

Parent / Guardian Information and Consent Form

Short name of project

Genetics of Speech and Language Disorders

Principal investigator

Prof Angela Morgan, Senior Research Fellow

Project number

37353



What is my child being invited to do?

We are inviting your child to take part in a project that looks at the genetic causes of speech and language disorders. They have been invited to take part because they:

- a) have a severe and persistent speech or language disorder (e.g. childhood apraxia of speech, stuttering, dysarthria); and/or
- b) have a proven family history of severe and persistent speech or language disorder (e.g. childhood apraxia of speech, stuttering); and/or
- c) have a diagnosed genetic condition which may impact speech or language

If your child takes part, we will ask them to perform screening assessments and to provide a saliva or blood sample for genomic testing and/or SNP genotyping.

Please read this information and ask us any questions. You can also talk to someone you trust, like a family member, friend, or your doctor. You can take time to make up your mind. You get to decide whether this project is right for your child.



What is the purpose of this project?

This project is studying the link between genes and speech and language disorders. Speech and language disorders disrupt the natural flow of communication. This can have an impact on self-esteem, school, work and general well-being.

Our bodies are made up of cells. In most of our cells we have a complete copy of our genetic information (genome). We all have about 20,000 genes in our genomes. Our genes are made of DNA and contain the instructions for development of the body. A gene change can disrupt instructions for development. Genes are arranged on chromosomes, and these

chromosomes are inside almost every cell of your body. Each cell has about 20,000 genes on 46 chromosomes. Genes get passed down in families from parents to children.

Because speech disorders can run in families, it is possible that speech disorders are caused by a change in a gene. In addition, other gene variants may be inherited which make it more likely that a person will recover, or a speech disorder will persist. Gene variants may also arise spontaneously in children with speech and language disorders.

To date, only a small number of genes have been identified for speech and language disorder. We will use genetic techniques to search for genes related to speech and language disorders.

By trying to identify gene changes associated with speech and language disorders, we will learn more about what causes someone to have speech problems, be able to identify people who are at high risk of speech disorder, and develop better targeted treatments.



Does my child have to take part and can I change my mind?

Taking part is up to you and your child

You get to decide whether your child takes part in this project. You can say yes or no.

Your decision will not affect your relationship with The Royal Children's Hospital and Murdoch Children's Research Institute and University of Melbourne.

You can change your mind at any time

If your child does take part, they can stop at any time. Simply tell someone in the project team. You do not have to give us the reason.

Once your child stops taking part, we will not complete further assessments or collect further information about them. However, we will keep the information we have already collected and the genetic data they have already supplied unless you tell us not to. This is so we can measure the project results properly.

The project might stop for other reasons

We might need to stop the project earlier than expected. If this happens, we will explain the reasons to you.

We may also ask your child to stop taking part in the project if it is no longer in their best interest. If this happens, we will discuss this with you.

Individuals who have co-occurring features where speech and language development is not the primary concern may not be eligible for this study. Where individuals have additional diagnoses/developmental concerns (e.g., intellectual disability, epilepsy, autism, and behavioural challenges) the multidisciplinary team will review referrals on a case-by-case basis to determine eligibility for this study, including genetic testing.



What do I/ my child have to do if we take part?

This section gives you more information about what you /your child will need to do. You and/or your child may need to spend 2-3 hours on this project.

a. Online survey/s

We may email you survey/s about your child's health. For example, the survey/s may ask questions about:

- Your child's birth and development history, medical history and history of communication or speech problems.
- If/when your child was diagnosed with speech/language disorder, the type of speech/language disorder they have, details of any treatments they have had
- Whether they have any other diagnoses or medical conditions
- If they see other medical professionals, e.g. a physiotherapist, audiologist or occupational therapist.

b. Information gathering

We may also like to obtain information from:

- Health/personal records e.g. speech therapy, paediatrician, psychology, school records etc. We will ask you to supply these records.
- If your child has previously had genetic testing (such as a microarray) then we will also ask to collect these results
- We may also collect Medicare Benefit Scheme, Pharmaceutical Benefit Scheme, quality of life and health economic data.

c. Study assessment/s

We may ask you and your child to take part in an assessment or assessments. This will either involve coming to The Royal Children's Hospital, University of Melbourne, completing some assessments over videocall (e.g. Teams or Zoom) or by completing online assessments (e.g. over online platforms or surveys).

For example, your child may be asked to:

- Perform a series of assessments that determine your child's ability to understand and produce words, sounds and sentences as well as make different movements with your mouth, e.g. poking out your tongue or pursing your lips
- Perform a non-verbal task that will test cognitive ability

If in person or via videocall, the assessments will be audio and video-recorded.

d. Genetic testing

The types of genetic testing we may do includes genomic testing (e.g. microarray, Fragile X testing, whole genome sequencing, exome sequencing) and/or Single Nucleotide Polymorphism (SNP) genotyping.

Microarray is a test that looks for extra or missing segments of DNA in a person's genome. Fragile X testing will look to see if there is a section of DNA that has been expanded.

Whole genome sequencing tests the majority of the DNA including genes and the DNA between genes.

Exome sequencing tests genes that are related to speech or language disorders.

SNP genotyping finds common DNA changes across those with a speech or language disorder.

Your child will give a DNA sample (either saliva, buccal or blood sample depending on the type of analysis) so that we can complete genomic testing and/or SNP genotyping.

The buccal sample or cheek swab involves brushing a swab against the inside of the cheek for around 10 seconds with the same force as brushing your teeth.

A blood sample requires around 18mls of blood (about 4 teaspoons) from an elbow vein.

A saliva sample requires completing a "spit kit" and collect around 5ml (1 teaspoon) of saliva.

Exome or genome testing also requires **both** parents to participate with their DNA samples to be used as a reference to check if your child has any gene changes that have been inherited (passed down from parents).

Your child's genomic data will continue to be analysed using new methods and technologies to identify gene changes associated with speech/language disorder.

e. Results appointment

We may ask you and your child to come to the Royal Children's Hospital to receive the results for genomic testing. This may include a brief developmental assessment by a clinical geneticist. This may take up to 60 minutes. Results may also be discussed over the phone or via telehealth and by letter.

SNP genotyping results will not be communicated on an individual level.

Optional parts of this project

If your child takes part in this project we will ask you to think about letting us do a couple of extra things.

You can say no to one or all of these things. If you say no, your child can still take part in the rest of the project.

a. Optional consent: use of images and video-recordings

We are asking you to let us use your child's clinical photographs/videos in this research.

If you choose to do this, we can publish their photo/video in scientific literature or include in conference presentations. This photo/video might help other researchers look for similarities with other children. However, readers or viewers may then be able to recognise and identify your child.

You can say no to this if you want to. If you say no, your child can still take part in the project.

b. Optional consent: contact about future projects

We are asking you to let us contact you about future projects that may be suitable for your child. The information we send will give you the full details about the project. It is your choice whether your child takes part in these projects. If you say yes, we will contact you by email or by mobile.

c. Optional consent: summary report

We are asking if you would like to receive a summary report outlining your child's speech and language assessment results, if available. If you say yes, we will contact you by email or by mobile. This will only be available for face to face or telehealth consults, not online only studies.

Project findings

At the end of the project we will send you a final letter or email. This will explain what we found out in this project – in other words, our project results. The letter will not have any information specifically about your child.



What are the benefits of taking part?

By taking part, you and your child will help the researchers understand more about the genetics of speech and language disorders. This knowledge may help people in the future.

Your child may not directly benefit from taking part in this project.

The real benefit from this project will come in the future. We hope we will be able to provide better information to parents about whether their child who has speech/language disorder is likely to have ongoing difficulties, or whether the problems are likely to be short-lived. This is important so that we can begin to find out some of the reasons that speech disorders occur and it may help us to work out who will benefit most from early intervention and treatment.



What are the risks and discomforts of taking part?

Blood tests

There are no major risks associated with a blood test. It is possible your child may feel some discomfort during the blood test. Your child may feel a sting when the needle is put in their arm to take the blood. 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.

Buccal sample

There are no risks or side-effects from a buccal sample. The force required is the same as brushing your teeth, so is not associated with any possible bodily harm.

Saliva sample

There are no risks or side-effects from a saliva sample. Some people may be embarrassed about spitting into a jar. We will offer your child a private space for collection of the sample.

Genetic tests

The genetic tests we perform may tell us something about you or your child's wider family. This may have an impact on how family members relate to one another. It is possible that we may find genes responsible for other genetic conditions that you do not know about. If we find that your child has any genetic condition that you do not know about, we will contact you to discuss the findings and refer you to a genetic counsellor.

Your child may be required to inform insurance companies in the future of any genetic information that is learnt about them through this project.

By chance, we may discover that parents and children or siblings may not be biologically related. In this case, information regarding paternity or maternity will not be available through this project.

If any new information about possible risks becomes known during the project, we will tell you immediately. If you have any questions with regards to any of this information, then please contact us.

Please take time to consider the advantages and disadvantages of discovery of a health risk before deciding to take part in this research project.

There may be unforeseen or unknown risks. In the unlikely event that your child suffers an injury because of participating in this project, the public health service will provide hospital care and treatment at no cost to you.



How will my child's information and samples be used for this project?

This section tells you how this project will collect, store, use, and share and/or dispose of your child's information and samples. If you do not want us to collect this information, your child cannot participate in this project.

Collecting your child's information

We may ask whether we can collect information for the project from your child's medical record, speech pathologist, physiotherapist, audiologist, occupational therapist, psychologist, clinical geneticist, paediatrician, local doctor/GP and most commonly directly from you and your child.

Keeping your child's information and samples safe

To keep your child's information and samples safe, we will:

- follow all relevant privacy requirements
- store information securely on an electronic database, REDCap
- store samples securely at Victorian Clinical Genetics Services (VCGS) clinical laboratory, Australian Genome Research facility (AGRF) or external ethically approved collaborative sites in Australia
- take steps to prevent anyone from accessing information or samples that identifies your child unless they are authorised to do so, such as the project sponsor or ethically approved suppliers (e.g. Go Lab Pty Ltd).

- give samples a code and keep them separate from your child's name or contact information. Your child's DNA sample will be re-identifiable. This means that we will remove your child's name and give the DNA sample a special code number. Only the research team can match your child's name to their code number, if it is necessary to do so.

You can ask us to tell you what information we have collected about your child as part of this project. If your child's information is not correct, you can ask us to change it.

The following people may have access to your child's samples or information collected as part of this research project:

- The research team involved with this project
- The Royal Children's Hospital Human Research Ethics Committee
- Clinical laboratory Victorian Clinical Genetics Service (VCGS), Australian Genome Research Facility (AGRF), external ethically approved collaborative sites in Australia, approved suppliers (Go Lab Pty Ltd) to facilitate mail out DNA kits.

DNA samples collected as part of this study for genome testing will be stored in a laboratory at the Victorian Clinical Genetics Services located at the Royal Children's Hospital or at Australian Genome Research Facility or at external ethically approved research laboratories or locations. VCGS keep your child's information and DNA sample indefinitely for use in future projects related to this project.

DNA samples stored for extraction for SNP genotyping will be stored locally. SNP genotyping will be performed at Australian Genome Research Facility (AGRF). This DNA will be kept at least until your child turns 25 years of age and after this time it will be destroyed. Personally identifiable information (name, address) will be securely sent to a third-party supplier (Go Lab Pty Ltd) for the singular purpose of sending you a self-complete DNA kit in the mail. This is for SNP genotyping studies (Genome Wide Association Studies) only. You will be told if this is relevant to your study. Your information will be kept until the package has been delivered to your address, after which it will be deleted.

Sharing your child's information with others

We will share some of your child's information with these people

- **Your child's referring health professional:** we may tell your child's referring health professional (speech pathologist, paediatrician etc.) that they are taking part in this project. They may add this information to your child's records. If we find out information relevant for your child's ongoing care, we may share this information with their referring health professional.
- **Analysing samples:** Your child's genetic data will be analysed by the Victorian Clinical Genetics Service, Australian Genome Research Facility or ethically approved research laboratories. After testing is completed, any remaining DNA will be stored indefinitely in case they need to re-analyse samples for any reason in the future

including for quality control. Your child's SNP, genomic or exome data may be transferred to a team member for further analysis. Your child's sample will be labelled with an identifier code to protect their confidentiality. We may also provide data to the researchers including your child's year of birth, gender and affected status, but will not send personal details such as their name. The research team member may be based overseas. Genomic data sent overseas is not covered by Australian laws or regulations.

RCH and Murdoch Children's Research Institute are research partners. This means that the two organisations will always share research information with each other.

Publishing project information

We will share information from this project so that others can use the findings. This project information will be provided in such a way that your child cannot be identified, unless you have chosen to provide optional consent for your child's photo or video to be used in research publications or presentations. In that case, your child may be recognisable to readers, viewers, or researchers.

We would never release the video-recordings onto social media or other platforms.



How will my child's information and samples be shared for future research?

Sharing information

To advance science, medicine and public health, we may share your child's **deidentified information** with research projects, biobanks, medical journals or data research repositories. Some of these organisations may be located overseas. **Any data that we send overseas may not be protected by Australian laws and regulations.** By signing this consent form you are giving us permission to do this.

If we share your child's information, we will remove identifying details such as their name, date of birth and address. We will give this information a special code number. We will put security measures in place to prevent re-identification of your child's identity.

There is a small chance that your child could be re-identified by someone outside of this research project. In the unlikely event that this happens, someone from the research team will contact you. If, at any point, you think that your child may have been re-identified, please let us know.



Who is running and paying for this project?

This project is being conducted by the Speech and Language group at MCRI and University of Melbourne. The site is receiving funding from the National Health and Medical Research Council to run this project.



Who has reviewed and approved this project?

This project has been approved by the Human Research Ethics Committee of The Royal Children's Hospital in Melbourne. This is an independent committee that makes sure that this project meets Australian ethical standards for research that involves people.

Comments or complaints about how this project is being run

If you have any comments or complaints about this project, please contact the Director of Research Operations at The Royal Children's Hospital and quote this reference number 37353.

You can phone the Director on (03) 9345 5044 or email them at rch.ethics@rch.org.au.



Where can I find more information?

Thank you for taking the time to read this information. You can contact a member of the project team at any time to ask questions.

Prof Angela Morgan (03) 8341 6458

Signature Page

Short Name of Project	Genetics of Speech and Language Disorders
Principal Investigator	Prof Angela Morgan, Senior Research Fellow
Project number	37353

Consent to take part in this project:

By signing this consent form, I acknowledge that:

- I freely agree for my child to take part in this project
- I understand that my child can stop taking part in the project at any time
- I have read, or have had read to me, the information provided about this project and understand what is involved including the use of my child's personal information
- I have had the opportunity to consider the information, ask questions and am satisfied with the answers I received
- I agree to my child completing genetic testing as part of this project

Optional parts of this project	Yes	No
a. Optional consent: use of images and video-recordings	☐	☐
b. Optional consent: contact about future projects	☐	☐
c. Optional consent: summary report	☐	☐

Participant's name: _____

Parent / guardian of the person taking part in the project

Signature: _____ Date: _____

Name: _____