# Parent/Carer Information Sheet

**Project Title: Global Angelman Syndrome Registry**

**Principal Investigators:**

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**You are being invited to take part in a global registry as the parent/ caregiver of an individual (either a child or an adult) who has a diagnosis of Angelman Syndrome (AS). Before you accept or decline the invitation, it is important for you to understand why this registry is being built and what it will involve. Please read this information sheet and discuss it with relatives, friends and clinicians caring for your child/ adult with AS, if you wish. If you have any further questions, please contact a member of the research team above.**

**Participation in the registry is voluntary. Please take the time to decide whether you would like to take part, or not.**

## Project Overview

The Global Angelman Syndrome Registry is being initiated by the Foundation for Angelman Syndrome Therapeutics (FAST) Australia with support from the Mater Medical Research Institute in Brisbane, the Royal Children's Hospital in Melbourne, and the Centre for Comparative Genomics at Murdoch University in Perth.

The study is being coordinated in Queensland by the Mater Medical Research Institute, and is being supported by the Royal Children's Hospital in Melbourne. The lead researcher in Queensland is Associate Professor Honey Heussler from the Lady Cilento Children's Hospital. Professor Katrina Williams heads the contribution of the Royal Children's Hospital in Melbourne.

The aim of the registry is to establish a database, which will be an electronic report containing information about your child/ adult with AS and his/ her disorder. Having an electronic database of individuals with AS means that we can identify groups of individuals with AS from all over the world, who might be able to participate in clinical trials and other research to help develop new therapies for AS. The database will contain all the data that researchers will need, including each child/ adult's diagnosis, symptoms and other important aspects of their disorder.

Registry data collected over time will also be used to compile a natural history data set for individuals with Angelman Syndrome. Descriptive and statistical analysis techniques will be used to develop an understanding of the health and medical, behavioural and developmental outcomes of individuals with Angelman Syndrome according to phenotype, genotype or other characteristics of their diagnosis, as well as demographic factors. This may help to enhance understanding of the long term clinical outcomes and natural history of AS, which may help to shape future research in

treating this disorder. Third party researchers not affiliated with this project may also apply to the registry governance committee to 1) access de-identified (anonymised) data for research purposes or 2) access identifiable data with the permission of participants. See the sections "**Will information about me and my child/ adult be kept confidential?"** and **"How will my child/ adult's data be used?"** below.

The registry is patient driven, meaning that parents and caregivers take an active role in participating in the registry. Recruitment for the registry takes place online via Angelman Syndrome organisations, which provide links to the online registry.

Before you consider registering you child/ adult's details, it is important that you understand what is involved and what will be done with the information you provide. This form contains answers to some of the questions you might have on the database. At the end of the form, and after you have had some time to think about it, we shall ask if you wish to register.

If you do, we shall ask you to submit an online consent form saying that you agree to join to start entering data into the registry. If you have any questions, please contact us before consenting to join the registry.

**How will I benefit from registering?**

This database aims to benefit individuals living with AS. We will make contact with you as a parent/ caregiver of a child/ adult with AS when we are a receive requests from doctors or researchers to help assess possible new treatments (clinical trials). By holding secure records of your child/ adult's clinical details we will be able to decide whether or not such trials would be suitable for your child/ adult with AS. You will not receive any payment or any other financial benefit as a result of joining the database. The results of research arising from the database may have business potential (if for example utilised by a pharmaceutical company to develop a new drug) but you will not receive financial benefits from such development.

Nevertheless, there may be other benefits to joining, including the following:

- We will inform you as a parent/ caregiver about suitable clinical trials that your child/ adult with AS might be eligible to join.
- The details collected will also provide information for doctors interested in the best standards of care for Angelman Syndrome.
- Information collected in the registry may help to progress research into the study of Angelman Syndrome.
- The information may help with service planning for people with Angelman Syndrome and their families in various locations.
- Details collected may be used to compare Angelman Syndrome populations in different parts of the world.

**What information will I be asked to provide about my child/ adult with Angelman Syndrome?**

Once you have consented to participate in the registry you will be able to complete a series of modules that will take approximately 1.5 to 2 hours to complete. We suggest that you collect any information you have on your child/ adult's diagnosis, tests and development to make this process easier (however they can be added at a later date). Similarly if you find that you don't have enough time to complete all the information in one block you can login again and complete at a later date.

The current modules are;

- Fertility, prenatal and birth history
- Newborn and infancy history
- Epilepsy
- Medications and interventions

- History of diagnosis and results
- Illnesses or medical problems
- Medical history
- Behaviour and Development
- Sleep
- Sleep Disturbance Scale for Children
- Pathology and Diagnostics

You will be asked each year to review the information that you have provided about your child or adult with AS and update any changes to their condition. Additional modules may also be added over time.

**I want my child/ adult with AS to be involved in a clinical trial. If I register, is this guaranteed?**

No. Although one of the main aims of this database is to make it easier for individuals with AS to be recruited for clinical trials, there is no guarantee that registering your details will mean you will be automatically approached to take part in a clinical trial. The registry may also operate for some time before a clinical trial becomes available.

Doctors or researchers organizing a clinical trial will approach the registry governance committee to provide details about the clinical trial and eligibility requirements. If the clinical trial satisfies the requirements of the registry governance committee, we will review the details you have given and if the trial appears to be suitable for your child/ adult with AS we will provide you with contact details for the doctor/ researchers conducting the trial.

Please note that the doctors/ researchers conducting the trial may need to assess the child/ adult with AS in greater detail, during which it may be clear that other developments in their health or details not recorded on the database mean that the trial is not suitable for them.

**Will information about me and my child/ adult be kept confidential?**

All information we receive from you will be treated confidentially. We make every effort to ensure your data is kept safe but we remind you to be vigilant when entering information online. Details of your child/ adult's specific diagnosis as well as personal information about you and your child/ adult (name, age, address, gender) will be stored on the database. This information is all required to enable us to match you with criteria for prospective clinical trials and better understand Angelman Syndrome. Only doctors and scientists involved in this project who are given specific permission will be allowed to look at this information. The information you provide can be made available to your treating doctor if you wish. If we publish any research or other documents based on information from the database, this will not identify you by name. Please note that non identifiable data must be stored for 15 years as required by Australian government regulations (National Health & Medical Research Council Guidelines).

**Do I have to enrol my child/ adult with AS in the registry and can I withdraw if I change my mind?**

Enrolling your child/ adult with AS the registry is voluntary. Should you wish to withdraw your child/ adult's information from the database you will be free to do so at any time without having to provide any explanation. If you wish to withdraw your child/ adult with AS from the registry, you should get in touch with the staff in charge of the database. Contact details are provided above. Joining or leaving the database will in no way affect the care your child/ adult receives for his/ her condition.

**How will my child/ adult's data be used?**

The main aim of us asking you enrol your child/ adult in the registry is to help recruitment into clinical trials. However the data may also be used to learn more about people with Angelman Syndrome. The information you provide may be used to help researchers design future investigations. Any data released for research will not identify you or your child/ adult by name.

Your child/ adult's de-identified (anonymous) data may be accessed as part of the full Angelman Syndrome registry data set by third party researchers wishing to conduct their own research into Angelman Syndrome. Third party researchers must apply to the registry governance committee and satisfy the requirements of the registry governance committee regarding ethical use of the data and maintaining participant confidentiality in order to access de-identified data.

Third party researchers wishing to conduct clinical trials or other studies which require access to identifiable information must also apply for approval from the registry governance committee, and show evidence of ethical clearance to conduct their research from an independent ethical review board. If you provide consent for the research team to contact you about clinical trials or other research that your child/ adult with AS may be eligible to participate in, we will contact you directly with information about the study. Your personal information will never be provided to third party researchers.

**How will our details be updated?**

You will be able to update any registry details via the web page including parent/ caregiver contact details or any changes to your child/ adult with Angelman Syndrome's condition. We will send you a yearly reminder to update information about your child/ adult with Angelman Syndrome's condition. However, you may also need to update information about your child/ adult's condition at other times, such as following clinic visits. You may also contact us at any time if you need to amend your details. We will also ask you each year if you are still happy to have your details included on the database. You are free to have your child/ adult's information withdrawn from the database at any time. If you have provided consent for us to do so, we will also contact the clinician in charge of your child/ adult's medical care so we verify your child/ adult's Angelman Syndrome Diagnosis.

**Who is funding the research?**

This study has been funded by the Foundation for Angelman Syndrome Therapeutics (FAST) Australia and there is no conflict of interest on the part of any of the researchers.

**Who has reviewed this project?**

This study has been reviewed and approved by the Mater Health Services Human Research Ethics Committee (EC00332). Should you wish to discuss the study in relation to your rights as a participant, or should you wish to make an independent complaint, you may contact the Coordinator or Chairperson, Human Research Ethics Committee, Mater Health Services Level 2 Aubigny Place, Raymond Terrace South Brisbane 4101 or telephone +617 3163 1585, email: research.ethics@mmri.mater.org.au.

**What if I have any concerns?**

If you have any concerns or other questions about this study or the way it has been carried out, you should contact one of the investigators on the telephone numbers or email addresses provided on page 1 of this document.

**Thank you for taking the time to read this information sheet**

# CONSENT FORM FOR PARENTS/ CARERS

## Global Angelman Syndrome Registry

Name of Researcher:

A/Prof Honey Heussler Mater Medical Research Institute, Brisbane

**Please provide your informed consent by checking the boxes below. Consent questions 1 – 8 and 12 are required for inclusion in the registry, while consent questions 9 - 11 are optional.**

**A copy of the Parent/ Carer Information presented here is available by clicking on the "Information Sheet - Please Read!" link at the top of this page. There is no need to sign or upload this form unless you are giving consent for the research team to contact your clinician on your behalf (see item 9 below).**

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| 1. I confirm that I have read and understand the information sheet (V6) dated 26th of May 2016, for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily. | |
| 2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected. | |
| 3. By entering data into the registry, I understand that I give consent for the storage of data on my child/ adult with Angelman Syndrome in the Global Angeman Syndrome Registry. | |
| 4. I understand that the storing of data will allow contact to be made with me if a suitable clinical trial/ research study becomes available for my child/ adult with Angelman Syndrome. | |
| 5. However, I accept that allowing my data to be stored on this database does not mean my child/ adult with Angelman Syndrome will automatically be entered into future clinical trials/ research studies. | |
| 6. I understand that the data I provide may be used to inform and plan future research. | |
| 7. I understand that the results from future research may not have any direct implications for me or my family. | |

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| 8. I am happy to consent to be included in this registry. | |
| 9. I confirm I am happy for the specialist in charge of my medical care to be contacted to verify diagnostic information. | |
| 10. I consent to being contacted to complete additional modules/ for longitudinal follow up. | |
| 11. I consent to being contacted about clinical trials and research studies that my child/ adult with Angelman Syndrome may be eligible to participate in. | |
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| Please read the Consent, Disclaimer and Release document linked here (https://angelmanregistry.info/files/Angelman_Syndrome_Consent_Disclaimer_Release.pdf).<br><br>Consent question 12 is required for inclusion in the registry. | |
| 12. I have read, understood and agree to the Consent, Disclaimer, Indemnity and Release . | |

Please sign here (parent/ caregiver):

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Name of Patient:

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Date Signature:

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