

Information Sheet for Parents and Children

Does Genetic Variation predispose to Cerebral Palsy?

Background

Approximately one in every 500 children born in Australia has cerebral palsy. It is now recognised that most cases of cerebral palsy are associated with factors present before labour begins, and not as a result of events occurring during labour and delivery. However, what actually causes cerebral palsy is not clear. In order to determine these factors, it is important to conduct research into possible causes of cerebral palsy.

What This Study Adds

Our team is investigating whether cerebral palsy is associated with structural variation in the inherited pattern of a large number of genes across the genome. This includes genes involved in biological processes relevant to cerebral palsy as well as many others. Our study looks for genetic markers (genes) for some of these in blood taken from children and their parents. We shall study these genetic markers from children with cerebral palsy and their parents, and compare them with children who do not have cerebral palsy and their parents. We are hoping to obtain samples from all families who have participated in our cheek swab study. The study is completely anonymous and does not identify your DNA to outside parties.

Your Involvement

Cerebral palsy occurs in all ethnic groups, however for statistical and scientific reasons it is only feasible for us to study Caucasian families in this particular study. By Caucasian families, we mean where the ancestors of both parents of the child are of European origin. Please ignore this request if you do not think you are eligible.

We request permission to:

1. Collect a blood sample (11mL, approximately 1 tablespoon) from your child and up to 9mL from yourself as parents for the purpose of obtaining a DNA sample. This quality DNA allows us to better identify possible sub-microscopic variations in chromosomes that may affect brain function. It also enables us to look more closely at regions of the genome where the majority of genes that control major functions of the human body are located. From this we may be able to identify new genes that may be associated with cerebral palsy. The sample will be taken by an experienced and qualified nurse either in the convenience of your own home, or if you prefer to attend the Women's and Children's Hospital, at SA Pathology. Samples can also be collected under anaesthetic or sedation for some children with cerebral palsy who may be booked in for a procedure, by prior arrangement. Taxi transport will be arranged and paid for by the study, at no expense to you.
2. If applicable, link the results from this study with results of the cheek swab study if you participated in that study. By linking the results, researchers will be able to access data you have given approval to access such as the data held by the cerebral palsy register and Perinatal outcomes statistics unit, including the background health questionnaire completed by mothers.
3. For the mother to complete a short questionnaire about her medical and pregnancy history. When answering the questionnaire provided, please note that each piece of information we ask for is not necessarily a cause of cerebral palsy. For example, we may ask you about you taking medications during your pregnancy. This does not mean we expect these medications to cause cerebral palsy, we are simply interested in identifying an association and this may or may not be causal.
4. For us to review data collected on the Supplementary Birth Record form, a form filled in by midwives after each birth. This form also contains basic clinical information about the pregnancy, birth and hospital stay. These records are kept by the State Perinatal Data Collection Unit, and are also confidential. When collected it will be linked only by a code number to our research results.
5. To access any clinical information collected by the Cerebral Palsy Register if you are on this register. This information is confidential, and will be linked only by a code number to our research results. At all times the research results will not identify the families involved.
6. Store a portion of your child's blood and DNA samples at a research facility called 'Genetic Repositories Australia (GRA)'. GRA has the expertise to extract and store high quality DNA for us. It processes samples for a whole range of diseases to improve outcomes, and is supported by the National Health and Medical Research Council of Australia. *See extra information and consent form for this.*

As explained in the extra information from GRA, the stored DNA is anonymous i.e. not linked to your details, but can be used by GRA for any ethics committee approved research involving testing or comparison of the DNA for research in any medical disorder. This is a condition of the use of the GRA facilities for our research.

It is important for you to read the extra information attached and to sign the person specific consent forms (i.e. to provide us with separate consent for your child). We apologise for the large amount reading material. If you wish to be involved we need you to sign both the GRA consent form (white) and The University of Adelaide consent form (pink).

Your Privacy and Access to Results

In accordance with the above guidelines:

- You are free to refuse consent for this research without giving any reasons. You may withdraw from the research project at any stage. Refusal to participate will not affect you or your child's medical care.
- Your information will remain confidential except in the case of a legal requirement to pass on personal information to authorised third parties. This requirement is standard and applies to information collected in both research and non-research situations. Such requests to access information are rare; however we have an obligation to inform you of this possibility.
- There is an option for you to allow for you and your child's sample to be retained and used in future research projects, provided these projects have the approval of the Children, Youth and Women's Health Service Research Ethics Committee. If you agree to this, you and your child's sample will be archived in a re-identifiable manner (meaning that the research team only can link back the sample to the name). If you do not agree to this, any remaining samples will be destroyed in accordance with the hospital's guidelines for the safe disposal of biological specimens when the research is complete.
- Data arising from this research is required to be retained for 15 years, in a secure facility.
- You will not receive any payment for your participation in this research study. We have designed this study so that the impact on your time is minimal, and no hospital visits are required (except if you choose to attend the WCH for blood sample collection). Our research is mostly directed to improving understanding of disease. Sometimes the research will lead to findings that result in the development of a commercial test or treatment that may be overseen by pharmaceutical companies. Australian law indicates that there is no financial reward or payment to you in such an event.
- Analysis of results and their possible future clinical relevance will take about 4 years. A few of the tests in a small minority of children may have potential relevance to that individual's future health if a hereditary tendency to thrombosis (clotting) is suspected. In such cases it is possible for you to state on the consent form that you would like to be notified if such a possible clinically relevant result is obtained. It would be necessary for any test result to be re-checked by your doctor or specialist. Specific counselling about the ramifications of being tested and acting on that result would be offered at the time of notification. Alternatively you can request that all results are permanently disconnected from you and your child's details (i.e. non-identifiable without connecting codes) and that you receive no further information about your child's results. Where you have asked for a coded identification link to remain so that potentially relevant results can be offered to you, this information will not be released for other use without consent, unless required by law.
- It should be clearly understood that individual results (other than where there is a risk of thrombosis) cannot be identified and returned to you as their clinical relevance is not yet known.
- You may request a summary of the overall study findings.

Please feel free to discuss the research in detail with the investigating team. Where possible the consent of both of you and your partner is requested and we encourage you to discuss participation with your child. This research is conducted with the permission of the Adelaide Children, Youth and Women's Health Service Research Ethics Committee. If you have any concern, complaint or wish to discuss this committee's approval process please phone the secretary of the Ethics committee, Ms Brenda Penny, 8161 6521. The research is conducted within the guidelines of the National Health and Medical Research Statement on the Ethical Conduct in Research involving Humans (1999). We thank you for considering participation in this research project which may help determine the causes of cerebral palsy. This knowledge may lead to the prevention of cerebral palsy.

Professor Alastair MacLennan
Project Co-ordinators

for The South Australian Cerebral Palsy Research Team Ph: (08) 8161 7619
Gai McMichael & Corinne Reynolds (Mon - Thurs) Ph: (08) 8313 1401
Jessica Broadbent (Mon & Tue) Ph: (08) 8161 7616

Chief Investigators

Professor Alastair MacLennan
Dr Catherine Gibson

Discipline of Obstetrics and Gynaecology, The University of Adelaide at the Women's & Children's Hospital

Associate Professor Paul Goldwater

Department of Microbiology and Infectious Diseases, Women's & Children's Hospital