Core dataset for international DM1 registry

	Item	Self-report example
	Mandatory items	
1.	**Personal data** Sex First name Last name Date of birth Address Zip/post code Telephone Email	**Your personal data**: Sex: First name: Last name: Date of birth: Address: Zip/post code: Telephone: Email:
2.	**Clinical Diagnosis** o Congenital Myotonic Dystrophy o DM1 o DM1 asymptomatic mutation carrier o Other o Unknown	**What is your diagnosis, according to your doctor?** o Congenital Myotonic Dystrophy o Myotonic Dystrophy Type 1 (DM1) o Mutation carrier for DM1 without symptoms o Other o I don't know
3.	**Genetic test result** o DM1 mutation (triplet repeat expansion) o Other mutation:…….. o Result pending o Not tested	**What is your genetic test result?** o DM1 mutation (triplet repeat expansion) o Other mutation:…….. o I have been tested but I haven't received the result yet o I have not been tested
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	
	MUSCLE	
6.	**Myotonia** ○ Severe ○ Mild ○ None	**Does myotonia (cramping, difficulties releasing your grip, etc.) currently have a negative effect on your normal daily activities?** ○ Yes, severely ○ Yes, but only mildly ○ Not at all
7.	**Myotonia medication use** ○ Yes (specify...) ○ No ○ Unknown	**Do you currently take medication to treat or prevent myotonia?** ○ Yes (specify or choose from drop down list) ○ No ○ I don't know
	CARDIAC	
8.	**Heart condition** ○ Yes, not further specified (age...) ○ Arrhythmia or conduction block (age...) ○ Cardiomyopathy (age...) ○ No ○ Unknown	**Have you been diagnosed with a heart condition?** ○ Yes, not further specified (at age: ….) ○ Yes, with arrhythmia or conduction block (at age: ….) ○ Yes, with cardiomyopathy (at age: ….) ○ No ○ I don't know
9.	**Cardiac implant** ○ Yes, not further specified (age …) ○ Pacemaker (age…) ○ ICD (age….) ○ No ○ Unknown	**Have you had an operation to implant a device to control/normalize your heart rhythm?** ○ Yes, not further specified (at age: ….) ○ Yes, a pacemaker (at age: ….) ○ Yes, a combined cardioverter-defibrillator (ICD) (at age: ….) ○ No ○ I don't know
10.	**10. ECG** ECG done: yes/no/unknown Sinus rhythm: yes/no PR interval: ….. ms QRS duration: ….. ms Date	**Have you had an electrocardiogram (ECG)?** ○ Yes ○ No ○ I don't know **If yes, please fill in the ECG results:** Sinus rhythm: yes/no PR interval: ….. ms QRS duration: ….. ms Date of examination: ……
11.	**Echocardiogram** Echo done: yes/no/unknown LVEF: …% Date	**Have you had an ultrasound of the heart (echocardiography)?** ○ Yes ○ No ○ I don't know **If yes, please fill in the echocardiography results:** LVEF ……% Date of examination: ……

12.	**Cardiac medication use** o Yes (specify...) o No o Unknown	**Do you currently take any medication to treat or protect your heart (e.g. ACE-inhibitors, beta-blockers, or anti-arrhythmics)?** o Yes (specify or choose from drop down list) o No o I don't know
	PULMONARY	
13.	**Non-invasive ventilation** o Full-time o Part-time o None	**Do you regularly use a non-invasive ventilation device?** o Yes, all day o Yes, but only part-time (e.g. at night) o No, never
14.	**Invasive ventilation** o Full-time o Part-time o None	**Do you use invasive ventilation?** o Yes, all day o Yes, part-time o No
15.	**Pulmonary function testing** FVC done: yes/no/unknown FVC: …% Date	**Have you had pulmonary function testing?** o Yes, o No o I don't know **If yes, please fill in the results of the test:** FVC .....% (predicted value) Date of the test:…….
	DIGESTIVE	
16.	**Dysphagia** o Yes o No o Unknown	**Do you have difficulty swallowing (food gets stuck in your throat, choking, etc)?** o Yes o No o I don't know
17.	**Gastric/nasogastric tube** o Yes o No o Unknown	**Do you have a tube (gastric/nasal) for feeding?** o Yes o No o I don't know
	OTHER	
18.	**Cataract surgery** o Yes (age …) o No o Unknown	**Have you had eye surgery for cataract removal**? o Yes (at age: ….) o No o I don't know
19.	**Fatigue/sleepiness** o Severe o Mild o No	**Does fatigue or daytime sleepiness currently have a negative effect on your normal daily activities?** o Yes, severely o Yes, but only mildly o Not at all

20.	**Fatigue medication use** ○ Yes ○ No ○ Unknown	**Do you currently take any medication to treat or prevent fatigue or daytime sleepiness?** ○ Yes (specify or choose from drop down list) ○ No ○ I don't know
21.	**Age of onset** ○ Congenital ○ Age ….. ○ Asymptomatic ○ Unknown	**At what age did the first medical problems occur that may be related to your myotonic dystrophy?** ○ At birth or within the first 4 weeks of life ○ At age ….. ○ I have no symptoms of myotonic dystrophy ○ I don't know
22.	**Genetic details/repeat size** ○ date of test ○ name of laboratory ○ method of testing ○ repeat size ○ No ○ Unknown	**Are details of your genetic test available?** ○ Yes ▪ date of test…… ▪ name of laboratory ▪ method of testing (Southern, PCR, RP-PCR) ▪ repeat size: ….. bp ○ No ○ I don't know
23.	**Positive family history** ○ Yes ○ No ○ Unknown	**Has anybody else in your family been diagnosed with the same disease?** ○ Yes ○ No ○ I don't know
24.	**Ethnic origin** ○ Caucasian ○ Black African/African American ○ Asian ○ Mixed ○ Other ○ Declined	**How would you describe your ethnic origin?** ○ White - European origin (Caucasian) ○ Black African/African American ○ Asian ○ Mixed ○ Other ○ I choose not to answer this question
25.	**Other registry** ○ Yes (specify...) ○ No ○ Unknown	**Have you signed up for any other myotonic dystrophy registry?** ○ Yes (if yes, please specify: ……) ○ No ○ I don't know