

Core dataset for international DM1 registry – final draft 1 August 2009

	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 <input type="radio"/> Congenital Myotonic Dystrophy <input type="radio"/> DM1 <input type="radio"/> DM1 asymptomatic mutation carrier <input type="radio"/> Other <input type="radio"/> Unknown	What is your diagnosis, according to your doctor? <input type="radio"/> Congenital Myotonic Dystrophy <input type="radio"/> Myotonic Dystrophy Type 1 (DM1) <input type="radio"/> Mutation carrier for DM1 without symptoms <input type="radio"/> Other <input type="radio"/> I don't know
3.	Genetic test result <input type="radio"/> DM1 mutation (triplet repeat expansion) <input type="radio"/> Other mutation:..... <input type="radio"/> Result pending <input type="radio"/> Not tested	What is your genetic test result? <input type="radio"/> DM1 mutation (triplet repeat expansion) <input type="radio"/> Other mutation:..... <input type="radio"/> I have been tested but I haven't received the result yet <input type="radio"/> I have not been tested
4.	Current best motor function <input type="radio"/> Ambulatory (unassisted) <input type="radio"/> Ambulatory (assisted) <input type="radio"/> Non-ambulatory	Which of the following options describes the best motor function you are currently able to achieve? (please tick the most appropriate answer) <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
5.	Wheelchair use <input type="radio"/> No <input type="radio"/> Part-time (age...) <input type="radio"/> Full-time (age ...)	Do you use a wheelchair? (please tick the most appropriate answer) <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		
MUSCLE		
6.	Myotonia <ul style="list-style-type: none"> <input type="radio"/> Severe <input type="radio"/> Mild <input type="radio"/> None 	Does myotonia (cramping, difficulties releasing your grip, etc.) currently have a negative effect on your normal daily activities? <ul style="list-style-type: none"> <input type="radio"/> Yes, severely <input type="radio"/> Yes, but only mildly <input type="radio"/> Not at all
7.	Myotonia medication use <ul style="list-style-type: none"> <input type="radio"/> Yes (specify...) <input type="radio"/> No <input type="radio"/> Unknown 	Do you currently take medication to treat or prevent myotonia? <ul style="list-style-type: none"> <input type="radio"/> Yes (specify or choose from drop down list) <input type="radio"/> No <input type="radio"/> I don't know
CARDIAC		
8.	Heart condition <ul style="list-style-type: none"> <input type="radio"/> Yes, not further specified (age...) <input type="radio"/> Arrhythmia or conduction block (age...) <input type="radio"/> Cardiomyopathy (age...) <input type="radio"/> No <input type="radio"/> Unknown 	Have you been diagnosed with a heart condition? <ul style="list-style-type: none"> <input type="radio"/> Yes, not further specified (at age:) <input type="radio"/> Yes, with arrhythmia or conduction block (at age:) <input type="radio"/> Yes, with cardiomyopathy (at age:) <input type="radio"/> No <input type="radio"/> I don't know
9.	Cardiac implant <ul style="list-style-type: none"> <input type="radio"/> Yes, not further specified (age ...) <input type="radio"/> Pacemaker (age...) <input type="radio"/> ICD (age....) <input type="radio"/> No <input type="radio"/> Unknown 	Have you had an operation to implant a device to control/normalize your heart rhythm? <ul style="list-style-type: none"> <input type="radio"/> Yes, not further specified (at age:) <input type="radio"/> Yes, a pacemaker (at age:) <input type="radio"/> Yes, a combined cardioverter-defibrillator (ICD) (at age:) <input type="radio"/> No <input type="radio"/> 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)? <ul style="list-style-type: none"> <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> 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)? <ul style="list-style-type: none"> <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> I don't know If yes, please fill in the echocardiography results: LVEF% Date of examination:

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

20.	Fatigue medication use <ul style="list-style-type: none"> <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown 	Do you currently take any medication to treat or prevent fatigue or daytime sleepiness? <ul style="list-style-type: none"> <input type="radio"/> Yes (specify or choose from drop down list) <input type="radio"/> No <input type="radio"/> I don't know
21.	Age of onset <ul style="list-style-type: none"> <input type="radio"/> Congenital <input type="radio"/> Age <input type="radio"/> Asymptomatic <input type="radio"/> Unknown 	At what age did the first medical problems occur that may be related to your myotonic dystrophy? <ul style="list-style-type: none"> <input type="radio"/> At birth or within the first 4 weeks of life <input type="radio"/> At age <input type="radio"/> I have no symptoms of myotonic dystrophy <input type="radio"/> I don't know
22.	Genetic details/repeat size <ul style="list-style-type: none"> <input type="radio"/> date of test <input type="radio"/> name of laboratory <input type="radio"/> method of testing <input type="radio"/> repeat size <input type="radio"/> No <input type="radio"/> Unknown 	Are details of your genetic test available? <ul style="list-style-type: none"> <input type="radio"/> Yes <ul style="list-style-type: none"> <input type="checkbox"/> date of test..... <input type="checkbox"/> name of laboratory <input type="checkbox"/> method of testing (Southern, PCR, RP-PCR) <input type="checkbox"/> repeat size: bp <input type="radio"/> No <input type="radio"/> I don't know
23.	Positive family history <ul style="list-style-type: none"> <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown 	Has anybody else in your family been diagnosed with the same disease? <ul style="list-style-type: none"> <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> I don't know
24.	Ethnic origin <ul style="list-style-type: none"> <input type="radio"/> Caucasian <input type="radio"/> Black African/African American <input type="radio"/> Asian <input type="radio"/> Mixed <input type="radio"/> Other <input type="radio"/> Declined 	How would you describe your ethnic origin? <ul style="list-style-type: none"> <input type="radio"/> White - European origin (Caucasian) <input type="radio"/> Black African/African American <input type="radio"/> Asian <input type="radio"/> Mixed <input type="radio"/> Other <input type="radio"/> I choose not to answer this question
25.	Other registry <ul style="list-style-type: none"> <input type="radio"/> Yes (specify...) <input type="radio"/> No <input type="radio"/> Unknown 	Have you signed up for any other myotonic dystrophy registry? <ul style="list-style-type: none"> <input type="radio"/> Yes (if yes, please specify:) <input type="radio"/> No <input type="radio"/> I don't know