



**SECTION B: INFORMATION ABOUT THE PERSON WITH SMA**

Name of patient: \_\_\_\_\_  
(First) (Middle) (Maiden) (Last)

Date of Birth: \_\_\_\_\_ Sex: \_\_\_\_\_  
(Month) / (Day) / (Year) (M or F)

Living: Yes \_\_\_\_ No \_\_\_\_  
If deceased, date of death: \_\_\_\_\_

Birthplace (City or County, and State): \_\_\_\_\_

State of Last or Current Residence: \_\_\_\_\_

Racial Background (circle one):

- a. American Indian or Alaskan Native
- b. Asian
- c. African American, not of Hispanic origin
- d. Caucasian, not of Hispanic origin
- e. Native Hawaiian / Pacific Islander
- f. Hispanic
- g. Other (Please Specify) \_\_\_\_\_
- h. Unknown

**SECTION C: CLINICAL HISTORY**

At what age was the patient diagnosed as having SMA? \_\_\_\_\_

Did a physician diagnose the disease? \_\_\_\_\_

If so, please give us the name and address of the physician and sign the medical release form attached to this questionnaire. This will allow us to obtain copies of the medical records concerning the patient; these records are useful in confirming the diagnosis and form of SMA.

Physician's name: \_\_\_\_\_

Physician's address: \_\_\_\_\_

What tests were used to make the diagnosis of SMA? (Check all that apply.)

- 1. \_\_\_\_\_ Serum enzymes, i.e. creatine phosphokinase or CPK
- 2. \_\_\_\_\_ Electromyography
- 3. \_\_\_\_\_ Muscle biopsy
- 4. \_\_\_\_\_ DNA test done for SMN deletion
- 5. \_\_\_\_\_ Clinical (medical opinion based upon exam)
- 6. \_\_\_\_\_ Other (give name of test): \_\_\_\_\_
- 7. \_\_\_\_\_ Do not know

According to the patient's physician, what form of spinal muscular atrophy does the patient have? (Circle one.)

- A. Infantile muscular atrophy (also known as Werdnig-Hoffmann Disease, spinal muscular atrophy I and SMA I. Patients typically can not sit without support)
- B. Intermediate muscular atrophy (also known as spinal muscular atrophy II and SMA II. Patients typically can sit without support and may stand without bracing but can not walk without bracing)
- C. Juvenile muscular atrophy (also known as Kugelberg-Welander syndrome, KWS, spinal muscular atrophy III and SMA III. Patients typically can, or were able to at one time, walk without assistance of a walker or bracing)
- D. Adult type proximal spinal muscular atrophy (also known as Finkel late-adult type SMA)
- E. Spinal and bulbar muscular atrophy (also known as Kennedy disease and bulbospinal muscular atrophy)
- F. Other (please specify) \_\_\_\_\_
- G. Do not know

What were the first signs of the disease? (Circle all that apply.)

- 1. Difficulty in feeding or swallowing
- 2. Floppiness or poor muscle tone
- 3. Breathing difficulty
- 4. Other (please list) \_\_\_\_\_
- 5. Do not know

The patient has achieved which of the following motor (physical) skills? (Circle all that apply.)

- 1. While lying on his/her back, patient brings hand to mouth
- 2. Patient can fully roll onto his/her side
- 3. Patient can sit with support on a straight back chair
- 4. Patient can sit with support on the floor
- 5. Patient can sit without support on a straight back chair
- 6. Patient can sit without support on the floor
- 7. When sitting, patient can lift one hand above shoulder without assistance
- 8. When sitting, patient can lift both hands above shoulder without assistance
- 9. Patient can get into the sitting position, without assistance, from lying down
- 10. Patient can crawl on hands and knees
- 11. Patient can get into the standing position, without assistance, from the sitting position
- 12. Patient can stand with support, using a walker, assistance of a person, braces and/or stander
- 13. Patient can stand without support
  - If so, for how long (circle one)
  - a. less than one minute
  - b. 1 to 3 minutes
  - c. as long as desired
- 14. Patient can walk with assistance, using a walker, assistance of a person, braces and/or stander
- 15. Patient can walk without assistance

At what age were the patient's motor skills maximized (i.e.: at what age did he/she have the most physical skills)?

Current height:

What was/is the patient's maximum height (feet and inches)? \_\_\_\_\_

At what age did this occur? \_\_\_\_\_

Current weight (pounds): \_\_\_\_\_

What was/is the patient's maximum weight (pounds)? \_\_\_\_\_

At what age did this occur? \_\_\_\_\_

Has the patient received physical therapy (PT) for SMA? Yes / No

If yes, how often (circle one)

- a. Once a month
- b. Twice a month
- c. Three times a month
- d. Four times a month
- e. Other, please specify: \_\_\_\_\_

Has the patient received occupational therapy (OT) for SMA? Yes / No

If yes, how often (circle one)

- a. Once a month
- b. Twice a month
- c. Three times a month
- d. Four times a month
- e. Other, please specify: \_\_\_\_\_

Has the patient received speech therapy (ST) for SMA? Yes / No

If yes, how often (circle one)

- a. Once a month
- b. Twice a month
- c. Three times a month
- d. Four times a month
- e. Other, please specify: \_\_\_\_\_

Has the patient had surgery related to SMA involving the following:

|                             | Circle yes or no | If yes, at what age was this surgery performed |
|-----------------------------|------------------|--|
| 1. Hip                      | Yes / No         |  |
| 2. Foot                     | Yes / No         |  |
| 3. Ankle                    | Yes / No         |  |
| 4. Spinal fusion            | Yes / No         |  |
| 5. Other (specify)<br>_____ | Yes / No         |  |

Does the patient have difficulty swallowing? Yes / No

Does the patient use a feeding tube? Yes / No

If yes, circle the type of feeding tube used

- a. G tube  
age it was used: \_\_\_\_\_
- b. Ng tube  
age it was used: \_\_\_\_\_
- c. Other, please specify: \_\_\_\_\_  
age it was used: \_\_\_\_\_

Is patient currently using any of the following respiratory therapies? (Circle all that apply.)

- a. ventilation using a trach
- b. ventilation using a mask
- c. cpap using a trach
- d. cpap using a mask
- e. bipap using a trach
- f. bipap using a mask
- g. ThAIRapy™ vest
- h. In-exsufflator/cough machine

Is patient currently on oxygen? Yes / No

Has patient been on oxygen previously? Yes / No

Is patient taking any of the following supplements? (Circle all that apply.)

- a. vitamins/minerals (some examples are multivitamin, vitamin C, zinc)  
If yes, please list them: \_\_\_\_\_
- b. herbal (some examples are St. Johns wort, rosemary, ginkgo biloba)  
If yes, please list them: \_\_\_\_\_
- c. protein/amino acids (some examples are soy proteins, creatine, glycine, coenzyme Q10)  
If yes, please list them: \_\_\_\_\_
- d. nutritional (examples are Ensure, Carnation )  
If yes, please list them: \_\_\_\_\_
- e. Other (specify): \_\_\_\_\_
- f. No supplements are taken

Is patient taking any prescription medication for SMA? Yes / No

If yes, please list them: \_\_\_\_\_

Does patient have any major medical problems other than SMA? Yes / No

If yes, please list them: \_\_\_\_\_

**SECTION D: FAMILY INFORMATION**

Have other family members been diagnosed with SMA? Yes / No

If yes, please list their relationship to the patient (e.g. brother, mother) and type of SMA

|    | Relationship to patient | Type of SMA (refer to pg 3) |
|----|-------------------------|-----------------------------|
| 1. |                         |                             |
| 2. |                         |                             |
| 3. |                         |                             |

Are other family members participating in the International SMA Patient Registry? Yes / No

Thank you for completing this questionnaire. If you have questions or if other family members are interested in participating in the International SMA Patient Registry, please have them contact either the International SMA Patient Registry at Indiana University School of Medicine, Department of Medical and Molecular Genetics at (317) 274-5745 or Families of SMA at (800) 886-1762.