

**Core dataset for international FSHD registry – final revision 24 May 2011**

	<b>Item</b>	<b>Self-report example</b>
	<b>Mandatory items</b>	
<b>1.</b>	<b>Personal data</b>  Sex First name Last name Date of birth Address Zip/post code Telephone Email Country	<b>Your personal data:</b>  Sex: First name: Last name: Date of birth: Address: Zip/post code: Telephone: Email: Country:
<b>2.</b>	<b>Genetic test result</b>  <input type="radio"/> Confirmed FSHD1 (with details) <input type="radio"/> Confirmed FSHD2 (with details) <input type="radio"/> Result pending <input type="radio"/> Not tested	<b>What is your genetic test result?</b>  <input type="radio"/> I have been told I have genetically confirmed FSHD and I can provide a copy of my genetic test result <input type="radio"/> I have been told I have genetically confirmed FSHD and I give the registry permission to ask my doctor for my genetic test result <input type="radio"/> I have been tested but I haven't received the result yet <input type="radio"/> I have not been tested
<b>3.</b>	<b>Clinical Diagnosis</b>  <input type="radio"/> no signs or symptoms <input type="radio"/> Facial weakness <input type="radio"/> Periscapular shoulder weakness <input type="radio"/> Foot dorsiflexor weakness <input type="radio"/> Hip girdle weakness	<b>Which of these symptoms do you have? (Tick all that apply)</b>  <input type="radio"/> I have no signs or symptoms of muscle weakness <input type="radio"/> Facial weakness (weakness of muscles in the face causing e.g. inability to smile, to whistle, or to close your eyes fully at night) <input type="radio"/> Shoulder weakness (weakness of the muscles around the shoulder blades causing e.g. inability to raise your arms sideways above the level of your shoulder) <input type="radio"/> Foot weakness (weakness of the muscles that help you lift your feet up, causing e.g. foot drop (where the foot tends to hang with the toes pointing down), steppage gait (lifting the feet high when walking), or frequent tripping) <input type="radio"/> Hip girdle weakness (weakness of the muscles of the pelvis and top of the legs, causing e.g. difficulties in going up stairs or ladders, rising from a chair or getting up from the floor)
<b>4.</b>	<b>Current best motor function</b>  <input type="radio"/> Ambulatory (unassisted) <input type="radio"/> Ambulatory (assisted) <input type="radio"/> Non-ambulatory	<b>Which of the following options describes the best motor function you are currently able to achieve? (please tick the most appropriate answer)</b>  <input type="radio"/> I can walk unaided (without an assistive device) <input type="radio"/> I can walk with an assistive device (walker, brace, cane, etc) <input type="radio"/> I cannot walk
<b>5.</b>	<b>Wheelchair use</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: .....) .....

Highly encouraged items		
PULMONARY		
<b>6.</b>	<b>Non-invasive ventilation</b> <ul style="list-style-type: none"> <li><input type="radio"/> Full-time (start date month/year)</li> <li><input type="radio"/> Part-time (start date month/year)</li> <li><input type="radio"/> None</li> </ul>	<b>Do you regularly use a non-invasive (mask) ventilation device?</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes, all day (started in ....)</li> <li><input type="radio"/> Yes, but only part-time, e.g. at night (started in ....)</li> <li><input type="radio"/> No, never</li> </ul>
<b>7.</b>	<b>Invasive ventilation</b> <ul style="list-style-type: none"> <li><input type="radio"/> Full-time (start date month/year)</li> <li><input type="radio"/> Part-time (start date month/year)</li> <li><input type="radio"/> None</li> </ul>	<b>Do you use invasive ventilation?</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes, all day (started in ....)</li> <li><input type="radio"/> Yes, part-time (started in ....)</li> <li><input type="radio"/> No</li> </ul>
OTHER		
<b>8.</b>	<b>Age of onset for selected FSHD symptoms (taken from question 3)</b> <ul style="list-style-type: none"> <li><input type="radio"/> Facial weakness (start date month/year)</li> <li><input type="radio"/> Periscapular shoulder weakness (start date month/year)</li> <li><input type="radio"/> Foot dorsiflexor weakness (start date month/year)</li> <li><input type="radio"/> Hip girdle weakness (start date month/year)</li> </ul>	<b>At what age did symptoms related to your FSHD first occur (give date for all that apply, as in question 3)?</b> <ul style="list-style-type: none"> <li><input type="radio"/> Facial weakness (first occurred in ....)</li> <li><input type="radio"/> Shoulder weakness (first occurred in ....)</li> <li><input type="radio"/> Foot weakness (first occurred in ....)</li> <li><input type="radio"/> Hip girdle weakness (first occurred in ....)</li> </ul>
<b>9.</b>	<b>Retinal vascular disease attributable to FSHD</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes (start date month/year)</li> <li><input type="radio"/> No</li> <li><input type="radio"/> Unknown</li> </ul>	<b>Have you been diagnosed with retinal vascular disease (problems with the retina of your eye causing e.g. loss of vision) that your doctors think may be related to your FSHD?</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes (first occurred in ....)</li> <li><input type="radio"/> No</li> <li><input type="radio"/> I don't know</li> </ul>
<b>10.</b>	<b>Hearing loss</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes (start date month/year)</li> <li><input type="radio"/> No</li> <li><input type="radio"/> Unknown</li> </ul>	<b>Do you have hearing loss?</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes (first occurred in ....)</li> <li><input type="radio"/> No</li> <li><input type="radio"/> I don't know</li> </ul>
<b>11.</b>	<b>Scapular fixation</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes, bilateral (surgery dates month/year)</li> <li><input type="radio"/> Yes, unilateral (surgery date month/year)</li> <li><input type="radio"/> No</li> </ul>	<b>Have you had scapular fixation (an operation to fix your shoulder blade to your ribcage)?</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes, both shoulders (operated in ..... and in ....)</li> <li><input type="radio"/> Yes, one shoulder (operated in ...)</li> <li><input type="radio"/> No</li> </ul>
<b>12.</b>	<b>Pregnancy (only females)</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes <ul style="list-style-type: none"> <li>• Number of pregnancies</li> <li>• Number of childbirths</li> </ul> </li> <li><input type="radio"/> No</li> </ul>	<b>(For women only:) Have you ever been pregnant?</b> <ul style="list-style-type: none"> <li><input type="radio"/> Yes <ul style="list-style-type: none"> <li>• If yes, how many times have you been pregnant? (count the number of pregnancies even if you did not have the baby)</li> <li>• If yes, how many children do you have?</li> </ul> </li> <li><input type="radio"/> No</li> </ul>

<p><b>13.</b></p>	<p><b>Positive family history</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Affected mother</li> <li><input type="radio"/> Affected father</li> <li><input type="radio"/> Affected sibling(s)</li> <li><input type="radio"/> Other affected relative</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 FSHD (tick all that apply)?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Yes, mother</li> <li><input type="radio"/> Yes, father</li> <li><input type="radio"/> Yes, one or several of my siblings (brothers and sisters)</li> <li><input type="radio"/> Yes, further relatives (other than parents and siblings)</li> <li><input type="radio"/> No</li> <li><input type="radio"/> I don't know</li> </ul>
<p><b>14.</b></p>	<p><b>Ethnic origin</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> Caucasian</li> <li><input type="radio"/> Black African/African American</li> <li><input type="radio"/> Asian</li> <li><input type="radio"/> Mixed</li> <li><input type="radio"/> Other</li> <li><input type="radio"/> Declined</li> </ul>	<p><b>How would you describe your ethnic origin?</b></p> <ul style="list-style-type: none"> <li><input type="radio"/> White - European origin (Caucasian)</li> <li><input type="radio"/> Black African/African American</li> <li><input type="radio"/> Asian</li> <li><input type="radio"/> Mixed</li> <li><input type="radio"/> Other</li> <li><input type="radio"/> I choose not to answer this question</li> </ul>
<p><b>15.</b></p>	<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 FSHD 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>