

Registration form for the TREAT-NMD (UK: / Jennifer Trust) SMA Patient Registry

To be included in the SMA Patient Registry, you need to fill in this registration form and also read and sign the patient information and informed consent form.

Before you start filling in this form, please read the instructions provided. The instructions provide explanations of a lot of the things that might sound a little complicated at first.

MANDATORY ITEMS

(If you don't answer all of these questions, we won't be able to include your details in the global registry.)

I am:

- The patient
 The patient's representative *(please tick as appropriate)*

1. Patient's personal details:

Sex: male / female *(delete as appropriate)*

First name(s): _____

Family name: _____

Date of birth: ___ / ___ / _____ (dd / mm / yyyy)

(UK) NHS number: _____

Address
(incl. postcode): _____

Email: _____

Phone: _____

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

Name: _____

Address: _____

Email: _____

Phone: _____

Relationship to
patient: _____

3. What is the patient's genetic test result?

Mutation name in *SMN1* gene following HGVS rules (based on cDNA Ref Seq):

c. _____ (on one allele) c. _____ (on other allele)

4. What is the patient's diagnosis, according to your doctor?

- Spinal Muscular Atrophy
- Other
- I don't know

5. Current best motor function (please tick the most appropriate answer):

- Patient is currently able to walk (with or without help/support)
- Patient is currently not able to walk, but is currently able to sit independently without support
- Patient is currently neither able to walk nor to sit independently

6. Best motor function ever achieved

(please tick the most appropriate answer and specify an age range where appropriate):

- Patient was able to walk (with or without help/support)
(**from** age ____years _____ months **until** age ____years _____ months)
- Patient was never able to walk, but was able to sit independently without support
(**from** age ____years _____ months **until** age ____years _____ months)
- Patient was never able to walk or to sit independently

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

- Patient uses a wheelchair permanently (started full-time use at age: _____ years)
- Patient uses a wheelchair part-time/intermittently (started at age: _____ years)
- Patient has never used a wheelchair
- Unknown

8. Has the patient had spinal surgery for scoliosis?

- Yes
- No
- Unknown

9. Does the patient have a tube (gastric/nasal) for feeding?

- Yes
- No
- Unknown

10. Is the patient currently included in a clinical trial?

- Yes, he/she is currently included in one of the following clinical trials:
 - CARNIVAL Type 1 (Valproic Acid and Carnitine in SMA Type 1)
 - Pilot Study of Growth Hormone in SMA Type 2 and 3
- He/she is currently not included in a clinical trial, but has previously participated in the following trial:
(Currently there is nothing to be listed here.)
- No, he/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 can't answer all of these questions, but please answer as many of them as you can.)

11. Does the patient regularly use a non-invasive ventilation device?

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

12. Does the patient use invasive ventilation?

- Yes, all day
- Yes, part-time
- No
- Unknown

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

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

13. Has the patient signed up for any other SMA registry?

- Yes (if yes, please specify: _____)
- No
- Unknown

14. Does anybody else in the patient's family have the same kind of disease?

- Yes (if yes, please specify: _____)
- No
- Unknown

15. Has the patient been classified into an SMA subgroup?

- SMA Type 1
- SMA Type 2
- SMA Type 3
- Unknown

16. Has the patient's SMN2 copy number been tested?

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