

	Item	Self-Report Example
	<b>Mandatory Items</b>	
1.	<b>Personal data</b> Sex First name Last name Date of birth Address Zip/post code Telephone Email	<b>Your personal data:</b> Sex: First name: Last name: Date of birth: Address: Zip/post code: Telephone: Email:
2.	<b>Genetic Test Result:</b>  Mutation name in SMN1 gene following HGVS rules (based on cDNA Ref Seq)	<b>What is your genetic test result?</b> Mutation name in SMN1 gene following HGVS rules (based on cDNA Ref Seq): c. _____ (on one allele) c. _____ (on other allele)
3.	<b>Clinical Diagnosis</b> <input type="radio"/> Spinal Muscular Atrophy <input type="radio"/> Other (specify...) <input type="radio"/> Unknown	<b>What is your diagnosis, according to your doctor?</b> <input type="radio"/> Spinal Muscular atrophy (SMA) <input type="radio"/> Other <input type="radio"/> I don't Know
4.	<b>Currently Able to Walk</b> <input type="radio"/> Yes <input type="radio"/> No	<b>What is your Current best motor function (please tick the most appropriate answer):</b> <input type="radio"/> I am currently able to walk (with or without help/support) <input type="radio"/> I am currently not able to walk, but is currently able to sit independently without support <input type="radio"/> I am currently neither able to walk nor to sit independently
5.	<b>Best motor function achieved</b> <input type="radio"/> Walking [specify age of acquisition] <input type="radio"/> Sitting independently [specify age of acquisition] <input type="radio"/> Never able to walk or sit independently	<b>What is the best motor function you have ever achieved?</b> <input type="radio"/> Walking From age...years...months until age...years....months <input type="radio"/> Sitting Independently From age...years...months until age...years....months <input type="radio"/> Never able to walk to sit independently
6.	<b>Wheelchair use (if over 3 years of age)</b> <input type="radio"/> No <input type="radio"/> Part time (age...) <input type="radio"/> Full-time (age...)	<b>Do you use a wheelchair? (please tick the most appropriate answer)</b> <input type="radio"/> No, not at all <input type="radio"/> I use a wheelchair part-time (I started at age: .....) <input type="radio"/> I use a wheelchair all the time (I started full-time use at age: .....)
7.	<b>Gastric/nasal feeding tube</b> <input type="radio"/> Yes <input type="radio"/> No	<b>Do you use a gastric or nasal tube for feeding?</b> <input type="radio"/> Yes <input type="radio"/> No
8.	<b>Scoliosis Surgery</b> <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown	<b>Have you ever had surgery for Scoliosis?</b> <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown

SMA Registry Core Data

9.	<p><b>Currently included in a clinical trial</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes, Currently (name of drug.....)</li> <li><input type="radio"/> No, but preciously</li> <li><input type="radio"/> Never</li> <li><input type="radio"/> Unknown</li> </ul>	<p><b>Are you currently taking part in a clinical trial?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes, Currently</li> <li><input type="radio"/> No, but previously</li> <li><input type="radio"/> Never</li> <li><input type="radio"/> Unknown</li> </ul> <p><b>If yes please specify the name the drug being tested.....</b></p>
Highly Encouraged Items		
Pulmonary Function		
10	<p><b>Non-invasive ventilation</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Full-time</li> <li><input type="radio"/> Part-time</li> <li><input type="radio"/> None</li> </ul>	<p><b>Do you regularly use a non-invasive ventilation device?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes, all day</li> <li><input type="radio"/> Yes, but only part-time (e.g. at night)</li> <li><input type="radio"/> No, never</li> </ul>
11	<p><b>Invasive ventilation</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Full-time</li> <li><input type="radio"/> Part-time</li> <li><input type="radio"/> None</li> </ul>	<p><b>Do you use invasive ventilation?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes, all day</li> <li><input type="radio"/> Yes, part-time</li> <li><input type="radio"/> No</li> </ul>
12	<p><b>Pulmonary function testing</b></p> <p>FVC done: yes/no/unknown</p> <p>FVC: ...%</p> <p>Date</p>	<p><b>Have you had pulmonary function testing?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes,</li> <li><input type="radio"/> No</li> <li><input type="radio"/> I don't know</li> </ul> <p><b>If yes, please fill in the results of the test:</b></p> <p>FVC .....% (predicted value)</p> <p>Date of the test:.....</p>
Other Registry		
13	<p><b>Other registry</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes (specify...)</li> <li><input type="radio"/> No</li> <li><input type="radio"/> Unknown</li> </ul>	<p><b>Have you signed up for any other SMA registry?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes (if yes, please specify: .....)</li> <li><input type="radio"/> No</li> <li><input type="radio"/> I don't know</li> </ul>
Family History		
14	<p><b>Positive family history</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes</li> <li><input type="radio"/> No</li> <li><input type="radio"/> Unknown</li> </ul>	<p><b>Has anybody else in your family been diagnosed with the same disease?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes</li> <li><input type="radio"/> No</li> <li><input type="radio"/> I don't know</li> </ul> <p>If yes please specify the relation to you.....</p>
SMA Classification		
15	<p><b>SMA Classification</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> SMA1</li> <li><input type="radio"/> SMA2</li> <li><input type="radio"/> SMA3</li> </ul>	<p><b>Have you been classified into an SMA subgroup?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> SMA Type 1</li> <li><input type="radio"/> SMA Type 2</li> <li><input type="radio"/> SMA Type 3</li> <li><input type="radio"/> Unknown</li> </ul>
Molecular Data		
16	<p><b>Number of SMN2 copies</b></p>	<p><b>Has your SMN2 copy number been tested?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes (if yes, please enter the result here: _____)</li> <li><input type="radio"/> No</li> <li><input type="radio"/> Unknown</li> </ul>