Core dataset for international FSHD registry

	Item	Self-report example
	Mandatory items	
1.	**Personal data** Sex First name Last name Date of birth Address Zip/post code Telephone Email Country	**Your personal data**: Sex: First name: Last name: Date of birth: Address: Zip/post code: Telephone: Email: Country:
2.	**Genetic test result** o Confirmed FSHD1 (with details) o Confirmed FSHD2 (with details) o Result pending o Not tested	**What is your genetic test result?** o I have been told I have genetically confirmed FSHD and I can provide a copy of my genetic test result o I have been told I have genetically confirmed FSHD and I give the registry permission to ask my doctor for my genetic test result o I have been tested but I haven't received the result yet o I have not been tested
3.	**Clinical Diagnosis** o no signs or symptoms o Facial weakness o Periscapular shoulder weakness o Foot dorsiflexor weakness o Hip girdle weakness	**Which of these symptoms do you have? (Tick all that apply)** o I have no signs or symptoms of muscle weakness o Facial weakness (weakness of muscles in the face causing e.g. inability to smile, to whistle, or to close your eyes fully at night) o 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) o 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) o 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)
4.	**Current best motor function** o Ambulatory (unassisted) o Ambulatory (assisted) o Non-ambulatory	**Which of the following options describes the best motor function you are currently able to achieve? (please tick the most appropriate answer)** o I can walk unaided (without an assistive device) o I can walk with an assistive device (walker, brace, cane, etc) o I cannot walk
5.	**Wheelchair use** o No o Part-time (age...) o Full-time (age ...)	**Do you use a wheelchair? (please tick the most appropriate answer)** o No, not at all o I use a wheelchair part-time (I started at age: ……..) o I use a wheelchair all the time (I started full-time use at age: ……)

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

13.	**Positive family history** o Affected mother o Affected father o Affected sibling(s) o Other affected relative o No o Unknown	**Has anybody else in your family been diagnosed with FSHD (tick all that apply)?** o Yes, mother o Yes, father o Yes, one or several of my siblings (brothers and sisters) o Yes, further relatives (other than parents and siblings) o No o I don't know
14.	**Ethnic origin** o Caucasian o Black African/African American o Asian o Mixed o Other o Declined	**How would you describe your ethnic origin?** o White - European origin (Caucasian) o Black African/African American o Asian o Mixed o Other o I choose not to answer this question
15.	**Other registry** o Yes (specify...) o No o Unknown	**Have you signed up for any other FSHD registry?** o Yes (if yes, please specify: ……) o No o I don't know