PARTICIPANT INFORMATION SHEET  
ADULT (16+)

Study title  
*A study of movement disorders in adults with 22q11 deletion (DiGeorge syndrome)*  
*Date: 19/02/2015 version 1.0*

We would like to invite you to take part in our research study. Before you decide we would like you to understand why the research is being done and what it would involve for you. **One of our team will go through the information sheet with you and answer any questions you have.** Talk to others about the study if you wish.

Part 1 tells you the purpose of this study and what will happen to you if you take part.  
Part 2 gives you more detailed information about the conduct of the study.

Ask us if there is anything that is not clear.

**Part 1 – to give you first thoughts about the project**

1. **What is the purpose of the study?**  
   Recently a research study has shown that people with 22q11 deletion (DiGeorge syndrome) may have an increased chance of developing a movement disorder. These movement disorders include a condition known as Parkinson's disease and isolated tremors and jerky movements of the limbs. The evidence for this is just a single research study and does not definitely prove this association. The purpose of the current project is to recruit a large number of adults (>18 years old) with 22q11 deletion syndrome (DiGeorge syndrome) and perform assessments for movement disorders. It is hoped that this will help clarify whether 22q11 deletion syndrome does predispose to movement disorders.

2. **Why have I been invited?**  
   You have been chosen because you have DiGeorge syndrome (22q11 deletion syndrome). We hope to have around 100 people with DiGeorge syndrome in this study. You were identified as being eligible for the study by your local Clinical Genetics service. There is a UK wide research agreement which allows a single NHS clinic or University research centre to identify people who are eligible for a particular research study through the records of all the NHS Clinical Genetics units. This explains why you have been contacted by a research team in Sheffield.
3. **Do I have to take part?**
   It is up to you to decide to join the study. We will describe the study and go through this information sheet. If you agree to take part, we will then ask you to sign a consent form. You will be given a copy of the information sheet and the signed consent form to keep for your records. You are free to withdraw at any time, without giving a reason. This would not affect the standard of care you receive.

4. **What will happen to me if I agree to take part?**
   You will be assessed by a clinical researcher at a single visit which will last about 1 hour. The whole research project is going to recruit participants over a 5 year period. If you agree to take part a clinical researcher will take a full medical history from you. A neurological examination of you will then be performed. This will involve testing the movements of your arms and legs, testing your sense of smell and asking you to complete some short questionnaires. None of these assessments are embarrassing or uncomfortable.

   We may ask to take a blood sample from you (10 mL or 2 teaspoonfuls of blood). We will grow cells from this blood sample in a University of Sheffield laboratory for further study.

5. **Expenses and payments**
   We can reimburse reasonable travel expenses.

6. **What will I have to do?**
   Attend a 1 hour clinical assessment at the Clinical Genetics Department at the Northern General Hospital in Sheffield or have a home visit from a researcher. You will be asked to answer some questions about your general health. The clinical researcher will perform a neurological examination of you (to assess the strength and movements of your arms and legs) and test your sense of smell (with test cards which release an odour when scratched and you then have to choose what the odour is from a list of four). You may be asked to give a blood sample.

7. **What are the alternatives for diagnosis or treatment?**
   This research project will have no influence upon your medical treatment as we are not performing a drug trial.
8. **What are the possible disadvantages and risks of taking part?**

Every effort will be made to reduce the anxiety felt during a blood test. If at any time you feel that the actual or perceived distress is too great, please don’t hesitate to tell the research doctor/nurse. Blood samples will be taken by qualified and experienced staff.

9. **What are the side effects of any treatment received when taking part?**

No drug treatments will be given.

10. **What are the possible benefits of taking part?**

We cannot promise the study will help you but the information we get from this study will help improve the treatment of people with 22q11 deletion (Di George syndrome) better.

11. **What happens when the research study stops?**

We will collect all the information together and we will decide if it is useful in telling us if the doctors can manage 22q11 deletion syndrome (Di George syndrome) better in the future.

12. **What if there is a problem?**

Any complaint about the way you have been dealt with during the study or any possible harm you might suffer will be addressed. The detailed information on this is given in Part 2.

13. **Will my taking part in the study be kept confidential?**

Yes. We will follow ethical and legal practice and all information about you will be handled in confidence. The details are included in Part 2.

**This completes Part 1.**

If the information in Part 1 has interested you and you are considering participation, please read the additional information in Part 2 before making any decision.
Part 2 of the information sheet

14. **What if relevant new information becomes available?**

If the study is stopped for any other reason, we will tell you and arrange your continuing care.

15. **What will happen if I don't want to carry on with the study?**

If you withdraw from the study, we will destroy all your identifiable samples if you wish, but we will need to use the data collected up to your withdrawal.

16. **What if there is a problem?**

**Complaints**

If you have a concern about any aspect of this study, you should ask to speak to the researchers who will do their best to answer your questions.

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If you remain unhappy and wish to complain formally, you can do this by contacting:

Mrs Linda Towers  
Patient Advice & Liaison Co-ordinator  
Sheffield Children’s NHS Foundation Trust  
Tel: 0114 271 7594

**Harm**

In the event that something does go wrong and you are harmed during the research and this is due to someone’s negligence then you may have grounds for a legal action for compensation, but you may have to pay your legal costs. The normal NHS complaints mechanisms will still be available to you.

17. **Will my taking part in this study be kept confidential?**

All information which is collected about you during the course of the research will be kept strictly confidential. Any information about you which leaves the hospital will have your name and address removed so that you cannot be recognised from it. Once the study is complete all information will be *kept for 5 years and then destroyed* or kept in your confidential notes.

Our procedures for handling, processing, storage and destruction of data are compliant with the Data Protection Act 1998.
We will keep the data from the history and examination we perform for 5 years. This will be held in an anonymised database on a password protected University of Sheffield computer. Only authorised persons (clinically qualified members of the research team and NHS clinicians) will be able to see any identifiable information.

Your medical notes may also be looked at by other people within the hospital involved in the running and supervision of the study to check that it is being carried out correctly.

18. **What will happen to any samples I give?**
If we take a blood sample from you we will use it to produce cells for study in a University of Sheffield laboratory. You will not be identifiable as a donor of these cells. The cells will remain at the University of Sheffield. They will be used to study how the 22q11 deletion might alter cell function and how this might explain the symptoms of 22q11 deletion syndrome.

19. **Will any genetic tests be done?**
No genetic testing will be performed.

20. **What will happen to the results of the research study?**
When the study has finished we will present our findings to other researchers, and we will put the results in medical magazines and websites that researchers read. We would also like to put a brief summary on the hospital research website so that you will be able to read about our results too. This will be available at the end of the study, in 2020, on www.sheffieldchildrens.nhs.uk/research-and-innovation.htm. All of these reports will be anonymous, which means that you will not be able to be identified from them.

21. **Who is organising and funding the research?**
Researchers at Children’s NHS Foundation Trust are organising this study. They will not get any extra money for doing this research. The Royal College of Physicians of Edinburgh has provided a research grant, awarded in a competition, to the research team, to fund the project.

22. **Who has reviewed the study?**
All research in the NHS is looked at by an independent group of people, called a Research Ethics Committee, to protect your interests. This study has been reviewed and given a favourable opinion by a Research Ethics Committee. It has also been given approval by the Research Department to run at this hospital.

23. **How can I find out more?**
If you would like to find know more about research in general, the Clinical Research Facility at this hospital has an **Information for families** section on its website www.sheffieldchildrens.nhs.uk/research-and-innovation.htm or you could contact the hospital Clinical Research Facility:

**Ms Wendy Swann**
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Updated by Wendy Swann 20.05.14
If you would like to know more specific information about this research project, please contact the project co-ordinator:

Name: Alisdair McNeill  
Title: Senior Clinical Fellow  
Hospital/Department: Clinical Genetics, Northern General Hospital  
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07732380747

If you would like advice as to whether you should participate you could contact the project team, or one of your health care professionals.

If you have any concerns during the study, you should contact the project team.

If you decide to take part in this study, you will be given this information sheet and signed consent form to keep.

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