

Patient Information and Informed Consent

The Australian National SMA Registry is a member of the TREAT-NMD global network of national patient registries for Spinal Muscular Atrophy

INFORMATION FOR PATIENTS

Before you agree to register in the SMA Patient Registry, it is important that you understand what is involved and what will be done with the information you provide. This form contains answers to some of the questions you might have. At the end of the form there is a section for you to sign to confirm that you agree to participate. If you have any questions after reading this form, please contact us before signing the form. You will find our contact details on page 5.

“What is a patient registry and why would you want to participate in one?”

Scientific advances over recent years have led to substantial changes in the treatment of many diseases. For some potential new treatments plans for large studies involving patients from more than one country are already in place.

Several new treatment strategies for neuromuscular diseases, like SMA, target specific genetic defects. When a clinical trial is being planned, it is very important that patients suitable for that trial can be found and contacted quickly. The best way of ensuring this can happen is to collect patients' details in a single database, or patient registry, which contains all the information that researchers will need, including each patient's particular genetic defect and other key information about their disease. The TREAT-NMD network is creating this kind of registry in countries across Europe. As well as each national registry, TREAT-NMD is also creating a single global registry which will combine the information from each of the national registries around the world, including those in Australia; this will ensure that patients who register in their national registry anywhere in the world can be contacted if their profile fits a clinical trial. In addition, these registries will help researchers to answer questions such as how common diseases like SMA are in Europe, America, Australia, Japan and other member countries. This information will also support other activities to improve patient care, such as the assessment of standards of care.

“Whose data is being collected in this registry?”

This registry is for patients affected by the disease Spinal Muscular Atrophy (SMA). In the future we will be creating registries for other neuromuscular diseases.

Because it is primarily designed to identify persons who might be suitable for new treatments, and to help researchers find the best ways of caring for patients with SMA, this registry is intended for patients currently living with SMA, and not as a record of those who have died from this condition.

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"Who should fill in this form?"

If you are the patient, you can fill in and sign the form yourself provided you are 16 years of age or older. If you are 15 years of age or younger but can understand this information, you can sign the form, but we also require your parent or guardian to sign it with you. Whatever your age, please discuss registration with your family and/or your doctor, and don't hesitate to contact us if you have any questions. If you are the parent or guardian of a child who is not old enough to understand this form, please sign the form yourself if you want your child's data to be included in the registry.

"What do I have to do and where will my data go?"

If you agree to take part in this project, you should read this patient information and sign the consent form at the end. Then you should complete the registration questionnaire with your doctor or a Clinical Coordinator, and return it to us. In the questionnaire we ask you for some personal data and some information about your disease. It is very important that the Registry is able to collect your clinical information and also details from the genetic test of the gene error in the SMN1 (survival motor neuron 1) gene that produces the SMN protein that is critical for survival of spinal cord nerves that direct muscles throughout the body and it is a mutation in this gene that has caused your SMA. If you have not already had your SMN1 genetic test this will need to be performed by a designated laboratory before your registration can be completed. The information you provide and the genetic information about your SMN gene mutation will then be entered into the Australian National SMA Registry.

The SMN1 disease gene is closely related to the SMN2 gene. Different people have a different number of copies, or dosage, of the SMN2 gene. There is evidence that the SMN2 gene dosage may be a useful indicator of how the disease might progress and also how a patient might respond to treatment of the disease. We are proposing where possible to collect information on your SMN2 gene dosage.

The Registry will be supervised by the Office of Population Health Genomics. Your data will be stored securely and no unauthorized people will be able to gain access to any information about you. The data about all patients in each country's national registry will then be fed into the TREAT-NMD global registry, which is accessible to researchers worldwide. When planning clinical trials, researchers can search this global registry for participants eligible for their trial, based on the patients' clinical and genetic data. **Only researchers who have been approved by their own local ethics committee and by the TREAT-NMD governing board and ethics council can access the registry.**

In the TREAT-NMD global registry, your clinical and SMN1 gene sequence data and the SMN2 gene dosage will be identified only by an anonymous code, not by your name. This means that when researchers search the registry, they will not be able to access your personal information (name, address etc.), but only the information they need about your disease that will help them decide whether you might be suitable for the trial. If they think you meet the criteria and might benefit from the trial, they will contact the person in charge of the Australian National SMA Registry. Staff working for the Australian National SMA Registry will "de-code" the data to find out your personal details and will ask your nominated doctor to contact you to give you information about the trial or about any other issues relevant to your disease. Neither the Australian National SMA Registry nor the TREAT-NMD global registry will give your name or any other personal information to the researchers. If you are

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interested in the information you receive about a particular clinical trial, you will be given additional information about the trial by your doctor. You are completely free to make your own decision about your participation in any trial we inform you about¹. If you decide not to take part in a particular trial, your data will still be kept in the registry and we will continue to inform you, through your doctor, about other trials unless you tell us not to. If, after careful consideration and discussion with your doctor, you decide to take part in the trial you will need to review and sign a separate consent form. Your doctor will then contact us at the Australian National SMA Registry, and we will in turn, send the required information to the researchers running the trial.

“How can I update my data if it changes?”

We will need to review data in the registry regularly, to ensure that it is up to date. To do this, we will request your doctor update your clinical records at least once per year. To help us and your doctor with this we ask you inform your doctor of any changes in your contact details (for example change of address) and any major changes in your medical condition,, for example the loss of ability to walk unassisted.. Your DNA does not change and the information from your SMN1 genetic test, once entered, will not need to be updated.

“Who will have access to my medical records?”

Staff in charge of the Australian SMA Registry might need to gain access to your medical records to obtain information necessary to the project (for example, we might need to ask your neurologist or geneticist to give us access to a copy of your genetic report). Only people specifically authorised by the Registry will be able to do this.

“How will I be identified in the Registry?”

Your personal details (name, address etc.) and those of your doctor have to be stored in the Australian National SMA Registry so that we can contact your doctor to inform you about possible clinical trials or anything else that might be relevant to your disease. This data will be stored in a secure manner and your records will be assigned **a unique code**. When we transfer your data to the global Treat-NMD registry, we will not transfer any of your personal details, and your records will only be identifiable by the code they have been assigned. Researchers searching in the global registry therefore cannot identify you personally from the information they can access. Only the person in charge of the Australian National SMA Registry (Dr Hugh Dawkins, Office of Population Health Genomics, Department of Health in Western Australia) or a person explicitly appointed by him will be able to “de-code” the data to get access to your personal details in relation to Treat-NMD or other enquiries.

“Will my data be kept confidential?”

Your data will be kept for an indefinite period in the Australian National SMA Registry, under the responsibility of the Director of Population Health Genomics, Department of Health in Western Australia.

“Will my relationship to any affected family members or relatives be linked to my record?”

¹ As clinical trial information becomes available the Registry will forward this information to support groups and doctors. In performing this service we are not making any recommendation about the trial or suggesting that it might be of any benefit to you. The Registry will simply act as a contact organisation through which international companies and overseas researchers can have information about their work and any clinical trials distributed to people affected by SMA.

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It is very useful for the Registry to have a record of your family history. The Registry proposes to link your record, using only your unique registry code. The link will show your unique registry code, your relationship to all consented and registered affected family member. Only those people with access specifically to your records will be able to see your details. They will not see any details about your relative other than their unique identifier and their relationship to you. The same restrictions will apply to their records and the registry link to you.

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A registry is a stored electronic record of a patient's personal and clinical data and the genetic test result of the SMN1 gene that has caused their muscular atrophy and their SMN2 gene dosage. These data are stored together in a coded file. This file will be subject to the regulations on data protection^{2,3} and we will only transfer non-identifiable data to the TREAT-NMD global operates under national laws^{1,2} and bearing relation to the EU directive 95/46 on personal data protection. All information we receive from you will be treated confidentially, encrypted and stored securely.

If we publish any research or other documents based on data from the registries, this research will never identify you by name.

Third parties wishing to have access to data in the Australian National SMA Registry or the TREAT-NMD global registry (such as researchers or companies planning clinical trials or conducting research on new treatments) will **only have access to anonymous information identifiable only by a code**. Before they are granted access even to this anonymous information, they have to have the approval of a Human Research Ethics Committee. Your data will not be made available to employers, governmental organizations, insurance companies or educational institutions.

"How will I benefit from registering?"

This registry is intended as a public service for the benefit of patients living with SMA. You will not receive any payment or any other financial benefit as a result of submitting your data to the registry. The results of research facilitated by the registry may be patentable or may have commercial potential. However, you will not receive patent rights and will not receive financial benefits from future commercial development. Nevertheless, there may be other benefits to participating, including the following:

- We will inform you if (on the basis of the information you provide) you might be a suitable candidate for a certain clinical trial.
- We will also inform you if we receive any new information on your disease which might be of interest to you – for example if we find better ways of caring for patients with SMA.
- The data collected might also provide benefits to other patients with your disease, for example by revealing statistics on how many people in Europe and in each of the other contributing countries have the same condition, or providing information for researchers interested in the best standards of care for your disease.
- We will publish some general statistical information from the Australian registry and from the other European and national registries on our website.

"I want to be involved in a clinical trial. If I register, is this guaranteed?"

Although one of the main aims of this registry is to make it easier for patients to be recruited for clinical trials, there is no guarantee that registering your details will

² The Federal Privacy Act 1988 is Australia's national law for the protection of personal information when handled by Federal and ACT Government Agencies and many private sector organisations. Within the Act, eleven Information Privacy Principles have been developed to govern things such as the collection, storage, use and disclosure of personal information. The Principles also provide individuals with certain rights to access their personal information and correct any errors.

³ National Health & Medical Research Council (NHMRC) National Statement on Ethical Conduct in Human Research (2007); The primary purpose "... is the protection of the welfare and the rights of participants in research..." and the secondary purpose "... is to facilitate research that is or will be of benefit to the researcher's community or to humankind..".

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ensure you will be involved in a clinical trial. If you are interested in receiving details of trials you might be eligible for, please tick the appropriate box at the end of this form. However, it is important that you understand that mounting clinical trials is very complex and criteria for inclusion are sometimes subject to change to meet regulatory and trial design. Consequently, even if you are contacted or believe that you might be eligible for a specific trial based on your registry data, it is still possible that during the assessment process you might not meet all the essential trial inclusion criteria after all.

"I don't want to be involved in a clinical trial. Should I still register?"

We hope you will be interested in registering even if you don't want to take part in a trial. Your information will still be useful to researchers who are trying to find out more about patients living with SMA, and we will still provide you with other information that might be relevant to your disease. If you do not want to receive any information about clinical trials that you might be eligible for, please tick "no" in question 3 of the informed consent section at the end of this form.

"Do I have to participate in the registry and can I withdraw if I change my mind?"

Your participation in this project is completely voluntary. The Federal Privacy Act and associated principles and guidelines^{1,2} grants you the right to rectify your data or withdraw from further participation in the registry at any time. Should you wish to withdraw from the registry you will be free to do so without having to provide any explanation. If you wish to withdraw, you should get in touch with the staff in charge of the Australian National SMA Registry. Contact details are provided below.

"Who should I contact if I have any questions?"

If you would like any additional information or need to tell us about any change in your data, or if you wish to withdraw your data from the registry, please contact the Office of Population Health Genomics
tel. (08) 9222 6888
email genomics@health.wa.gov.au.

The current coordinator of the National SMA Registry is Dr Hugh Dawkins.

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INFORMED CONSENT

1. Do we have your permission to store your personal & clinical data in the Australian National SMA Registry and to transfer it (in a form identifiable only by a code) to the global TREAT-NMD registry in which it may be used for research and for the planning of clinical trials? NO YES
2. Do we have your permission to obtain your SMA genetic test (SMN1 gene sequence and SMN2 gene dosage) result from the relevant testing laboratory and to store this information with your clinical and personal information in the Australian National SMA Registry and to transfer it (in a form identifiable only by a code) to the global TREAT-NMD registry where it may be used for research and for the planning of clinical trials? NO YES
3. If we receive information on TREAT-NMD projects or other information related to your disease which might be relevant to you, would you like to be informed about this? NO YES
4. If your doctor receives information about a clinical trial which you might be eligible for, would you like to be informed about this? ⁴ NO YES
5. So that we can keep the registry up to date, we will need to update your records once a year. Do you agree to receive follow-up forms once a year which you will be asked to complete in order to register any changes in your medical condition or contact details? NO YES
6. To improve the quality of the family history data on the Registry, we propose to link your record to any other affected family member or relative on the Registry. The link will only show your Unique identification number and your relationship to the affected relative. Do you agree to have your record linked to any other affected relatives on the Registry? NO YES
7. If there are any major changes in your data (for example change of address or changes in your medical condition, such as loss of ability to walk unassisted) that occur in the period between updates, are you willing to inform us? NO YES

⁴ **Please note** that if we inform your doctor about the existence of a trial, this does not imply that we endorse it. In order to participate in any trial, you will need to discuss it with your family and your doctor and will be required to fill out a separate informed consent form that relates to that specific trial.

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The nature of the registry has been fully explained to me. I have understood the patient information and informed consent form and have received a copy to take away with me. I have had the opportunity to ask questions, and all my questions have been answered to my satisfaction. Upon reflection, I agree to participate in this registry.

Signature of participant

Date

Signature of parent/guardian

Date

(Required if the participant is a child 15 years old or younger)

First name:

Family name:

Address:

Telephone:

Mobile phone:

Email:

If you are wanting to initiate registration by directly contacting the SMA Registry please provide the name of your doctor below giving us permission to contact your doctor if we need further information to complete your registration

You have my/our permission to contact my doctor for my personal/our child's/ details:

Doctors First name:

Doctors Family name:

Clinic / Medical Practice Address:

Clinic / Medical Practice Telephone:
