

Project Information and consent form

The Neurofibromatosis Type 1 (NF1) Cutaneous Neurofibroma Consortium Project Identifying genetic modifiers of disease burden to inform treatment pathways

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We would like to invite you to enrol in the **NF1 Cutaneous Neurofibroma Consortium Project** at nf1project.com

Why are we doing this research?

Neurofibromatosis type 1 (NF1) is the most common neurogenetic condition, affecting 1 in 2500 people. NF1 causes brown skin marks (café au lait spots) and benign (non-cancer) growths around the nerves called neurofibromas.

Skin (cutaneous) neurofibromas are lumps in or under the skin. These skin lumps are usually small in size but may increase in number throughout adulthood. People with NF1 can have different numbers of skin neurofibromas.

It is difficult to know how many skin neurofibromas someone will develop. These skin lumps can cause distress for those affected, particularly if they appear in large numbers.

There is currently no drug treatment for these skin neurofibromas.

This research aims to improve our understanding of:

1. How genetics affects the number of skin neurofibromas a person with NF1 will develop.
2. How to treat skin neurofibromas in the future.

Who can take part in this project?

Adults (age 18 or older) with a diagnosis of NF1.

What will you be asked to do?

You will be asked to register on the secure study website at nf1project.com and provide:

- Photographs of the skin on your abdomen, neck and arms. Photographs of your back, face and chest are optional.
- Medical information. (Your doctor, nurse or genetic counsellor can do this for you if you prefer).
- Consent to provide a blood or saliva sample or use a stored DNA sample for this study.

Participants both in Australia and overseas will be sent a saliva kit in the post with a reply paid envelope to return the sample.

If you would prefer to provide a blood sample, this option is available. You will be provided with the address of the closest public pathology collection centre to your home. The amount of blood needed is 5mls (1 teaspoon).

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What will we do with your photographs?

- The photographs will be securely stored in the University of Newcastle cloud server.
- The photographs will be given a code. Your name will not be stored with them.
- The photographs will help us understand how genes affect the number of skin neurofibromas.

What will we do with your saliva / blood sample?

- We will extract DNA from your saliva / blood sample. Your DNA sample will be labelled with a code (not your name) and safely stored in the research team's laboratory at the University of Newcastle.
- We will do studies on your DNA called genome-wide association studies (GWAS) and whole exome sequencing (WES) to try and understand how genes interact to produce neurofibromas. This type of genetic testing will help us to study many different genes and their interactions. It will also help us to look at changes within genes that may influence the severity of neurofibromas.
- We will study biomarkers in your saliva / blood sample to understand more about the effects of NF1 on the body.
- We will compare results from biomarker and genetic studies with the information from your photographs and medical history.
- You can choose to provide consent for us to use your DNA and information for other ethically approved studies about NF1 in the future. Otherwise, your DNA will be destroyed at the end of this project.

What are the possible benefits of participating in this project?

This research is not likely to benefit you personally. The main benefit of this project is to help us:

1. Understand how genes affect the number of skin neurofibromas a person with NF1 will develop.
2. Improve our understanding of how to treat skin neurofibromas in the future.

Will I be given my genetic test results?

NF1 gene test:

- Your NF1 gene will be tested. However, we will not send you your results because this testing will be done in a research laboratory, rather than an accredited clinical laboratory.
- Adults with NF1 planning a family can see their local clinical genetic service for genetic counselling and possibly genetic testing in an accredited clinical laboratory.

Incidental findings:

- We will not be looking for gene changes that affect your general health. However, because of the type of genomic testing we are doing, there is a very small chance of identifying a genetic change unrelated to your NF1, which could be important to your health. This result is referred to as an "incidental finding".
- An example of a possible incidental finding would be finding a gene change that indicated you have a high risk of developing cancer and would benefit from early

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screening for a particular type of cancer (for example, the breast cancer [BRCA] gene).

- An "incidental finding" would be a research result and must be confirmed in an accredited clinical laboratory.
- You can consent to have "incidental findings" returned to your doctor, who may then refer you to the local genetic services to have the research "incidental finding" confirmed in an accredited clinical laboratory.
- You can also decide that you would prefer not to be given any "incidental finding" result. To learn more about incidental findings, ask your doctor for more information or go to the NF1 Project Website

What are the possible risks of participating in this project?

- A blood collection may cause pain or a bruise.
- There is a very small chance that we might find a genetic change important to your health that was not related to this research study.
- For some people, completing a questionnaire about their genetic condition may cause distress. Support is available through the research team or the NF1 support organization (The Children's Tumour Foundation) if this happens for you.

What will be done to keep your information safe and confidential?

- Your information will be stored in a secure cloud-based database within the University of Newcastle.
- Your name will be removed from your information and given a special code.
- Only Dr Dudding-Byth will be able to link your special code with your name.
- Only Dr Dudding-Byth, Dr Lovell and the research assistant will be able to see your photographs.
- Your photographs and name will not be used in any talks or papers.

What if I change my mind?

- It is your choice to take part in the project.
- You do not need to agree to enrol if you do not want to.
- If you consent and then change your mind, you can withdraw your consent to use your DNA, photographs and information at any time by emailing our team at info@nf1project.com. We will dispose of your saliva/ blood /DNA sample and delete your information.
- You do not need to tell us the reason why you want to withdraw.

Will I be told about the results of this project?

- The results of this research will be reported in a medical journal.
- This report will be put on the NF1 CNC website.
- We will send you a yearly newsletter about the project.

Ethics

This research has been approved by the Hunter New England Human Research Ethics Committee of Hunter New England Local Health District.

Complaints about the research

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If you are worried about your rights in this research or you have a complaint about the research, please contact the Hunter New England Health Research Office on HNELHD-ResearchOffice@health.nsw.gov.au and quote the reference number: 2021/ETH10963

Your Details			
Name		Date of birth	
Address		Postcode	
		Country	
Email		Phone	

- I have read (or had read to me) this information sheet.
- I understand the study.
- I consent to the research assistant/nurse uploading my photographs and information.
- I understand that the photographs and information will be stored safely.
- I have made my own choice to enrol.

Tick the box if you consent to enrol in this NF1 project.

I consent to enrol in this NF1 project.

Options for additional consent. It is your choice to give the extra consent.

I consent for my de-identified saliva or blood, DNA, and medical information to be used for other ethically approved NF1 research.

I consent for my de-identified photographs to be used for other ethically approved NF1 research.

I consent to be contacted about other NF1 research?

Tick the box below if you want to have incidental findings important to your health returned to your doctor. It is your choice to give the extra consent.

You can read about incidental findings in the document [_ More information about incidental findings](#).

Please contact the research team on [email address] if you need more information about incidental findings.

***I consent for any incidental findings to be given to my doctor (provide doctors details below)**

**[The online version of the information and consent form has a link to a document more information about incidental findings]*

If the research assistant prints off this form to consent face to face, they can print off the incidental findings document to discuss with the participant]

Your doctor's details.		
Name		
Address		
City		Postcode

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Country		
<u>Your Name</u>	<u>Signature</u>	<u>Date</u>