Participant Information and Consent Form.

Royal Children's Hospital

Project Title: The National Muscle Disease Bio-databank (NMDB)

Short Name of Project: Biobank

HREC Project Number: 90530

Principal Investigator/s: Professor Catriona McLean and Dr Peter Houweling

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1. Introduction

Thank you for taking the time to read this **Participant Information Statement and Consent Form**. We would like to invite you to participate in the National Muscle Disease Bio-databank (NMDB) which is explained below.

What is an information statement?

This information is to help you to decide whether or not you would like to take part in the research. Please read it carefully and ask us any questions about anything that you don't understand.

Before deciding to take part or not, we encourage you to also talk about this with a relative, friend, your local doctor and/or treating neurologist.

Important things you need to know.

It is your choice whether you take part in the research. You do not have to agree if you do not want to.

If you decide you do not want to take part, it will not affect the treatment and care you receive.

If you would like take part in the research project, please sign the consent form at the end of this information statement. By signing the consent form you are telling us that you:

- understand what you have read;
- had a chance to ask questions and received satisfactory answers; and
- consent to taking part in the research.

We will give you a copy of this information and consent form to keep. You can withdraw at anytime, see section 7.

2. What is the project about?

We are inviting you to take part in a project called the National Muscle Disease Bio-databank (referred to as biobank thereafter.)

Biobanks collect and store people's samples, data or clinical information to use for research. If you consent to be part of the biobank we will collect and store your blood (and skin cells, which is optional) as well as data from your medical records to study your condition in greater detail.

The knowledge gained from this research will be used to help us understand more about the cause of your muscle disease.

3. Why are we asking you to take part?

We are asking you to take part in this research project because you have received a diagnosis of a genetic muscle disease.

4. What does participation in this project involve?

If you take part in this biobank, we will ask to collect your medical information, blood test, optional skin biopsy and access to your muscle biopsy. This section gives you more information about this.

a. Medical information

We are asking to collect and store your medical information or data including age, sex, diagnosis, length of illness, treatment, imaging data and follow up. This information will be collected by NMDB staff from medical records and stored on a secure database at MCRI.

b. Blood test

We are asking for blood test from you (approximately 1 teaspoon). This will be taken when you come into hospital for visits and will be taken with routine blood collection where possible.

Commented [MM1]: Just stick to one teaspoon, rather than giving them the measurement in ml as well. Giving people two different types of measurements forces them to work harder to understand your meaning.

Optional consents

In this project, we will also ask you to do a couple of optional things. These are a skin biopsy and accessing your muscle biopsy, if available. This section explains what these optional things involve. You can say no to these things. If you say no, you can still do the other parts of the project.

c. First optional consent – skin biopsy

We are asking for an optional skin biopsy from you (2-3 mm section in size). This would be taken at a routine visit and would occur during routine procedure where possible.

d. Second optional consent – accessing your muscle biopsy

You may have had a muscle biopsy done for diagnostic reasons. If so, this tissue gets stored in a relevant pathology department. We are asking to retrieve this biopsy for research.

Commented [MM2]: Take the reader through the steps that your project involves. Instead of skipping ahead to what you will do with their child's samples, first tell them exactly what they will need to do in this project.
I've created these sub-headings based on the sub-headings in your table.
In each sub-heading, tell them what the test or procedure will involve and about how long it will take and so on.

Commented [MM3]: I've put these in the same order that they're in in the table. However, I'm wondering if the order is chronological? For example, should medical information come before the blood test? If so, please rejig the order of the subheadings here and in the table, so they appear in the same order.

5. What we will do with your samples

Once we have collected your samples, we will analyse your blood, available tissue, genetic analysis and creating cell lines. This section gives you more information about what this involves.

a. Analysis of blood, tissue and creation of cell lines

We will use your samples- blood and available tissue- to create what is called a 'cell line'. A cell line consists of cells from different types of tissue or blood that can be grown in the laboratory. This allows the creation of a continuous supply of material for research purposes and reduces the need to collect further samples. Creating cell lines often involves growing human cells in other species such as mice to provide a proper setting for cells to grow.

Cell lines are used to discover how muscle disease develops and to test new therapies in preclinical models, prior to testing in children. Creating these cell lines means we can study the specific type of cell that is not working properly– for example, your muscle cells.

b. Research genetic analysis

You have a set of genetic instructions that make you who you are. These are your genome. You can think of your genome as your book. Your genome can be broken down into smaller parts called chromosomes. These are the chapters in your book. Smaller still, are genes. These are the sentences in your book. Like a sentence, a gene has a beginning, middle and end. It contains information that shapes you. For example, your genes can determine the colour of your eyes and your blood type.

Each person has about 23,000 gene pairs. Genes are arranged along a chemical substance called DNA. Sometimes a gene message contains a 'spelling mistake'. This can change the gene's coded message which is called RNA. This gene change is also called a variation. If this makes the gene not work properly it is known as a mutation.

Muscle diseases are caused by a change in one or more genes. These conditions may emerge at birth or may appear later in life. You would have already received a genetic diagnosis.

The genetic analyses in this study are performed in research laboratories and involves testing a genetic material, including DNA and RNA from your child's sample.

The results of these additional research genetic analyses will not be given to you as the significance of such findings at this time are unclear from a clinical viewpoint.

Table One: What the Project Involves

Part of study	What does this involve?	Is this optional in the project?
Blood test	A blood test which will be taken with routine collection where possible.	**No**
Medical Information	Collection and storage on a secure database of my medical information including age, sex, diagnosis, length of illness, treatment, imaging data and follow up.	**No**

Commented [MM4]: What's the difference between a cell line and an iPSC line? Can you explain this in plain language?

Commented [MM5]: Note that the first sentence now has personal pronouns to engage the reader. It's also in the active voice.

Using the active voice is the key step you can take to make your writing clearer.

To translate your sentences to the active voice, lead with the person or thing who is performing the action. In some cases, you will need to insert a pronoun such as 'we' or a noun such as 'The Royal Children's Hospital' to translate your sentences to the active voice.

For example, this is a passive sentence: 'At the end of the study you will be sent the study results.' In the active voice, this becomes: 'We will send you the study results at the end of the study.'

Commented [MM6]: The National Statement makes clear that researchers must respect the **developing capacity** of children and young people to be involved in decisions about participation in research.
Also consider the National Statement's guidelines on research with children. The statement says:

A child or young person's maturity and capacity to understand the project varies from person to person. It can also change over the course of the project, as children age and mature.

The National Statement places value on the ethical principles of **respect**. It emphasises that even young children with very limited cognitive capacity should be engaged at their level in discussion about the research and its likely outcomes.

The National Statement is also guided by the principle of **best interests**. It says that before including a child or young person in research, researchers must establish that there is no reason to believe that such participation is contrary to that child's or young person's best interest. See section 4.2.13 of the National Statement.

The National Statement goes on to say that a child or young person's refusal to participate in research should be respected wherever he or she has the capacity to give consent to that same research. Where a child or young person lacks this capacity, his or her refusal may be overridden by the parents' judgement as to what is in the ...

Commented [MM7]: Can you recast this sentence in the active voice?

For example,

Researchers will use the cell lines to find out xxxx

Commented [MM8]: Put this in the active voice

We will not xxxx

Commented [MM9]: Split this into separate sentences and avoid the use of bracket.

Skin Biopsy	Skin biopsy 2-3 mm section in size from me. This would be collected during my routine procedures where possible.	**Yes**
Access to your Muscle Biopsy	Accessing your muscle biopsy, if available. It is likely that you had this done for diagnostic reasons and this tissue would be stored in the relevant pathology department. We are asking to retrieve this biopsy for research if it is available.	**Yes**

Please note: as part of your routine care, multiple appointments and procedures are required. During these visits, we will try to ensure sample collection for research is performed to minimise discomfort.

6. Do I have to take part in this biobank?

Participation in any research project is voluntary. It is your choice if you participate in the biobank. You do not have to agree to this.

7. Can you withdraw?

You can stop taking part in this biobank at any time. You just need to tell us so. You do not need to tell us the reason why.

If you leave the biobank, we may be able to destroy your stored samples and information. However, we will not be able to destroy samples and information already used. This is because the samples and information may already be published, and it may affect the accuracy of the research results.

Cell lines will not be generated immediately. This means that destroying these lines will be possible for up to a month after sample collection. However, once generated it is not possible to destroy cell lines that have been used or shared with other researchers.

8. What are the possible benefits of taking part?

You may not benefit directly. There may be benefits for future people with this condition. This can happen if the research increases our understanding of why people develop genetic muscle disease and we discover new and more effective treatment options.

9. What are the possible risks, side-effects, discomforts and/or inconveniences to me?

There are no major risks associated with a blood test or skin biopsy. It is possible you may feel some discomfort. You may feel a sting when the needle is put in their arm to take the blood. We can use a cream to numb the skin before the blood is taken. 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.

When taking a skin biopsy, you may also feel a sting when the skin is collected. We can also use a cream to numb the skin before the sample is taken. It is possible there may be some bruising, swelling or bleeding where the skin is collected. A bandage will be placed over the site. The bandage will be in place for 1 – 2 days to protect the location from dirt. After this time the area can be washed and kept clean until the area heals. Risks for this procedure include tissue

Commented [MM10]: Split this into separate sentences

Commented [MM11]: ??

Commented [MM12]: Any other risks you need to address?
What about risks around privacy?
Risks around the person / family learning new genetic information?
See also our Standard Wordings document for guidance on writing about risks

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Commented [MM13]: The identifiability of the data or information is a factor for researchers to consider when informing participants about the potential risks of the project. See chapter three of the National Statement.

wounds and bruising, bleeding due to insertion of medical tools with sharp edges, formation of subtle scars, and infection and inflammation in the area of the biopsy.

Despite our best efforts to protect your information, there is a small chance that you could be identified by someone outside of the NMDB team. In the unlikely event that this happens, someone from the research team will contact you. If, at any point, you think that you may have been identified, please let us know.

10. Incidental findings because of additional research genetic analyses.

An incidental finding can occur when a medical test is performed in a patient for a particular purpose, which then identifies an unexpected abnormality that is not related to the initial reason for doing the test. Such findings can be surprising to both patient and doctor.

Genetic analyses may reveal an increased risk of developing a condition that is independent from your muscle disease, but cannot predict whether somebody will develop the condition. If you give consent and we find a change in your genes that might be an important risk factor for disease based on current knowledge, you will be contacted by your treating clinician who will arrange a referral to an approved genetic counsellor.

Possible risks of being informed of incidental findings include anxiety, other psychological distress, and the possibility of insurance and job discrimination. You may need to inform insurance companies or future employers of such information that is discovered in this project or any possible future projects.

You should take time to consider the advantages, disadvantages and health risk before deciding to know about any incidental findings with your samples. Such results are only passed to families by qualified clinicians.

Genetic tests for research

We are doing the genetic tests for research purposes. We will look at your genes for features relevant to the research project. **We will not give you the test results**. This is because we do not know how the results impact you. This means that the results would not change the way we care for you.

11. Who is funding the project?

The project is funded by the Medical Research Future Fund (MRFF) – 2021 Chronic Musculoskeletal Conditions in Children and Adolescents Grant Opportunity.

12. What will be done to make sure my information is confidential?

Your privacy is very important to us and we will make every effort to protect it. Any information we collect that can identify you will be treated as confidential and clinical information will be stored securely at MCRI. We can only disclose information with your permission, except as required by law.

Samples are kept in a controlled and secure storage facility at the MCRI, housed within the RCH. We will keep your samples indefinitely. Your samples will be stored in a re-identifiable manner. **Re-identifiable** means that we will remove name, date of birth, address and UR number and this is replaced with a unique study code. Only the researchers who are working on

Commented [MM14]: See our Standard Wordings document for guidance on how to write about genetic tests in plain language

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Commented [MM15]: Split this into shorter sentences

Try to keep each sentence to 25 words or fewer. Use one main idea per sentence.

See the RCH Communication Style Guide for further information.

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Commented [MM16]: This information primarily affects the child. So 'your child' rather than 'you'.

Commented [MM17]: You've probably done this already, but it's worth revisiting the National Statement guidelines about biospecimens to make sure you've covered off the necessary requirements. The statement says:

Human biospecimens are any biological material obtained from a person including tissue, blood, urine and sputum. The term also includes any derivative material, such as cell lines. See chapter 3.2 of the National Statement.

The PICF should tell people how their biospecimens will be stored, used and disposed of. See section 3.2.12(b) of the National Statement.

It should also tell people whether their biospecimens are reasonably identifiable, and how their privacy and confidentiality will be protected. See section 3.2.12(c) National Statement.

The PICF should tell people whether the research with the biospecimens is likely to provide information that may be important to their health or to the health of their relatives or their community. If the research is likely to impact on these people's health, the PICF should tell participants whether they have a choice in receiving this information, and if they do receive it, how will this be managed. See section 3.2.12(e)-(f) of the National Statement.

The PICF should tell people whether their biospecimens will be accessed by other researchers, including researchers ...

Commented [MM18]: Data sharing?
The National Statement recognises that data sharing has become an important aspect of contemporary research. Increasingly, it is the norm that researchers need to make their data open, accessible and reusable. There are a number of reasons for doing this, such as increasing the transparency of the data and building trust in research. Most importantly, sharing data allows other researchers to build on your research, and potentially bring benefits to a greater number of people. The National Statement says that, unless there ...

Commented [MM19]: See our Standard Wordings document for how to write about data sharing here

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this project will be able to re-identify you by linking their code number to your name and other personal information.

The following people may access identifiable information/samples collected:

- the Coordinator and national NMDB team including clinicians and scientists; and
- the RCH Human Research Ethics Committee.

To advance science, medicine and public health, we may share your **reidentified** data with any current and future funders, research projects, other biobanks, medical journals or data research repositories. Some of these organisations may be located overseas. **Any data that we send overseas is not protected by Australian laws and regulations.** By signing this consent form you are giving us permission to do this. If we share your data, we will remove identifying details such as your name, date of birth and address and give the data a special code number.

13. Will my samples and data be used for Australia-based research only?

It is common for international and interstate researchers to collaborate. In rare diseases such as genetic muscle disease, international collaboration allows bigger sample numbers.

Your samples and information will only be used in projects approved by a Human Research Ethics Committee. A legal agreement between the NMDB and research organisation will be signed prior to the transfer of samples and clinical information. This is a written contract that allows the sharing of research materials between two organisations letting the recipient use the samples and clinical information for their own research purposes. The information released to external researchers would be provided in a re-identified manner, as outlined above. **Any sample and/or data that we send overseas is not protected by Australian laws and regulations.**

14. Will I be informed of the results of any research done using my samples?

We will not give you individual results from the research using your samples or information. Research updates will be posted on our website, link: www.nmdb.org.au

Commented [MM20]: I've rewritten this in the active voice

If you want to get an annual newsletter on the biobank, please give us your email address. If you are interested in hearing more you can contact the Coordinator.

15. Could the biobank close?

The biobank could close due to insufficient funding. In this case, all your information will be put into an archive that will be overseen by NMDB team. Another option may be that your data and samples get transferred to another biobank.

When all your samples are no longer required, they will be destroyed in a safe manner according to laboratory procedures.

16. Is there any financial benefit from me participating in this research?

Your sample may be used for research that has commercial benefit, for example, the development of new technology. We will not pay you for this.

17. Who has reviewed the research project?

All research in Australia involving humans is reviewed by an independent group of people called a Human Research Ethics Committee (HREC). The ethical aspects of this research project have been approved by the HREC of The Royal Children's Hospital Melbourne.

This project will be carried out according to the *National Statement on Ethical Conduct in Human Research (2007)*. This statement has been developed to protect the interests of people who agree to participate in human research studies.

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18. Contact.

If you would like more information about the project or if you need to speak to a member of the research team please contact:

Name	Ms Emily Galea
Position	Research Nurse Coordinator
Telephone	(03) 99366626
Email	Emily.Galea@mcri.edu.au

If you have any concerns about the project or the way it is being conducted, and would like to speak to someone independent of the project, please contact: about any aspect of the project, the way it is being conducted or any questions about being a research participant in general, then you may contact:

Director, Research Ethics and Governance at Royal Children's Hospital

Telephone: (03) 9345 5044

In case of a medical emergency, you should call 000 or attend your nearest hospital's emergency department.

Consent Form

Project Title: The National Muscle Disease Bio-databank (NMDB)

Short Name of Project: Biobank

HREC Project Number: 90530

Principal Investigator/s: Professor Catriona McLean and Dr Peter Houweling.

Site name: Royal Children's Hospital

Declaration by Participant

- I have read the Participant Information Sheet or someone has read it to me in a language that I understand.
- I understand the purposes, procedures and risks of the research described in the project.
- I give permission for my doctors, other health professionals, hospitals or laboratories outside this hospital to release information to NMDB concerning my disease and treatment for the purposes of this project. I understand that such information will remain confidential.
- I understand the risks I could face because of my involvement in this project.
- I voluntarily consent to take part in this research project.
- I have had an opportunity to ask questions about the project and I am satisfied with the answers I have received.
- I understand that this project has been approved by The Royal Children's Hospital Melbourne Human Research Ethics Committee. I understand that the project and any updates will be carried out in line with the National Statement on Ethical Conduct in Human Research (2007).
- I understand I will receive a copy of this Information Statement and Consent Form.

Consent:

1. I consent for my medical information to be collected and stored including age, sex, diagnosis, length of illness, treatment, imaging data and follow up	
2. I consent for a blood test (approximately 1 teaspoon) with routine collection where possible	

Optional Consent

3. I consent a skin biopsy 2-3 mm section which would occur during routine procedure where possible.	☐ I consent	☐ I do not consent
4. I consent for the NMDB team to access my muscle biopsy, if available.	☐ I consent	☐ I do not consent

5. I consent for the research team to contact me regarding future research applicable to me.	☐ I consent	☐ I do not consent
6. If research with my samples were to reveal an incidental finding, I wish to be informed.	☐ I consent	☐ I do not consent

Commented [MM21]: You need to outline this optional consent in the body of the PICF.
The optional consents in the body of the PICF should match the optional consents in the consent form.
See the Example PICF for how to set this out.

Participant Name	Participant Signature	Date

Email Address (optional)

Name of Witness to Participant (if applicable)	Witness to Participant Signature (if applicable)	Date

* Witness is not to be the investigator, a member of the study team or their delegate. If an interpreter is used, the interpreter may not act as a witness to the consent process. Witness must be 18 years or older.

Declaration by Researcher: I have explained the project to the parent/guardian who has signed above. I believe that they understand the purpose, extent and possible risks of their child's involvement in this research project.

Research Team Member Name	Research Team Member Signature	Date

Note: All parties signing the consent section must date their own signature.