Registration form for the NZ NMD Disease Registry
Spinal Muscular Atrophy [SMA_DATA]

Thank you for agreeing to participate in the New Zealand Neuromuscular Disease Registry. Please ensure you have read and signed the participant information sheet and consent form. To complete your registration you will need to fill in this form and return this form. You may like to complete it with the assistance of your doctor. Alternatively, if you are not certain about the answer to any question please discuss this with the registry curator.

MANDATORY ITEMS
(For your details to be included in the global registry you must complete all of these questions.

I am: (please tick as appropriate)

	The participant
	The participantôs representative

All of the following questions relate to the participantôs with the condition

1. Participant's personal details:

Sex:	male / female (delete as appropriate)		
First name(s):			
Family name:			
Date of birth:	/ / (dd / mm / yyyy)		
	NZ European	Maori	Samoan
	Cook Island Maori	Tongan	Niuean
	Chinese	Indian	Other
NHI number:			
Address			
Postcode:			
Email:			
Home Phone:			
Mobile:			

2. Please provide the name of your GP below giving us permission to contact your GP directly if we require further information to complete your registration.

GPs Full name:	
Medical Practice Address	
Email:	
Medical Practice Phone:	

3. If you are the participant's representative (parent/guardian), please provide your details:

Full name:	
Address	
Email:	
Phone:	
Relationship to participant	

4. What is the participant's genetic test result?

It is very important that your genetic test result is entered correctly into the registry. If you do not know your exact result, or you are not sure how to fill it in correctly, please provide the name of the hospital or doctor who requested your test. The registry curator will contact them to ask for the result on your behalf. Please include a copy of your test result if you have one.

Mutation name in SMN1 gene following HGVS rules (based on cDNA Ref Seq):
c._____ (on one allele) c._____ (on other allele)

5. What is the participant's diagnosis, according to your doctor?

	Spinal Muscular Atrophy
	Other
	I don't know

6. Current mobility (please tick the most appropriate answer): Walking

	The participant is currently able to walk (with or without help/support)
	The participant is not currently able to walk

7. Sitting

	The participant is currently able to sit independently (without the support of their arms, or without leaning against the back of the chair)
	The participant is not currently able to sit independently

8. Best motor function ever achieved
What is the best level of movement the participant has ever managed? Please provide an age range. (This question is not about the participant's current condition, but about the time when his/her motor skills were at their best.)

	The participant was able to walk (with or without help/support) (**from** age ____years _____ months **until** age ____years _____ months)

	The participant was never able to walk, but was able to sit independently (without the support of their arms or without leaning against the back of the chair) (**from** age ____years _____ months **until** age ____years _____ months)
	The participant was never able to walk or to sit independently

9. If the participant is 3 years old or more, does he/she have to use a wheelchair?

	The participant uses a wheelchair permanently (started full-time use at age: _____ years) (The participant uses a wheelchair any time he/she needs to get around)
	The participant uses a wheelchair part-time/intermittently (started at age: _____ years) (The participant can walk short distances without the wheelchair and uses it only for longer distances)
	The participant has never used a wheelchair
	Unknown

10. Has the participant had spinal surgery for scoliosis?
Scoliosis is a deformation or ñbendingò of the spine which may have required surgery.

	Yes
	No
	Unknown

11. Does the participant have a tube (gastric/nasal) for feeding?
Does the participant have a tube that goes into their stomach either through the nose, or directly into the stomach through an incision in their stomach?

	Yes
	No
	Unknown

12. Is the participant currently included in a clinical trial?

	He or she is currently not included in a clinical trial, but has previously participated in the following trial:
	No, he or she has never participated in a clinical trial
	Unknown

HIGHLY ENCOURAGED ITEMS
(We can still include your details in the global registry even if you canôt answer all of these questions, but please answer as many of them as you can)

13. Does the participant regularly use a non-invasive ventilation device?
A non-invasive ventilation device is a device to support breathing, that has not required an operation to use, for example a mask worn on the face (CPAP, BIPAP or VPAP)

	Yes, all day
	Yes, but only part-time (e.g. at night)
	No, never
	Unknown

14. Does the participant use invasive ventilation?
An invasive ventilation device requires an operation to use, for example a tracheostomy.

	Yes, all day
	Yes, part-time
	No
	Unknown

If the participant has had pulmonary function testing, please fill in the result if you know it:

FVC (Forced Vital Capacity) % (predicted value)	
Date of the test:	

15. Has the participant signed up for any other SMA registry?

	Yes (if yes, please specify:
	No
	Unknown

16. Does anybody else in the participant's family have the same kind of disease?

	Yes (if yes, please specify:
	No
	Unknown

17. Has the participant been classified into an SMA subgroup?

	SMA Type 1
	SMA Type 2
	SMA Type 3
	Unknown

18. Has the participant's SMN2 copy number been tested?

Testing the SMN2 copy number is another form of genetic testing which is sometimes done in addition to the test for deletion/mutations of the SMN1 gene.

	Yes (if yes, please enter the result here:
	No
	Unknown

Thank you for completing this form if you have any questions please do not hesitate in calling the Curator of the registry on **09-815-0247** or talk to your health care provider.