staff collecting and using personal health information about you and your child for the research project. The personal information collected in this study may include, but not be limited to:

- Your child's medical history including information about your child's condition and other medical conditions
- Your child's test results (e.g. blood tests, x-rays, scans or other test reports)
- Your child's information from Medicare and Pharmaceutical Benefits Scheme relating to medical investigations ordered and medications prescribed – a separate consent form will be provided for this data as required by the Department of Human Services.
- Reports about your child's treatment and side effects
- Family history of any related medical conditions
- Genetic and genomic testing results and information

Any information obtained in connection with this research project that can identify you or your child will remain confidential, except as required by law. Details of how you and your child's confidentiality will be protected and who will have access to you and your child's information are provided below.

All information that is collected/recorded about you and your child during the research study ("study data") will not be labelled with any personal details (such as name, date of birth or hospital number) but instead will be identified by a unique study identifier number (UIN) assigned to your child by the central coordination site of the AGHA when they join the study. Only the study doctors and their relevant research staff will be able to re-identify the UIN and study data back to you and your child.

Data will be stored for at least 5 years after the publication of the results to comply with the recommendations of the Australian Code for Responsible Conduct of Research. The laboratory performing the test according to standard clinical laboratory guidelines stores data generated from the genomic sequencing. These data may also be stored in a national data repository, as your child's individual data in combination with others' may assist in further discoveries for the genetic health of others. For this reason your and your child's data may also be shared in an anonymised way with approved genomic databases nationally and internationally. Further information about this is provided below under large-scale data sharing.

All study data will be stored in a secure, password-protected electronic database/registry. Only the study doctors, study researchers and personnel working directly with this study will have access to the database/registry.

Data related to MBS, PBS, and hospital and emergency datasets will be stored on the Murdoch Children's Research Institute's and University of Sydney's secured server and access to the data will be limited to authorised researchers in this study via assigned login password. Hard copy consent forms will be stored in a locked cabinet where it is only accessible by authorised researchers in this study. All data related to MBS, PBS, and hospital and emergency datasets will be destroyed after 7 years from the publication of the final project report. Hard copies will be shredded and destroyed by secured destruction service provider. Non-identifiable data will be

stored indefinitely as the economic model will be critically appraised by the scientific community, and update and running of the model will require access to the non-identifiable data files.

Your child's health information will be used for the purpose of this research project, and subject to your agreement, may also be used in future research projects which are approved by a Human Research Ethics Committee.

Information about your child's participation in this research project may be recorded in your child's health records.

Your child's health records and any information collected and stored by the study doctors during the research project may be reviewed for the purpose of verifying the procedures and the study data. This review may be done by the ethics committee that approved this research project, regulatory authorities, or as required by law. In these circumstances, these parties will review only, not collect or record, your child's personal information. By signing the consent form, you authorise release of, or access to, this confidential information.

In accordance with relevant Australian and/or State privacy and other relevant laws, you have the right to request access to your and your child's information collected and stored by the study team. You also have the right to request that any information with which you disagree be corrected. Please contact the study team member named at the end of this document if you would like to access your information.

It is anticipated that the results of this research project will be published and/or presented in scientific and medical meetings. In any publication and/or presentation, information will be provided in such a way that you or your child cannot be identified.

Large-scale data sharing

To do more powerful research, it is helpful for researchers to share information they get from studying human samples. They do this by putting it into one or more scientific databases, where it is stored along with information from other studies. Researchers can then study the combined information to learn even more about health and disease.

If you agree to participate in this study your and your child's de-identified or anonymised genetic and health information may be placed into one or more scientific databases. It may allow study investigators to identify the cause of your child's condition, as features of your child's rare condition can be matched to other individuals with the same genetic variants or similar features. There are many different kinds of scientific databases; medical research institutions maintain some while governments maintain others. For example, the National Institutes of Health (an agency of the US Government) maintains a database called "dbGaP." Other databases in which your or your child's data may be shared in a de-identified way include but are not limited to PhenomeCentral, PhenoTips, MatchMaker Exchange and Genematcher. These secure databases are shared by the international rare diseases community including Canada, the United States, Europe and Australia. They store descriptions of the features of the condition and genetic variation(s) that may identify the cause of that individual's rare condition.

Access to these databases is restricted to approved medical researchers. A researcher who wants to study the information must apply to the database. Different databases may have different ways of reviewing such requests. Researchers with an approved study may be able to see and use your and your child's information, along with that from many other people.

Your and your child's information will be handled so that you will not be readily identifiable. These safeguards to protect your privacy include:

- Personal identifiers are removed (i.e. name/date of birth);
- Personal details are kept separate;
- Data is coded;
- Only information relevant to the question being asked will be shared;
- Stringent security measures prevent unauthorized access or misuse.

This use of your or your child's information has the potential to improve understanding of normal and abnormal variants, as it will allow the variants in the genomes of thousands of people to be compared. Your child may not benefit directly from this work, but your and your child's information may be used for advancement of knowledge generally.

You will only be contacted about this data sharing if researchers from the AGHA find a result that is relevant to your clinical care. Results from research carried out by other approved researchers on information available in these databases will not be returned to you as they will not have access to your child's identifying information.

When your child reaches the age of 18 and if your child is able to provide informed consent, we will attempt to contact your child for their acknowledgement and continued consent to be involved in this research, if it is ongoing at that time.

12 What will happen to the samples collected for these studies?

Samples (blood and/or saliva) will be collected at an appropriately located pathology collection facility and will be sent directly onto an accredited diagnostic laboratory for DNA extraction for both your and your child's samples and testing on your child's sample only. In the diagnostic setting these samples will be identifiable as a requirement of sample tracking.

Other samples (where required) will be collected by your clinical team and passed on to relevant diagnostic or research laboratories as mentioned above.

As part of this study you are also consenting to your and your child's samples being accessed by researchers on this study where further testing may be carried out as described above in sections 2 and 5. These samples will be provided to researchers with your child's study UIN, so any information relevant to your child's condition may be passed back to your child's clinician. These samples will also be stored in a research laboratory in an area with restricted access.

Your and your child's samples will be kept indefinitely unless you request the destruction of these samples. We will track your and your child's samples though our study database and hence will know where your samples are if you request this.

As part of this study for your child's specific condition, your child's samples will be tested at:

Department of Molecular Genetics, The Children's Hospital at Westmead (NSW) or Women's and Children's Hospital (SA)

with both your and your child's samples being sent to, research (as described above) carried out and stored at:

1. Kids Research Institute (NSW) and/or Murdoch Childrens Research Institute (VIC)

2. Murdoch Childrens Research Institute (VIC)

3. Harry Perkins Institute for Medical Research (WA) and/or the Kids Research Institute (NSW) and/or the Broad Institute (USA).

4. NSW Health Pathology Randwick Genetics (NSW)

5. South Australian Health and Medical Research Institute (SA)

6. The Garvan Institute for Medical Research (NSW)

7. The Centre for Personalised Immunology (ACT)

9. The Garvan Institute for Medical Research (NSW) and/or Kinghorn Cancer Centre (NSW) and/or Victor Chang Cardiac Research Institute (VCCRI) (NSW) and/or Royal Prince Alfred Hospital (NSW) and/or Centenary Institute (NSW) and/or PathWest (WA) and/or Victoria Clinical Genetics Services (VCGS) (VIC)

If you consent to your and your child's samples being used in future ethically approved research, your and your child's samples may be provided to other researchers who are not part of this project in an unidentified way.

13 Who has reviewed the research project?

All research in Australia involving humans is reviewed by an independent group of people called a Human Research Ethics Committee (HREC). The ethical aspects of this research project have been reviewed by the HREC of Melbourne Health

This project will be carried out according to the *National Statement on Ethical Conduct in Human Research (2007)*. This statement has been developed to protect the interests of people who agree to participate in human research studies.

14 Further information and who to contact

If you want any further information concerning this project, you can contact the Prof Adam Jaffe on 02 93825500 or any of the following people:

Clinical contact person

Name	Kirsten Boggs
Position	Genetic Counsellor
Telephone	0476 807 324
Email	kirsten.boggs@health.nsw.gov.au

If you have any complaints about any aspect of the project, the way it is being conducted or any questions about being a research participant in general, then you may contact:

Reviewing HREC approving this research and HREC Executive Officer details

Reviewing HREC name	*Melbourne Health*
HREC Executive Officer	*HREC manager*
Telephone	*03 9342 8530*
Email	*research@mh.org.au*

Local HREC Office contact (Single Site - Research Governance Officer)

This project has also been authorised to be conducted at The Children's Hospital at Westmead / Sydney Children's Hospital Randwick. If you have any concerns about the conduct of this study, at this site please do not hesitate to contact the Research Governance Officer on (02) 9845 3011.

SECTION 2 - CONSENT TO PARTICIPATE IN RESEARCH

This consent form is divided into 2 parts. The first part is for participation in the research and the second part is the consent for the clinical genomic sequencing test. You must sign both section 2 and 3 to consent to the clinical testing and the research.

Consent Form for participation in research

Title	*Australian Genomics Health Alliance: Preparing Australia for Genomic Medicine*
HREC Number	*2016.224*
Coordinating Principal Investigator	*A/Prof Meredith Wilson (CHW), Dr David Mowat (SCH)*
Associate Investigator(s)	*Prof Adam Jaffe*
Location	*SCHN*

I have read, or had read to me in my first language, the information statement and I understand its contents.

I believe I understand the purposes, procedures and risks of the research described in the project.

I voluntarily consent for myself and my child to participate in this research project as described and understand that I am free to withdraw my consent for myself and my child any time during the project without affecting my child's future health care.

I have had an opportunity to ask questions and I am satisfied with the answers I have received.

I understand I will receive a copy of this Information Statement and Consent Form to keep.

I give permission for my child's doctors, other health professionals, hospitals, federal and state agencies or laboratories outside this hospital to release my child's re-identifiable information to the Australian Genomics Health Alliance (AGHA) concerning my child's condition and treatment for the purposes of this project. I understand that this information may be collected for up to 10 years following genomic testing. I understand that such information will remain confidential.

I give permission for the diagnostic laboratory undertaking the testing to release my child's identifiable genetic and genomic data associated with this study to the AGHA, and I acknowledge that the responsibility for the appropriate use and storage of the released data lies henceforth with the AGHA.

I understand that this project has been approved by the Melbourne Health Human Research Ethics Committee and will be carried out in line with the National Statement on Ethical Conduct in Human Research (2007).

Optional consent for future research:

☐ I do ☐ I do not consent to my and my child's data (health related, genomic and self-reported information) and samples being used for future ethically approved research projects.

☐ I do ☐ I do not consent to being re-contacted by the study team about related future research projects that me or my child may be eligible for.

Optional consent for parental samples:

☐ I do ☐ I do not consent for my blood and/or saliva to be collected and used in this research project.

_____	_____	_____
Mother's Name	Mother's Signature	Date

(a parent providing a sample must sign)

☐ I do ☐ I do not consent for my blood and/or saliva to be collected and used in this research project.

_____	_____	_____
Father's Name	Father's Signature	Date

(a parent providing a sample must sign)

PARENT/GUARDIAN CONSENT for participation in RESEARCH as part of this study

_____
Child's Name

_____	_____	_____
Parent/Guardian Name	Parent/Guardian Signature	Date
_____	_____	_____
Name of Witness to Parent/Guardian's Signature	Witness Signature	Date

Declaration by researcher: I have explained the project to the participant who has signed above, and believe that they understand the purpose, extent and possible risks of their involvement in this project.

_____	_____	_____
Research Team Member Name	Research Team Member Signature	Date

Note: All parties signing the Consent Form must date their own signature.

SECTION 3 - CONSENT TO PARTICIPATE IN CLINICAL GENOMIC TESTING

Consent Form for Genomic Sequencing

Genomic sequencing has been explained to me by a health professional and I have been given the information sheet describing the test.

I understand that my child's DNA will be tested for genes associated with

_____

by genomic sequencing. Only variants in genes known to be associated with the condition above will be analysed and interpreted in the first instance. **This test is NOT a general health test and will not identify all gene changes that could contribute to health problems in the future**.

I understand:

1. the potential outcomes of the test, including the potential risks and benefits, as outlined in Information Sheet 1: Exome sequencing.
2. knowledge about genetic conditions is likely to improve in the future. If new information comes to the attention of the laboratory that alters the meaning of the result for my child's health, the laboratory may inform my healthcare provider of this. Alternatively, I can ask my child's healthcare provider to contact the diagnostic laboratory to check if such information has become available.
3. there is a small chance that mutations may be identified that are associated with an unrelated condition that may develop in the future, or that may reveal carrier status of an unrelated condition. In this circumstance, my child's healthcare provider will arrange appropriate follow up care, as necessary.
4. the result can be used to facilitate the counselling and testing of other family members.
5. my DNA sample and genomic data will be stored by the testing laboratory in accordance with national laboratory guidelines.
6. the genomic data and associated healthcare information can be used and disclosed in accordance with the applicable health privacy laws.
7. the result and associated health information will be stored by my healthcare provider and may be made available for my child's ongoing treatment and healthcare.
8. my child's genomic data and associated health information may be shared, in a way that does not identify my child (anonymised), for the purpose of advancing knowledge generally, including understanding of genetic variation in humans. I understand that my child is unlikely to gain any personal benefit from this and I will not be notified if the information is shared.
9. I may be contacted to obtain my consent for the use and disclosure of my child's DNA, genomic data and associated health information for other purposes not specified above.
10. Genomic testing will not affect the ability to obtain health insurance but may affect applications for some types of life or other insurance.

Additional use of DNA and genomic data.

Please specify if you agree or not to the following:

Tick one box:

10. ☐ I agree ☐ I do not agree for my child's DNA, genomic data and associated health information (information) to be shared with third parties other than the AGHA after the initial analysis for activities that may benefit healthcare. Examples of how the information may be shared are listed overleaf. If you agree, the information will be shared in such a way that it may be possible to identify your child so results can be returned (if necessary/applicable). If I do not agree, the information will not be shared for activities that may benefit my child's healthcare.

Examples of how information that identifies you may be used#

Some examples of how your / your child's or relative's information could be used in a way that may identify you are listed below. These uses may reveal information that may be important to your health, or the health of your blood relatives. For all the examples below, your information can be linked to you so any information with important health implications can be returned to you.

Example of use:
Activities to clarify the cause or management of your condition.* For example, your doctor may share your information with international groups trying to identify variants common to people who all have the same condition. The greater the number of patients with the same condition and the same variants, the more likely the variants are to contribute to the condition.
Research to identify new genes associated with your condition.* Research opportunities aimed at finding the cause of your condition may be offered. This may involve a re-analysis of your genomic data. With your permission, your doctor and the laboratory could share your information, meaning researchers may not need to re-sequence your genome and you would not need to provide another sample.
Trying to clarify what a specific variant does in the cells of the body.* To help clarify whether the variant affects cell functioning, the laboratory may ask for your information so they can do further work that may provide clues to the role of the variant.

*You may be asked to sign a separate consent form for this use.

PARENT/GUARDIAN CONSENT for Clinical Genomic Testing

_____
Child's Name

_____	_____	_____
Parent/Guardian Name	Parent/Guardian Signature	Date

Parent/Guardian email _____

_____	_____	_____
Name of Witness to Parent/Guardian's Signature	Witness Signature	Date

Declaration by researcher: I have explained the potential clinical utility (including the risks and benefits) of the requested test, as well as the clinical implications of not undergoing the test, and answered questions asked of me by the patient and/or the patient's guardian or authorised representative.

_____	_____	_____
Health Professional Name	Health Professional Signature	Date

Note: All parties signing the Consent Form must date their own signature.

NOTE: Copy of this clinical consent is to be sent to testing laboratory with sample.