

# AUSTRALIAN AUTISM BIOBANK

## Storage of information

The genetics of autism is complex, and these studies often take many years. We will retain all data collected for this study (including genetic information) indefinitely, with annual reviews.

As soon as you enter the study, your child and your family would be identified by a code number. The document matching your code numbers and names will be kept separately from the study data.

The information we collect from the questionnaires and the clinical assessment will be entered immediately into a secure electronic database. The hard copies of this information would be kept in a locked filing cabinet at the Centre for Children's Health Research.

The blood samples we collect will be labelled with a code number and will be sent within 24 hours to the University of Queensland. DNA, RNA and plasma will be extracted from the samples and then transported to Wesley Medical Research (Brisbane) and stored in a locked freezer at this site. You and your child are able to contact the study team at any time for further information, and also to request that the stored information be destroyed.

## Access and use of collected information

The information stored in the Australian Autism Biobank will be valuable to the autism research community. In the first instance, the data will be used by the research team specified on Page 2 of this Information Booklet to investigate the causes of ASD. Your biological samples will only be used for bona fide research studies. Some genetic information about you will be obtained and used with other data in your medical records to see how your genes relate to your diagnosis and general health. Your samples will NOT be used for research involving reproductive technology, human embryos, or cloning.

Second, genetic information about both parents and your child, as well as your child's clinical information, may be sent to other researchers around Australia and the world for collaborative research purposes. Where appropriate, your information may be sent interstate or overseas for collaborative research purposes. This can only happen when we are sure that requisite approvals have been obtained and the necessary ethical and privacy safeguards are in place. Sharing information between research groups is critical to making research advances, and we would like the Australian Autism Biobank to be part of this important process. There may be occasions where the data we collect from you and your child are published along with our scholarly articles. Importantly, we will never share any of your personal identifying information. Instead, your data will be labelled with the code number allocated to you at the start of the research.

Access to the data will be managed by the 'Biobank Access Committee', which will be chaired by Prof Sylvia Rodger from the Autism CRC. Access to data will only ever be granted by this committee when researchers have approval from an Ethics Committee and where the Biobank Access Committee can identify a clear potential benefit of the proposed research to the ASD community.

The law in Australia dictates that you may not be rewarded financially for donating biological samples. We are, however, allowed to profit from research outcomes that are ultimately successfully commercialised. Any money we receive from commercial ventures is always put back into medical research.

## Withdrawing consent

You are free to withdraw your consent to participate in this study at any time. Your decision to withdraw consent will not influence the care your child receives from any of the personnel involved in this study. If you withdraw consent, we will destroy the hard and electronic copies of the collected information as well as the biological samples.

Once your child turns 18 years of age, we will recontact your family to see if your child would like to continue their participation in this study. If they have the cognitive capacity to provide consent at this time, they will have the choice to continue participation in this study or to withdraw consent. The decision to withdraw consent will not affect their care by any personnel involved in this study.

## What happens to the results of the research study?

*Individual results:* You should not expect to get individual results or Anticipated Findings (related to autism) from research done through the Biobank. We would not normally give feedback about results for individual children or adults to anyone. Your child should remain under the care of their usual practitioner/s. We will not give the results to your doctor. We will not put them on your medical record. This Biobank will not report Incidental Findings (not related to autism) from the genetic studies that are carried out on your samples. Incidental Findings are those which are unrelated to the research but which may come up when genome sequencing is undertaken. If you want to obtain clinical predictive genetic testing, then you should discuss that with your usual doctor. *Group results:* We plan to publish our findings in scientific journals. You can get general news about the studies at [www.autismcrc.com.au](http://www.autismcrc.com.au)

## Who has approved the study?

This study has been approved by the Princess Margaret Hospital Ethics Committee, La Trobe University Human Research Ethics Committee, Sydney Local Health District Human Research Ethics Committee and Mater Health Services Human Research Ethics Committee.

## Who to contact if you have any concerns about the running of the study?

If you have any concerns or complaints regarding this study, you can contact the Director of Medical Services at PMH (Telephone No: (08) 9340 8222). Your concerns will be drawn to the attention of the Ethics Committee which is monitoring this study.

If you would like to take part in this research study, please contact our research team at the Olga Tennison Autism Research Centre on 03-9479-5421 or [autism.biobank@latrobe.edu.au](mailto:autism.biobank@latrobe.edu.au)



# The AUSTRALIAN AUTISM BIOBANK

Parent/Guardian Information Booklet

Your family is being invited to take part in a research project that seeks to gain more insights into the causes of autism. Before you decide, it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and discuss it with other family members. Do feel free to ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part.

This information Booklet contains the following information:

- Why are we doing this study?
- Who is carrying out the study?
- Why have I been chosen?
- Does my child have to take part?
- What will happen if I give permission to take part?
- What will happen with the blood samples?
- What are the possible disadvantages in taking part?
- What are the possible benefits in taking part?
- Storage of information
- Access and use of collected information
- Withdrawing consent
- What happens to the results of the research study?
- Who has approved the study?
- Who to contact if you have any concerns about the running of the study?
- Who to contact for more information or if you would like your child to take part in this research?

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## Why are we doing this study?

Autism is a developmental condition present in approximately 1% of the population. Diagnosis of autism is defined by (1) differences (deficits) in social communication, and (2) fixed and repetitive interests. Autism is a spectrum and individuals may have very varied capabilities and support requirements in both of these areas.

The aim of the Australian Autism Biobank is to collect detailed information on Australian children with autism and their families. Understanding how the many individual genes, gene combinations, and gene-environment interactions may result in the broad spectrum of autism may allow more accurate or earlier diagnosis. In the future, the information from this Biobank may potentially allow diagnosis and targeting of supports according to genetic subtypes of autism.

We are taking a long-term approach towards establishing a strong autism research program in Australia that is closely integrated with autism clinical services and also the training of future health care professionals providing services to individuals and their families. We expect that some of the discoveries that will emerge from the Australian Autism Biobank will contribute to international autism research.

## Who is carrying out the study?

This study is being funded by the Autism Cooperative Research Centre (Autism CRC; [www.autismcrc.com.au](http://www.autismcrc.com.au)), which is a national research program aimed to improve the lives of people with autism and their families.

The Australian Autism Biobank is conducted across four Australian states, and is led by Prof Andrew Whitehouse (WA), Prof Cheryl Dissanayake (VIC), Professor Valsamma Eapen (NSW) and Dr Honey Heussler (QLD). The biological data and samples will be stored at the Wesley Medical Research (WMR), Brisbane. Your samples will be under the custodianship of Autism CRC for the duration of the Autism CRC. If the Autism CRC is finalised, custodian will pass to WMR.

## Why have I been chosen?

You have been approached because your child has been diagnosed with autism. We are approaching families of children who have received this diagnosis and live in WA, VIC, NSW or QLD.

## Does my child have to take part?

It is up to you and your family to decide whether or not to take part. If you do decide to take part, you will be given this information sheet to keep and be asked to sign a consent form. You and your family are still free to withdraw at any time and without giving a reason. A decision to withdraw at any time, or a decision not to take part, will not affect your child's health care in any way and your child should still remain under the care of their usual health practitioner/s.

## What will happen if I give permission to take part?

If you decide to take part, a researcher will contact you via telephone. They will discuss the five parts of the study with you. These are:

### 1 Questionnaires

You will be sent via mail a series of questionnaires, which asks about your child's development (including puberty), as well as the medical history and social behaviours of the biological parents. The questionnaire asks some questions that could be considered 'personal', such as any illicit drug use. All information is kept strictly confidential and you can choose not to answer any question. These questionnaires can be completed at your home and will take about 2 hours to complete.

### 2 Clinical Assessment

You and your child will then be invited into the Olga Tennison Autism Research Centre at La Trobe University, for a clinical examination. In this session, your child will take part in various child-friendly games and activities. We will also take measurements of you and your child's head circumference, height and weight. This session will take around three hours and is designed to be fun for children. We will also ask parents to complete a structured parent/caregiver interview based upon your family history and child's development, and also complete a short cognitive ability test, taking about two hours in total.

### 4 Biological Samples

#### Blood sample

To obtain genetic information, we will ask to obtain a blood sample of up to 12mls from your child. We will also request to take a blood sample from the biological parents (approximately 15ml each). The procedure will take around 10 minutes and will be carried out by suitably qualified staff at La Trobe Medical Centre. If a blood sample cannot be collected, we will collect a saliva sample using a small tube to spit into or using a small sponge placed in your child's cheek.

### 4 Biological Samples

#### Stool (faecal) sample

We will ask you to collect a sample of your child's stool, a day or two before the appointment. We will send you instructions and a container for collection. This can be kept at room temperature in a sealed plastic bag. You can put the plastic bag in a container before bringing this sample to your appointment. We will then ask you to complete a short food questionnaire about your child.

#### Urine sample

We would like to collect a urine sample at the same time as your child's blood test. If this is not possible, we will ask you to collect a urine sample from your child up to a few days before the appointment which can be stored in your freezer and brought to your appointment. We will send you instructions for collection and a sample container.

#### Hair sample

We would like a small hair sample from your child. This will involve cutting 10 strands of hair from near the root of the hair follicle. This is not painful in anyway, and will not affect your child's appearance and it can be taken from the temple or back of head. This sample can be collected at your appointment or at home.

All biological samples will be transported to the University of Queensland for initial processing and placed in long-term storage at Wesley Medical Research (Brisbane).

### 5 Siblings of Children with ASD

With your consent, we would like to obtain clinical information and biological samples from any other children under the age of 18 years in your family who do not have autism. This would involve a short clinical assessment (about one hour), and a questionnaire about early childhood development. We would also like to obtain blood, stool, urine, and hair samples from these children.

The procedure for collecting blood for children is as follows:

- i. You and your child will be taken to a 'child-friendly' room.
- ii. Your child will be encouraged to sit in a chair or lie down on a bed.
- iii. A soft rubber band (tourniquet) is placed around your child's arm.
- iv. Blood will then be taken from his/her arm. Your child will feel a small 'prick' sensation as the needle enters the skin. The blood will then fill a test-tube for around 10 seconds.
- v. The needle is then withdrawn from the skin and a cotton wool bud is placed on your child's arm to stop the bleeding.

Importantly, the qualified staff at La Trobe Medical Centre are highly experienced in taking blood from children.

If you or your child would like to stop this procedure at any time, you may do so. If your child requires some 'numbing' cream prior to blood collection, we can provide this as well. If you cannot attend these appointments, you can still participate by completing these questionnaires at home and mailing these back along with a saliva sample using the provided tubes from each person in your family who would like to participate.

## What will happen with the biological samples?

Blood comprises many substances and we will be extracting two elements from each sample: DeoxyriboNucleic Acid (DNA) and plasma. Each element will be stored in secure freezers at the University of Queensland for later analysis. DNA is the chemical substance that carries the genetic information required for the reproduction, growth, development, and function of the human body, including the brain. These elements will be analysed in search of genes of possible importance to autism and related disorders. We may also look at the plasma for exposures to common chemicals in the environment. The stool samples will be sent to Queensland, where the gut bacteria (microbiome) will be analysed. Urine samples will be stored and urine products (urine metabolome) will be analysed. The hair samples will be tested for levels of certain proteins.

## What are the possible disadvantages in taking part?

Some discomfort may be experienced when a blood sample is taken. A local anaesthetic cream can be applied beforehand to minimise pain. Afterwards a small bruise at the site of blood collection may develop. The procedures will be carried out by qualified staff at La Trobe Medical Centre. Stool and urine samples may also be difficult to obtain in children. We will provide detailed instructions to help you attempt to collect this at home.

The Family Questionnaire asks some personal information about the pregnancy, your child and your family. You are able to answer as many of the questions as you like. Importantly, all information is kept confidential and secure.

The DNA analysis may identify instances where the father is biologically unrelated to the child. These results will be kept strictly confidential and not disclosed to anyone outside of the research team, unless there is written agreement from both the mother and father of the child.

There is no cost for taking part in this study

## What are the possible benefits in taking part?

The information we get from this study could help design better ways of assessing and helping future generations of people with autism. You and your child could be involved in an important scientific advance.

Your family may participate in as much or as little of this study as you would like.