Core data set for DMD national 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.	**Genetic Test Result:** Mutation name in DMD gene following HGVS rules (based on cDNA Ref Seq)	**What is your genetic test result?** Mutation name in DMD gene following HGVS rules (based on cDNA Ref Seq): c._____
3.	**Deletion: all exons tested** o Yes o No o Unknown	**Deletion of exon(s): Have all exons been tested?** o Yes o No o Unknown
4.	**Duplication: all exons tested** o Yes o No o Unknown	**Duplication of exon(s): Have all exons been** tested? o Yes o No o Unknown
5.	**Deletion/Duplication: boundaries known.** o Yes o No o Unknown	**If the result of the genetic testing shows a deletion or duplication: Are the boundaries known?** o Yes o No o Unknown
6.	**Point Mutation: all exons sequenced** o Yes o No o Unknown	**If the result of the genetic testing shows a point mutation: Have all exons been sequenced?** o Yes o No o Unknown
7.	**Targeted mutation testing in the patient but testing of all exons in a relative male patient** o Yes o No o Unknown	**If targeted mutation testing has been performed with the patient's DNA, have all exons been tested in a male relative who is also a DMD patient?** o Yes o No o Unknown
8.	**Clinical Diagnosis** o Duchenne Muscular Dystrophy (DMD) o Becker Muscular Dystrophy (BMD) o Intermediate Muscular Dystrophy (IMD) o Female Carrier o Unknown	**What is your diagnosis according to your doctor?** o Duchenne Muscular Dystrophy (DMD) o Becker Muscular Dystrophy (BMD) o Intermediate Muscular Dystrophy (IMD) o Female Carrier o Unknown
9.	**Current best Motor Function**	**Are you currently able to walk**

	o Can currently Walk o Cannot currently Walk	o Yes (without any help/support) o No
10.	**Wheelchair use (if over 3 years of age)** 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: ……)
11.	**Current Steroid Therapy** o Yes, Currently o No, but previously o Never o Unknown	**Are you currently taking steroids (glucocorticoids) for DMD?** o Yes, Currently o No, but previously o Never o Unknown
12.	**Scoliosis Surgery** o Yes o No o Unknown	**Have you had Scoliosis Surgery?** o Yes o No o Unknown
13.	**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
14.	**Currently included in a clinical trial** Yes, Currently (name of drug…..) No, but preciously Never Unknown	**Are you currently taking part in a clinical trial?** o Yes, Currently o No, but previously o Never o Unknown **If yes please specify the name the drug being tested**……..
	Highly Encouraged Items	
15.	**Currently able to sit without support** o Yes o no	Are you currently able to sit without support? o Yes o No
	Cardiac	
16.	**Heart condition** o Yes, not further specified (age...) o Arrhythmia or conduction block (age...) o Cardiomyopathy (age...) o No o Unknown	**Have you been diagnosed with a heart condition?** o Yes, not further specified (at age: ….) o Yes, with arrhythmia or conduction block (at age: ….) o Yes, with cardiomyopathy (at age: ….) o No o I don't know
17.	**Echocardiogram** Echo done: yes/no/unknown	**Have you had an ultrasound of the heart (echocardiography)?**

	LVEF: …% Date	o Yes o No o I don't know **If yes, please fill in the echocardiography results:** LVEF ……% Date of examination: ……
	Pulmonary	
18.	**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
19.	**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
20.	**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:……
21.	**Previous muscle biopsy** o Yes o No o Unknown	**Have you ever had a muscle biopsy?** o Yes o No o Unknown
22.	**Other registry** o Yes (specify...) o No o Unknown	**Have you signed up for any other DMD registry?** o Yes (if yes, please specify: ……) o No o I don't know
23.	**Positive family history** o Yes o No o Unknown	**Has anybody else in your family been diagnosed with the same disease?** o Yes o No o I don't know **If yes please specify the relation to you……..**