TREAT-NMD Core Dataset for FSHD v2

	Clinically Reported	Self-reported
	MANDATORY ITEMS	
1a	**Personal data**	**Your* personal data (*Or FSHD patient's, if you are a parent/guardian or caregiver entering data on behalf of a patient)**
	Section one is only collected at the local level and is not to be transmitted to TGDOC	
	Biological Sex at birth	Biological Sex at birth
	First name at birth:	First name at birth:
	Middle name	Middle name
	Surname at birth:	Surname at birth:
	Current surname	Current surname
	Date of birth:	Date of birth:
	Town/city of birth:	Town/city of birth:
	Country of birth	Country of birth
	Address:	Address:
	Zip/post code:	Zip/post code:
	Country:	Country:
	Telephone:	Telephone:
	Email:	Email:
1b	**Alternative contact**	**Alternative contact should you be unavailable**
	Next of kin	Next of kin
	First name	First name
	Last name	Last name
	Current address	Current address
	Zip/post code	Zip/post code
	Country	Country
	Telephone	Telephone
	Email	Email
1c	**Diagnosing physician**	**Physician who diagnosed you with FSHD**
	First name	First name
	Last name	Last name
	Medical institution	Medical institution
	Address	Address
	Zip/post code	Zip/post code
	Country	Country
	Telephone	Telephone
	Email	Email
2	**Diagnosis and genetic test result**	**Diagnosis and your genetic test result**
	Confirmed FSHD1 (D4Z4 contraction 1–10 repeats + 4qA)	I have been told I have genetically confirmed FSHD and I can provide a copy of my genetic test result **[UPLOAD]**
	FSHD2 (no contraction + 4qA + SMCHD1 mutation)	I have been told I have genetically confirmed FSHD but I do not have my genetic test result.**[FOLLOW-UP: OBTAIN GENETIC TEST REPORT FROM DIAGNOSING PHYSICIAN]**
	Clinically confirmed diagnosis but no genetic testing	I have been clinically diagnosed with FSHD but have not been genetically tested
		I have not been tested but wish to be tested
		I have not been tested and do not wish to be tested
3	**Signs or symptoms**	**Which of these symptoms do you have? (Tick all that apply)**
	No signs or symptoms	I have none of the signs or symptoms described above
	Facial weakness	Facial weakness (weakness of muscles in the face causing e.g. inability to smile, to whistle, or to close your eyes fully at night)
	Periscapular shoulder weakness	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)

	Foot dorsiflexor weakness	Foot or ankle weakness (weakness of the muscles that help you lift your feet up, causing e.g. foot drop (where the foot tends thang with the toes pointing down), steppage gait (lifting the feet high when walking), or frequent tripping)
	Hip girdle weakness	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**	**Which of the following options describes the best motor function you are currently able to achieve?**
	Ambulatory (unassisted)	I can walk unaided (without an assistive device)
	Ambulatory (assisted)	I can walk with an assistive device (walker, brace, cane, etc)
	Non-ambulatory	I cannot walk

5	**Wheelchair use**	**Do you use a wheelchair? (please select all that apply)**
	No	I don't use a wheelchair.
	Part-time (start date year)	I started using a wheelchair part-time from [YEAR]
	Full-time (start date year)	I use a wheelchair all the time since [YEAR]
		I started using an assistive device from [YEAR]
	Unknown	Unknown

6a	**Pulmonary function test**	**Has your respiratory capacity ever been evaluated (for example pulmonary function testing)?**
	No	No
	Yes	Yes
	Unknown	Unknown
6b	**Non-invasive ventilation**	**Do you regularly use a non-invasive (mask) ventilation device?**
	Full-time (start date year)	Yes, all day since [YEAR]
	Part-time (start date year)	Yes, but only part-time, e.g. at night, since [YEAR])
	None	No, never
	Unknown	Unknown
6c	**Invasive ventilation**	**Do you use invasive ventilation (requiring surgery, e.g. tracheostomy)?**
	Full-time (start date year)	Yes, full-time since [YEAR])
	Part-time (start date year)	Yes, part-time since [YEAR])
	None	No
	Unknown	I don't know

7	**Age of onset for selected FSHD symptoms (taken from question 3)**	**At what age did symptoms related to your FSHD first occur (give approximate year for all that apply, as in question 3)?**
	Facial weakness (start date **year**)	Facial weakness (first occurred in [**YEAR**])
	Periscapular shoulder weakness (start date **year**)	Shoulder weakness (first occurred in [**YEAR**])
	Foot dorsiflexor weakness (start date **year**)	Foot weakness (first occurred in [**YEAR**])
	Hip girdle weakness (start date **year**)	Hip girdle weakness (first occurred in [**YEAR**])
	Unknown	I don't know

8	**Retinal vascular disease attributable to FSHD**	**Have you been diagnosed with retinal problems or abnormal blood vessels at the back of your eye that your doctors think may be related to your FSHD? ("Coat's disease," retinal vascular disease)**
	Yes (start date **year**)	Yes, first occurred in [YEAR], but with no visual impairment
		Yes, first occurred in [YEAR], and has caused visual impairment
	No	No
	Unknown	I don't know

	Hearing loss	**Do you have hearing loss?**

9	Yes (start date **year**)	Yes, first occurred in [YEAR], but I don't use a hearing aid
		Yes, first occurred in [YEAR], and I use a hearing aid
	No	No
	Unknown	I don't know
10	**Scapular fixation**	**Have you had scapular fixation (an operation to fix your shoulder blade to your ribcage)?**
	Yes, bilateral (surgery dates **year**)	Yes, both shoulders operated in [**YEAR**] and [**YEAR**]
	Yes, unilateral (surgery date **year**)	Yes, one shoulder **[LEFT/RIGHT]** operated in [**YEAR**]
	No	No
	Unknown	I don't know
11	**Pregnancy (female at birth only)**	**(For females at birth only:) Have you ever been pregnant?**
	Yes, Number of pregnancies ____	Yes, ____ time(s) in [YEARS]
	No	No
	Unknown	I don't know
12	**Family history**	**Has anybody else in your family been diagnosed with FSHD (tick all that apply)?**
	Affected mother	Yes, mother
	Affected father	Yes, father
	Affected offspring	Yes, one or more children
	Affected sibling(s)	Yes, one or several of my siblings (brothers and sisters)
	Other affected relative	Yes, further relatives (other than parents and siblings)
	No	No
	Unknown	I don't know
13	**Epilepsy**	**Do you have a history of seizures/convulsions?**
	No	No
	Yes	Yes
	Unknown	I don't know
14	**Cognitive impairment**	**Do you have a history of delayed cognitive development or cognitive impairment?**
	No	No
	Yes	Yes
	Unknown	I don't know

HIGHLY ENCOURAGED ITEMS

15	**Ethnic origin**	**How would you describe your ethnic origin?**
	Caucasian	White - European origin (Caucasian)
	Black African/African American	Black African/African American
	Asian	Asian
	Mixed	Mixed
	Other	Other
	Declined	I choose not to answer this question
16	**Other registry**	**Have you signed up for any other NMD registry?**
	Yes (specify...)	Yes (if yes, please specify:)
	No	No
	Unknown	I don't know