

## Instructions for the Self-Report Form – DMD Patient Registry

This document contains additional information to help you fill in your registration form correctly. The numbers in this document (1-17) refer to questions 1-17 in the registration form. Before you start, you should be aware of the following:

- Before the registration process can be completed, you will have to read and sign a patient information and consent form as well as the registration form.
- If you have any questions or comments about this form or the registration process, please contact Professor Hanns Lochmüller at tel. 0191 241 8602 or email [hanns.lochmuller@ncl.ac.uk](mailto:hanns.lochmuller@ncl.ac.uk).
- If a question contains tick boxes, please only tick **one** box with the most appropriate answer.
- It is very important that your genetic test result is entered correctly in the registry. If you do not know your exact genetic test result or are not sure how to fill it in correctly (question 3), you can provide us with the name of the hospital or medical centre where you had your test. We will contact them to ask for the result on your behalf.

### PART I – MANDATORY ITEMS

The questions in this first section have to be answered completely. Data from this section will be kept in the national registry and a copy will also be sent to the global registry. However, your personal information will only be kept in the national registry – in the global registry your data will be identified only by a code.

#### 1. Patient's personal details

Please fill in the patient's name, date of birth, NHS number (UK), postal and email address, and phone number.

#### 2. If you are the patient's representative (parent/guardian), please provide your details

If you are filling in the form on behalf of somebody else, please give your name, postal and email address and explain your relationship to the patient (for example: mother/father).

#### 3. What is the patient's genetic test result?

Please fill in the result of the genetic test or tell us if the result is pending (if you have been tested but haven't received the result yet). If you don't know the result or aren't sure what to write, please tell us where the patient had the test. If you agree, we will ask them to send us the results.

Mutation name in *DMD* gene following HGVS rules (based on cDNA Ref Seq): This information can be found in the genetic report; for more information on the genetic nomenclature, please see the "Handbook on DMD genetics".

Deletion of exon(s): Have all exons been tested?

It is highly recommended to test all exons for deletions due to the occurrence of non-contiguous rearrangements.

Duplication of exon(s): Have all exons been tested?

It is highly recommended to test all exons for duplications due to the occurrence of non-contiguous rearrangements.

If the result of the genetic testing shows a deletion or duplication: Are the boundaries known?

It is recommended, that patients for whom the extents of the deletion have not been unambiguously defined should be further analyzed with an additional set of exons (multiplex PCR, semi-quantitative PCR) or MLPA. Many patients have been analyzed by using the standard multiplex PCR method, which is intended to detect deletions but not necessarily to define the borders of deletions. If only the Southern blot technique has been performed it is recommended to re-analyze the DNA using a PCR-based technique. For more information, see the Handbook on DMD genetics.

If the result of the genetic testing shows a point mutation: Have all exons been sequenced?

It is highly recommended to sequence all exons for point mutations due to the possible occurrence of multiple mutations.

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?

This item should be filled in ONLY when the "No" answer has been selected for one of the three boxes above (deletion, duplication, point mutation).

#### 4. What is the patient's diagnosis, according to your doctor?

When your doctor told you the diagnosis, did he/she name it "DMD" or something else?

#### 5. Current best motor function (please tick the most appropriate answer):

Motor function describes a person's ability to move their body.

What is the best motor function the patient is able to manage at the moment?

Can the patient walk (with or without help/support)?

6. If the patient is 3 years old or more, does he/she have to use a wheelchair?

You only have to answer this question if the patient is 3 years old or more.

We want to know if the patient has to use a wheelchair. If the patient does use a wheelchair, does he/she use it any time he/she needs to get around (=full-time), or can he/she walk short distances without the wheelchair and therefore uses it only for longer distances (this would be part-time)?

7. Is the patient taking steroids (glucocorticoids) for Muscular Dystrophy?

Clinical trials have shown that treatment with steroids (a different name for this group of medication is glucocorticoids) has a positive impact on muscle strength in patients with Duchenne muscular dystrophy.

Please tick if the patient is currently taking steroids, if he/she has taken steroids previously, or if he/she has never taken steroids for Muscular Dystrophy.

8. Has the patient had spinal surgery for scoliosis?



Some DMD patients suffer from weakness in their back muscles which results in a deformation or "bending" of their spine called scoliosis. In order to stabilize the spine, they often have surgery done. If the patient has had this kind of operation on their spine, please tick "yes". If the patient has not had this operation, even if you think they might be going to have it in future, please tick "no". You can update the information when we next send you an update form.

9. Is the patient currently taking any medication to treat or protect his/her heart?

Because the heart muscle is also affected by muscular dystrophy (see Q12), it is very reasonable that patients with Duchenne Muscular Dystrophy take medication to protect their heart at an early stage of the disease. These types of medication include Beta-blockers or ACE-inhibitors. If you know his/her medication, please tick/list them here.

10. Is the patient currently included in a clinical trial?

Please indicate if the patient is currently taking part in any of the clinical trials listed here. This information is of high relevance in the planning phase of a new trial!

## PART II – HIGHLY ENCOURAGED ITEMS

Please answer the following set of questions as far as you are able. Anonymous data from this section will also be sent to the global registry and will be useful for researchers studying particular aspects of DMD. But don't worry if you aren't able to answer every question. We can still include your information in the global registry even if this part of the questionnaire is not filled in completely.

### 11. Is the patient currently able to sit without support?

This question is also related to the motor function of the patient. Sitting without support means that the patient must be able to maintain the sitting position on a chair or a wheelchair without support of upper limbs or leaning against the back of the chair.

### 12. Has cardiomyopathy (affection of the heart muscle) been diagnosed in the patient?

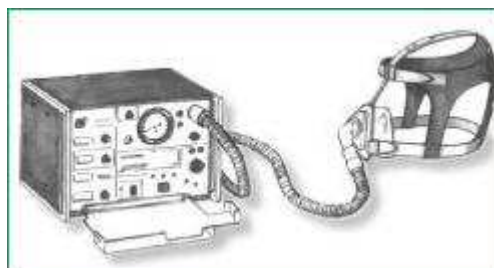
In patients with Muscular Dystrophy not only the skeletal muscle is affected, but also the heart muscle. In the course of the disease, patients are therefore often diagnosed with a weakness of the heart muscle called cardiomyopathy.

To monitor your heart function, your doctor might have done an ultrasound of your heart. One of the parameters measured in this test is known as the Left Ventricular Ejection Fraction (LVEF). LVEF is the percentage of blood volume ejected by the left ventricle of the heart per heart beat. In a healthy person, LVEF is usually higher than 65%.

If the patient has had an ultrasound of the heart and you know the result, please fill this in. Please also include the date of the test.

### 13. Does the patient regularly use a non-invasive ventilation device?

Some DMD patients have trouble with their breathing. To support their breathing, they get a ventilation device that they have to use either all day (=full-time) or only a few hours per day, e.g. at night (=part-time). "Non-invasive" means that they use this device without having had an operation (usually this means they wear a mask that can be removed at any time).



14. Does the patient use invasive ventilation?

See also the notes to Q13 above.

“Invasive ventilation” means that the patient had to have an operation (a tracheotomy or incision in the wind-pipe) to use the ventilation device. Again, this ventilatory support system can be used all day (=full-time) or a few hours per day (=part-time).



To monitor your breathing function, your doctor might have done pulmonary function tests. One of the parameters measured in these tests is known as the Forced Vital Capacity (FVC). To test the FVC, the patient has to breathe in as far as they can and then blow out into a machine that measures how much air is being exhaled. The FVC is the volume of air exhaled. The FVC varies with age, gender, weight and height and is measured in litres.

If the patient has had this test and you know the result, please fill this in. Please also include the date of the test.

15. Has the patient ever had a muscle biopsy?

In order to confirm the diagnosis “Muscular Dystrophy”, many patients undergo a muscle biopsy. Several tests can be performed on this small piece of muscle which is removed surgically.

16. Has the patient signed up for any other DMD registry?

Since we aim to harmonize international DMD registries and include data in one global registry, we want to make sure that patients do not appear in our global registry twice by mistake. If you are registered already in another DMD registry, please let us know, and we’ll make sure we only register you once in the global registry. If you are already registered in another registry, that doesn’t mean we don’t want you to register in this one – please do register, but let us know about the other registry.

17. Does anybody else in the patient’s family have the same kind of disease?

Since DMD is an inherited disease, we want to know if there are other relatives who have similar symptoms or the same diagnosis.