UK Myotonic Dystrophy Patient Registry User Guide

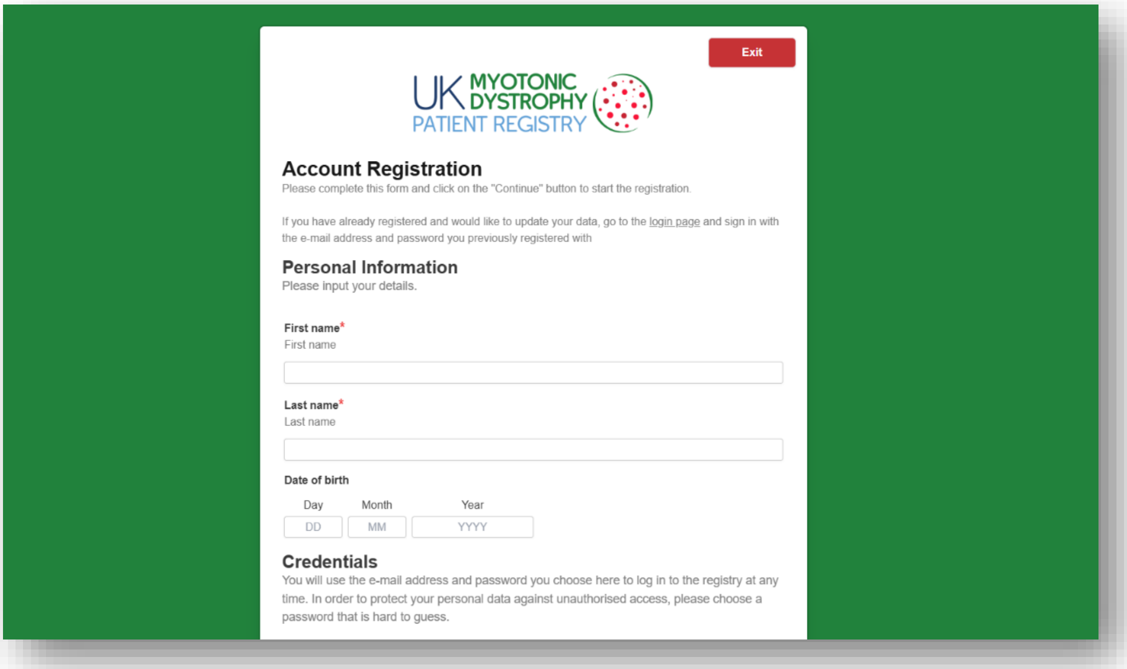
How to register and share updates with the UK Myotonic Dystrophy Patient Registry **as a patient, or as the parent/carer of a patient**.

A separate user guide exists for clinical users – contact the registry team for details.

Contents

Step 1 – Set up your user account

- Go to the registry website - https://www.dm-registry.org.uk/
- Click 'Register as a patient or parent'
- You will see the registration page that looks like the image below. Add your information, then click the blue **'Continue'** button.
- Please ensure you keep your email address and password somewhere safe – you will need to remember the details you have used in order to log in to the registry to provide updates in future.



Account Registration

Please complete this form and click on the "Continue" button to start the registration.

If you have already registered and would like to update your data, go to the login page and sign in with the e-mail address and password you previously registered with

Personal Information

Please input your details.

First name*
First name

Last name*
Last name

Date of birth
Day DD
Month MM
Year YYYY

Credentials

You will use the e-mail address and password you choose here to log in to the registry at any time. In order to protect your personal data against unauthorised access, please choose a password that is hard to guess.

Step 2 – Add a patient

- Tell us if you are registering yourself or your child as a patient on this page.
- **If you are registering both yourself and a child, please register yourself first, then create a patient profile for your child afterwards.**

Welcome to the UK Myotonic Dystrophy Patient Registry

Please Note:
This registry is for Myotonic Dystrophy patients living in the UK only. You can register yourself as an adult patient (aged 16 or over), and/or your affected child(ren) aged under 16. If you do not fit these criteria, please contact the registry team before continuing.

If you are an **adult patient** registering for the first time and you also want to register your affected child(ren), please **register yourself as a patient first.** You can add your child(ren)'s details later.

If you are under the age of 16, your parent or legal guardian must complete this registration process and provide consent for you to register. You can take control of your record when you turn 16.

If you are an adult patient (aged 16+) and require assistance managing your account, you can invite a helper to assist you after you have completed registration.

If you are not a patient or parent/guardian yourself, but would like to support an adult patient with registration, please contact the registry team.

Please contact the registry team if you have any questions or need help with the registration process.

Step 3 – Complete the patient consent form

- Please read through the consent form on this page. If you are registering a child, please ensure they understand all the information provided
- If you or the patient have any questions about anything in the consent form, please contact the registry team before continuing.
- If you need to pause registration at this stage, you can pick up the process from this page and won't need to start from the beginning.
- Take your time to read through the whole page…

Principal Investigator/Data Controller: Professor Chiara Marini Bettolo, Translational and Clinical Research Institute, Newcastle University

Information for adult patients (age 16 and over) (v2.1 March 2024)

You can print or save this page for your records and you can access it again at any time through the website.

Copy for your records (PDF file, opens in new window)

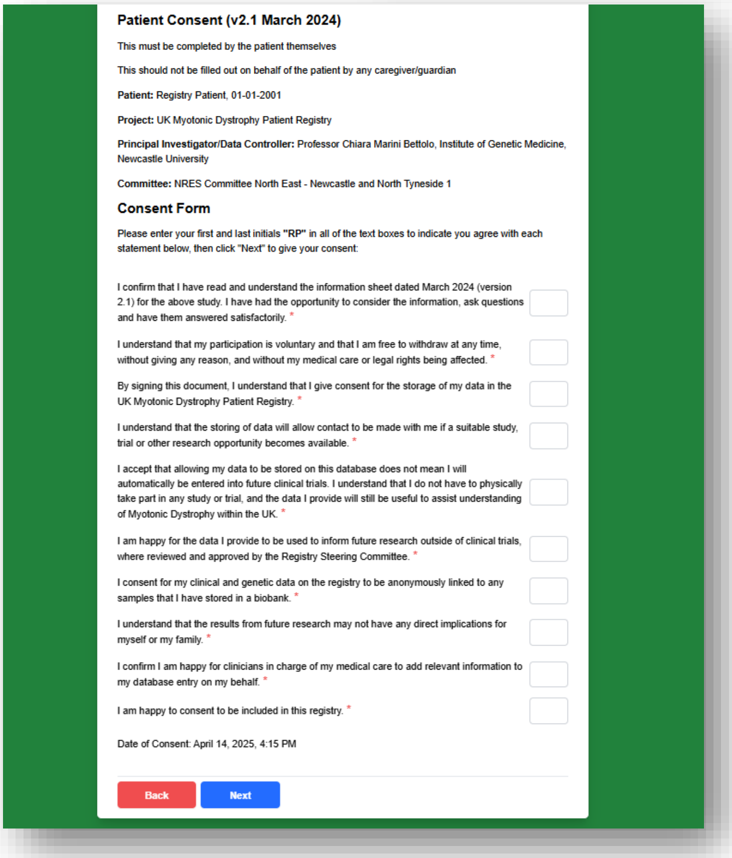
You are being invited to take part in an ongoing research project known as a 'Patient Registry'. This would involve you answering some questions about how Myotonic Dystrophy (sometimes shortened to 'DM'), affects you and giving consent for the registry team to keep a record of your answers in a secure, password-protected computer database. You can join if you have Myotonic Dystrophy, even if you don't have any symptoms. You can also nominate your medical team to add some extra information from your medical notes on your behalf.

Before you accept or decline the invitation, it is important for you to understand why the research is being done and what it will involve. Please read the following information and discuss it with relatives, friends and your GP, if you wish. If there is anything that is not clear, or if you have any further questions, please ask us (our contact details are at the end of this document, or on the website here).

Take time to decide whether you would like to take part, or not.

Why is this research being done?

It is important to collect information from people with rare conditions like Myotonic Dystrophy to help researchers and scientists trying to better understand the condition. More research is likely to improve the care patients receive in future. Specific examples of how the registry data has been used to further research and support the Myotonic Dystrophy community in the UK can be viewed on the registry website.



Patient Consent (v2.1 March 2024)

This must be completed by the patient themselves

This should not be filled out on behalf of the patient by any caregiver/guardian

Patient: Registry Patient, 01-01-2001

Project: UK Myotonic Dystrophy Patient Registry

Principal Investigator/Data Controller: Professor Chiara Marini Bettolo, Institute of Genetic Medicine, Newcastle University

Committee: NRES Committee North East - Newcastle and North Tyneside 1

Consent Form

Please enter your first and last initials **"RP"** in all of the text boxes to indicate you agree with each statement below, then click "Next" to give your consent:

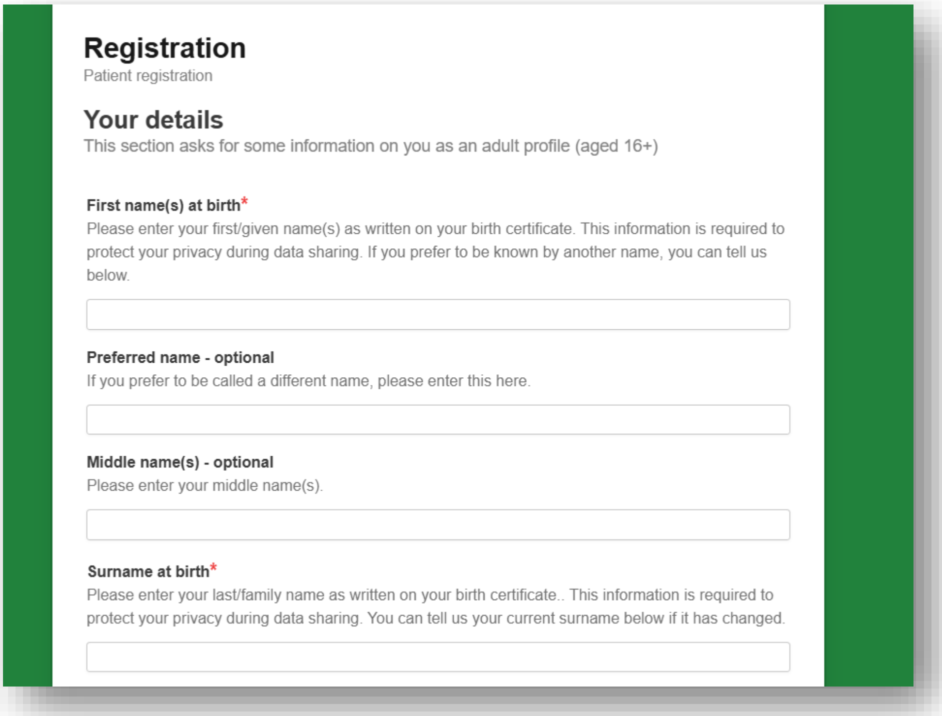
I confirm that I have read and understand the information sheet dated March 2024 (version 2.1) for the above study. I have had the opportunity to consider the information, ask questions and have them answered satisfactorily. *	☐
I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, and without my medical care or legal rights being affected. *	☐
By signing this document, I understand that I give consent for the storage of my data in the UK Myotonic Dystrophy Patient Registry. *	☐
I understand that the storing of data will allow contact to be made with me if a suitable study, trial or other research opportunity becomes available. *	☐
I accept that allowing my data to be stored on this database does not mean I will automatically be entered into future clinical trials. I understand that I do not have to physically take part in any study or trial, and the data I provide will still be useful to assist understanding of Myotonic Dystrophy within the UK. *	☐
I am happy for the data I provide to be used to inform future research outside of clinical trials, where reviewed and approved by the Registry Steering Committee. *	☐
I consent for my clinical and genetic data on the registry to be anonymously linked to any samples that I have stored in a biobank. *	☐
I understand that the results from future research may not have any direct implications for myself or my family. *	☐
I confirm I am happy for clinicians in charge of my medical care to add relevant information to my database entry on my behalf. *	☐
I am happy to consent to be included in this registry. *	☐

Date of Consent: April 14, 2025, 4:15 PM

- When you reach the bottom of the page, please enter your initials in each text box to indicate you agree with each statement.
- If you do not enter your initials in each box, you will be unable to continue with your registration.
- Press the blue **'Next'** button to proceed.

Step 5 – Add your/your child's personal details

- The next step is to complete some personal details.
- Questions that contain an asterisk at the end are mandatory and must be answered to proceed:



Registration

Patient registration

Your details

This section asks for some information on you as an adult profile (aged 16+)

First name(s) at birth*

Please enter your first/given name(s) as written on your birth certificate. This information is required to protect your privacy during data sharing. If you prefer to be known by another name, you can tell us below.

Preferred name - optional

If you prefer to be called a different name, please enter this here.

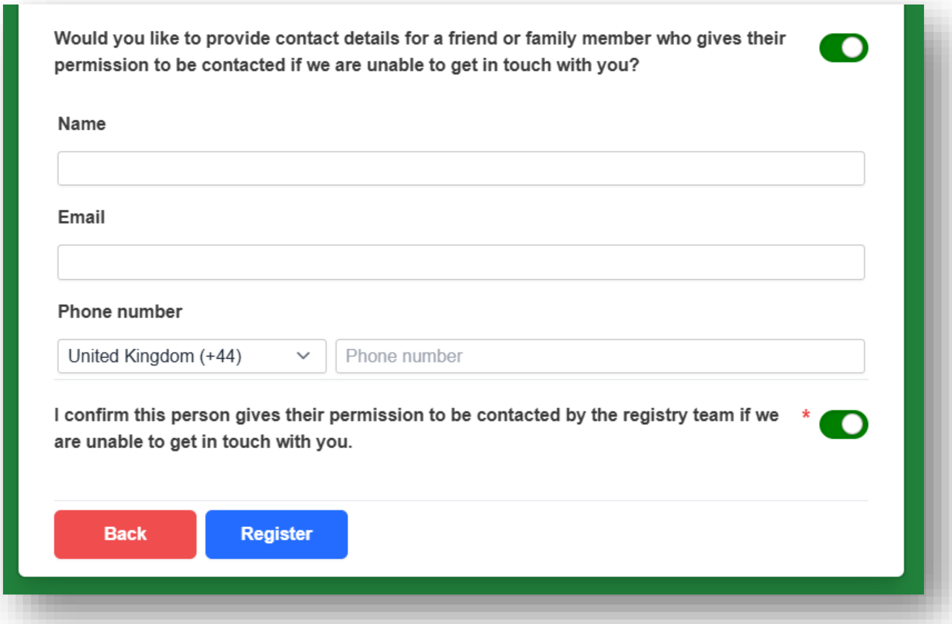
Middle name(s) - optional

Please enter your middle name(s).

Surname at birth*

Please enter your last/family name as written on your birth certificate.. This information is required to protect your privacy during data sharing. You can tell us your current surname below if it has changed.

- At the bottom of this page there is an option to share details of a friend or family member who can be contacted if we are unable to get in touch with you. If you wish to do this and have their permission, toggle the question on and enter their information.



Would you like to provide contact details for a friend or family member who gives their permission to be contacted if we are unable to get in touch with you?

Name

Email

Phone number

United Kingdom (+44) | Phone number

I confirm this person gives their permission to be contacted by the registry team if we are unable to get in touch with you. *

- Once the mandatory fields are complete, click the blue **'Register'** button.

Step 6 – Adding another patient

This step is only required if you will be providing information for at least one child patient aged 15 or younger (i.e., if you are a patient also registering their affected child, or an unaffected parent registering two children with the condition).

Adult patients should always complete their own initial registration. If they wish, they can then nominate someone to assist them with entering their patient questionnaire answers.

Register More Users

Would you like to register a child under your care? You can add more dependants to your account in the future if you are not currently ready to add another.

Click **'Yes'** if you wish to add another user and complete the consent and registration (steps 2-5) again.

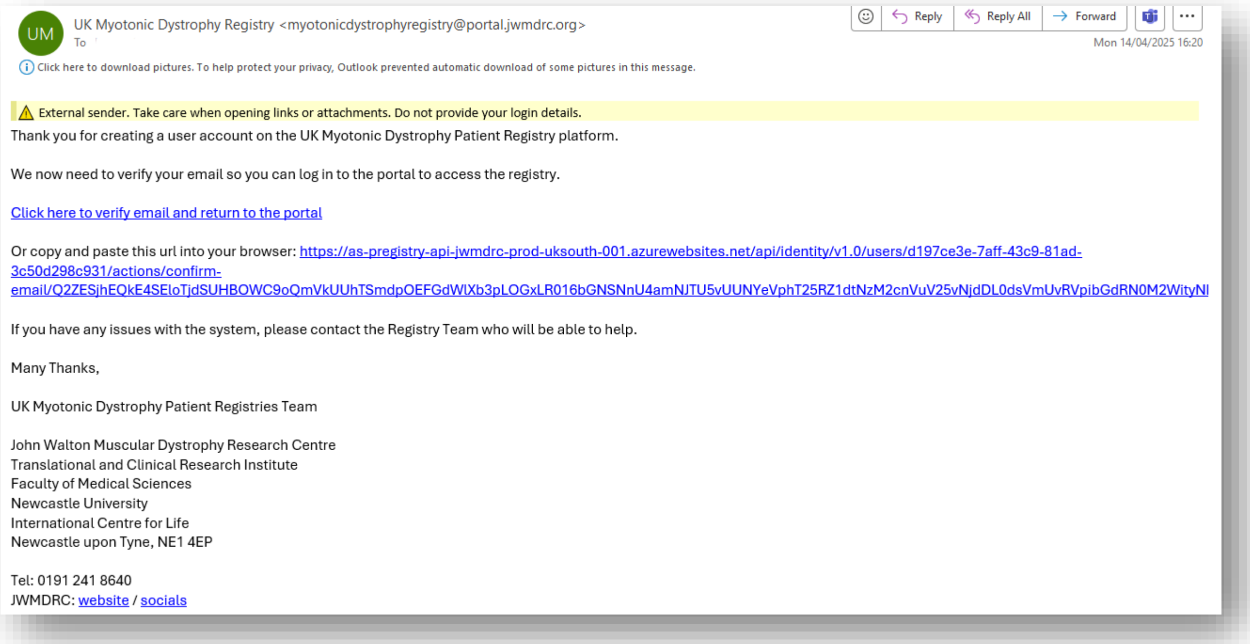
- You can add as many child patients for whom you are a parent or carer as you want by repeating this step. You will be responsible for keeping the information updated for any patients you have added.
- When a patient turns 16 years old, they must complete the consent process again as an adult. They will then create their user registration and take ownership of their patient record.

If you are only registering yourself, **click 'No'.**

Please contact the registry team (myotonicdystrophyregistry@newcastle.ac.uk) if you have any questions about registering or assisting another patient.

Step 7 – Confirm your email

- After pressing **'Continue'**, your user account will be set up and you will receive a confirmation email, but **your registration is not yet complete**.
- Please verify your email address by clicking the link in the email sent to your account (if you can't see it, first check your junk folder then contact the registry team). The message should look something like this:



UK Myotonic Dystrophy Registry <myotonicdystrophyregistry@portal.jwmdrc.org>

Mon 14/04/2025 16:20

Click here to download pictures. To help protect your privacy, Outlook prevented automatic download of some pictures in this message.

External sender. Take care when opening links or attachments. Do not provide your login details.

Thank you for creating a user account on the UK Myotonic Dystrophy Patient Registry platform.

We now need to verify your email so you can log in to the portal to access the registry.

Click here to verify email and return to the portal

Or copy and paste this url into your browser: https://as-pregistry-api-jwmdrc-prod-uksouth-001.azurewebsites.net/api/identity/v1.0/users/d197ce3e-7aff-43c9-81ad-3c50d298c931/actions/confirm-email/Q2ZESjhEQkE4SEloTjdSUHBOWC9oQmVkUUhTSmdpOEFGdWlXb3pLOGxLR016bGNSNnU4amNJTU5vUUNYeVphT25RZ1dtNzM2cnVuV25vNjdDL0dsVmUvRVpibGdRN0M2WityNl

If you have any issues with the system, please contact the Registry Team who will be able to help.

Many Thanks,

UK Myotonic Dystrophy Patient Registries Team

John Walton Muscular Dystrophy Research Centre
Translational and Clinical Research Institute
Faculty of Medical Sciences
Newcastle University
International Centre for Life
Newcastle upon Tyne, NE1 4EP

Tel: 0191 241 8640
JWMDRC: website / socials

- When you click to verify your email address, you will be taken directly to the patient dashboard:
- Your/your child's name will be displayed in the top right corner.

Welcome to the UK Myotonic Dystrophy (DM) Patient Registry

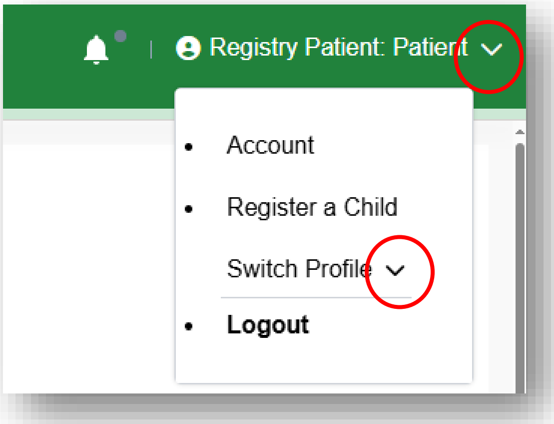
Please complete your Patient Questionnaire and Medical Questionnaire once a year to share ongoing updates about your health and wellbeing. You can find these in the Questionnaires section in the left menu.

You can access your account details by selecting your name at the top of this page. Here you can:

- Select or invite your doctor
- Update your contact details
- Add a helper
- View your consent forms
- Change your password

Contact the registry team if you have any questions or technical issues with any part of the registry.

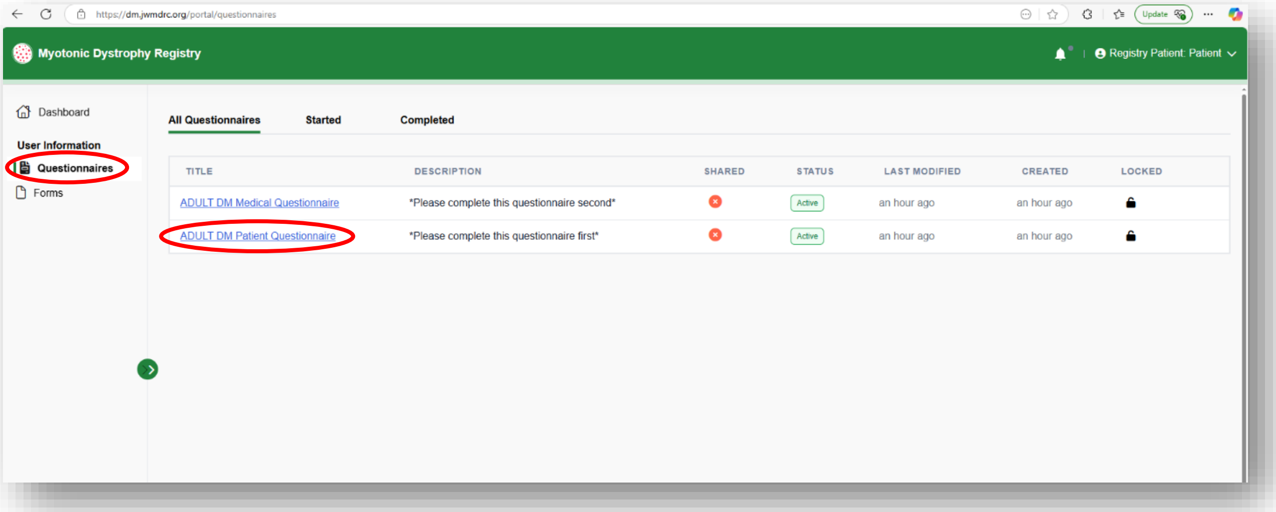
The JWMDRC Patient Registries Team are grateful for the donations received from Arrowhead Pharmaceuticals, AskBio, PepGen and Roche to support development of this registry platform. Industry do not receive any access to the system or data in return for this contribution, which is purely philanthropic in nature.



- If you have registered multiple patients from the same account, you will be able to switch between them by clicking on the down arrows.

Step 8 – Providing information about your condition/the patient’s condition

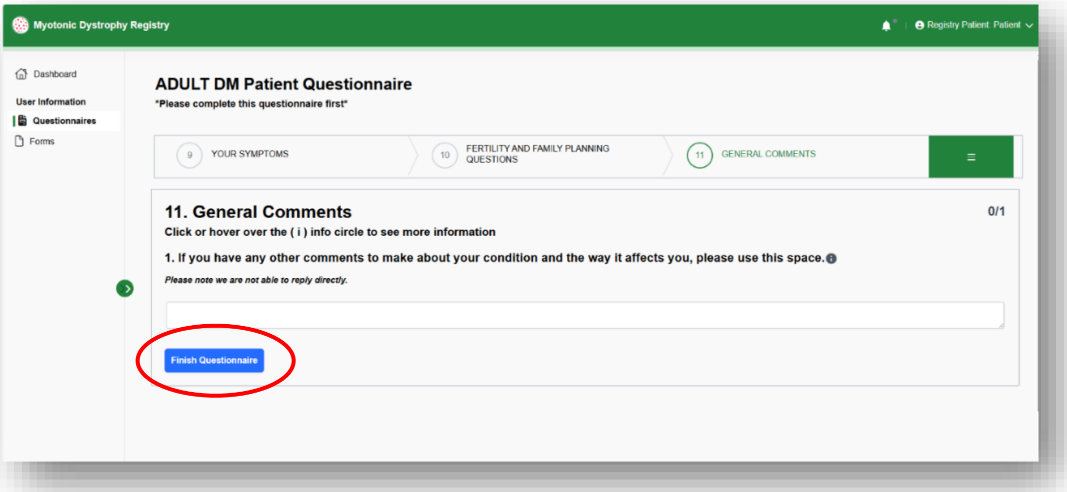
- At this point you have registered as a patient on the registry, but now we need to collect information about your condition/the patient’s condition.
- To access your questionnaires, click **‘Questionnaires’** then **‘Adult/Child DM Patient Questionnaire’.**



TITLE	DESCRIPTION	SHARED	STATUS	LAST MODIFIED	CREATED	LOCKED
ADULT DM Medical Questionnaire	*Please complete this questionnaire second*	✕	Active	an hour ago	an hour ago	🔓
ADULT DM Patient Questionnaire	*Please complete this questionnaire first*	✕	Active	an hour ago	an hour ago	🔓

- The questionnaires are split into sections asking about your condition, your pain levels, and your wellbeing/quality of life.
- Please complete each page as thoroughly and honestly as you can, then click the blue **‘Next section’** button at the end of the page to continue.

- If you need a break at any time, you can logout and come back to this section by logging back in and clicking on **'Questionnaires'** from the menu on the left of the screen



ADULT DM Patient Questionnaire

Please complete this questionnaire first

11. General Comments

Click or hover over the (i) info circle to see more information

1. If you have any other comments to make about your condition and the way it affects you, please use this space.

Please note we are not able to reply directly.

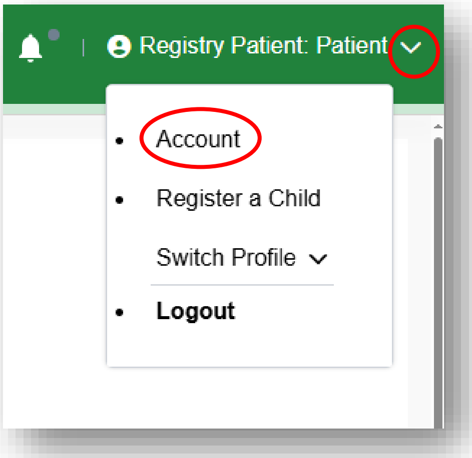
- **You must complete all of the mandatory (*) questions and click 'Finish Questionnaire' to submit your answers.**

- You will receive a reminder email in a year's time to ask you to log back on to the registry using the email address and password you entered in Step 1. You can log in to update your answers to these questions as often as you like, but we ask that this is done at least annually.

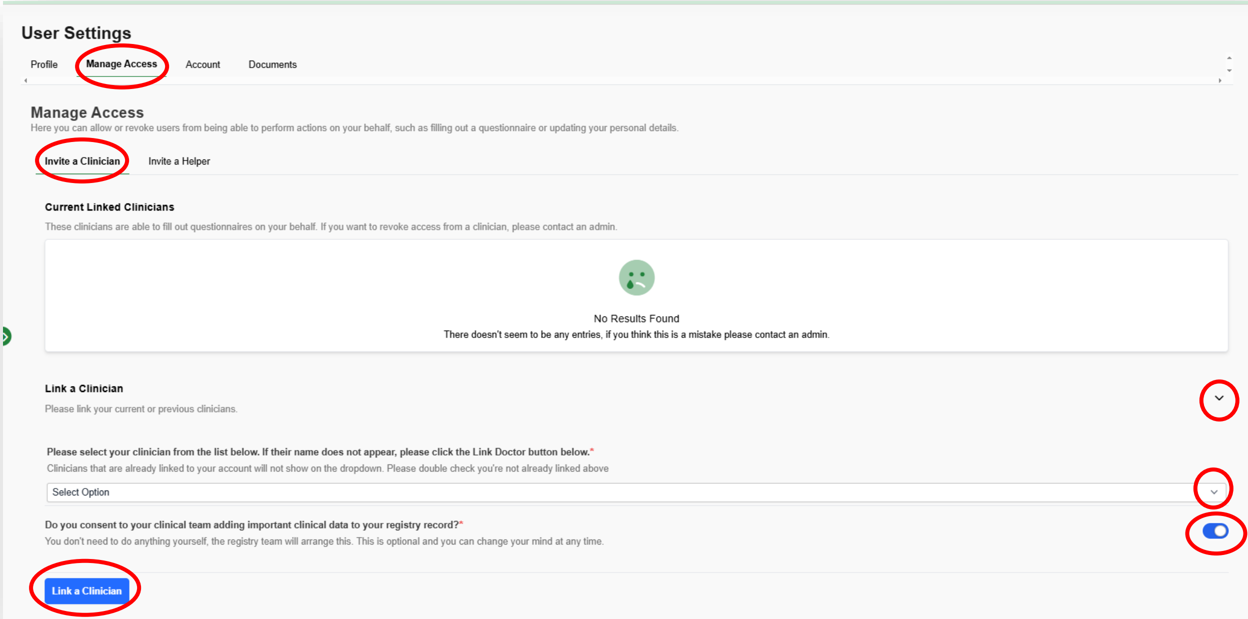
- You have now completed your registration! If you need to complete the questionnaire for another patient, switch patients (see p8) and repeat this step.

Step 9 – Adding your/your child's doctor

- To add a doctor, you must link their account to yours, even if they had previously been linked in the old registry system.
- You can manage which users have access to your patient data in your account settings.
- To link your doctor, click '**Manage Access'**, then '**Invite a Clinician'** and '**Link a Clinician**'
- When you click '**Link a Clinician**', a drop-down list of doctors will appear. If your doctor is on the list, please select them.



- You need to consent to them adding data to your patient profile by toggling on the consent button. Then click **'Link a Clinician'** and your doctor will be linked to your profile.



Manage Access

Here you can allow or revoke users from being able to perform actions on your behalf, such as filling out a questionnaire or updating your personal details.

Current Linked Clinicians

These clinicians are able to fill out questionnaires on your behalf. If you want to revoke access from a clinician, please contact an admin.

No Results Found

There doesn't seem to be any entries, if you think this is a mistake please contact an admin.

Link a Clinician

Please link your current or previous clinicians.

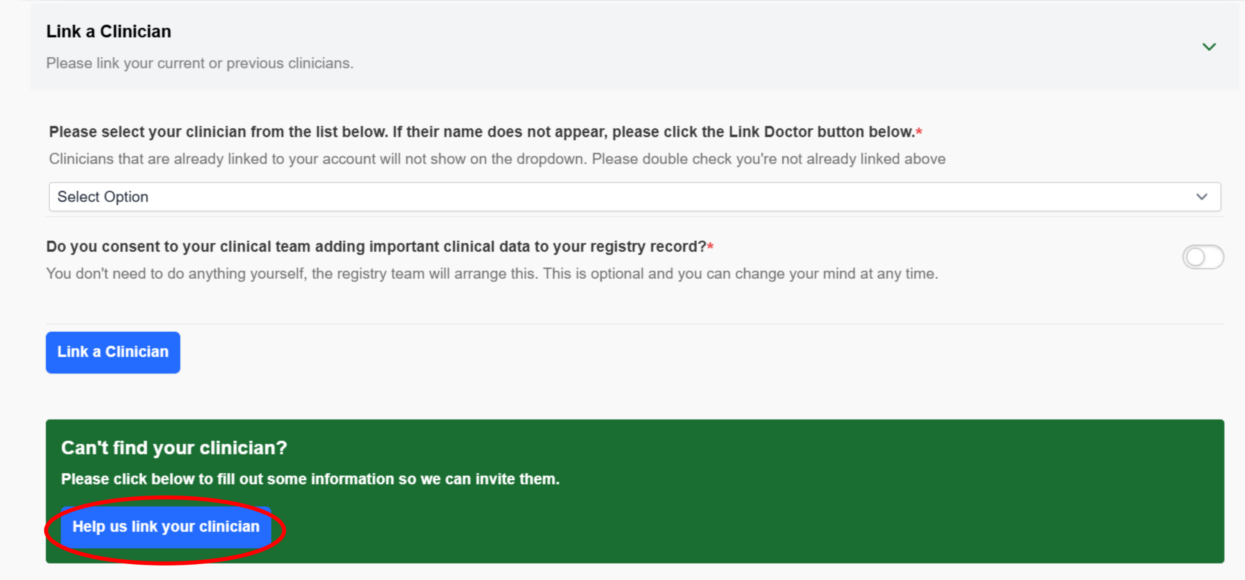
Please select your clinician from the list below. If their name does not appear, please click the Link Doctor button below.*

Clinicians that are already linked to your account will not show on the dropdown. Please double check you're not already linked above

Do you consent to your clinical team adding important clinical data to your registry record?*

You don't need to do anything yourself, the registry team will arrange this. This is optional and you can change your mind at any time.

- If your doctor is not on the list but you would like them to be invited to the registry, please click '**New Clinician Details'** at the bottom of the **'Invite a Clinician'** page.



Link a Clinician

Please link your current or previous clinicians.

Please select your clinician from the list below. If their name does not appear, please click the Link Doctor button below.*

Clinicians that are already linked to your account will not show on the dropdown. Please double check you're not already linked above

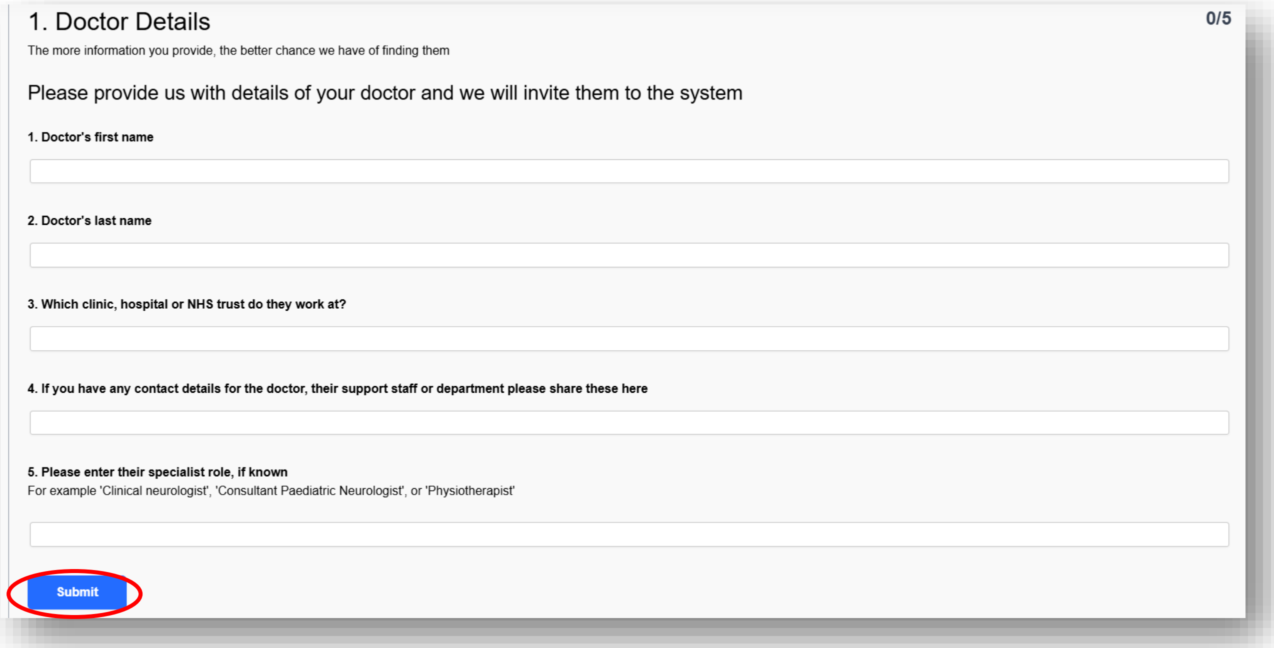
Do you consent to your clinical team adding important clinical data to your registry record?*

You don't need to do anything yourself, the registry team will arrange this. This is optional and you can change your mind at any time.

Can't find your clinician?

Please click below to fill out some information so we can invite them.

- When you click '**Help us link your clinician'**, you will be taken to a new form that helps us identify and invite your doctor. Complete the details and press '**Submit**' to finish the form.



1. Doctor Details

0/5

The more information you provide, the better chance we have of finding them

Please provide us with details of your doctor and we will invite them to the system

1. Doctor's first name

2. Doctor's last name

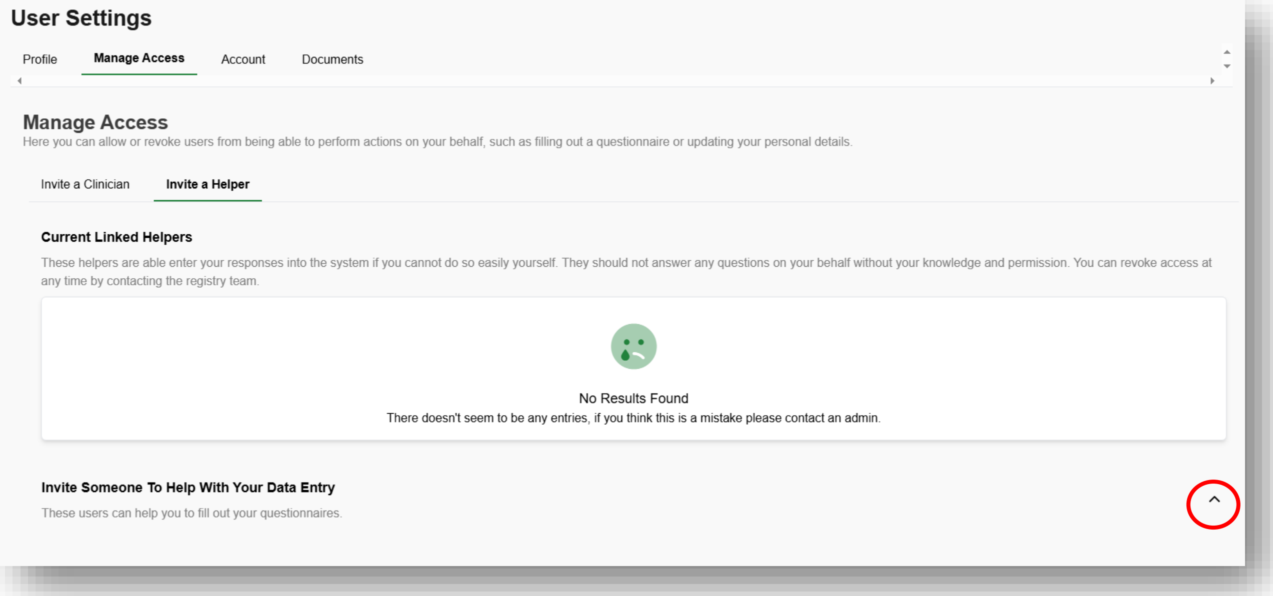
3. Which clinic, hospital or NHS trust do they work at?

4. If you have any contact details for the doctor, their support staff or department please share these here

5. Please enter their specialist role, if known
For example 'Clinical neurologist', 'Consultant Paediatric Neurologist', or 'Physiotherapist'

Step 10 – Adding a support user

- Our new registry platform allows you to invite a support user to input information on your behalf. You may wish to invite a trusted friend, carer or family member to help you with data entry. Unlike the old system, your support user will have their own log-in to access your patient profile.
- Details of any linked support users or helpers can be found in the **Manage Access** area of your **account** settings.



User Settings

Manage Access

Here you can allow or revoke users from being able to perform actions on your behalf, such as filling out a questionnaire or updating your personal details.

Current Linked Helpers

These helpers are able enter your responses into the system if you cannot do so easily yourself. They should not answer any questions on your behalf without your knowledge and permission. You can revoke access at any time by contacting the registry team.

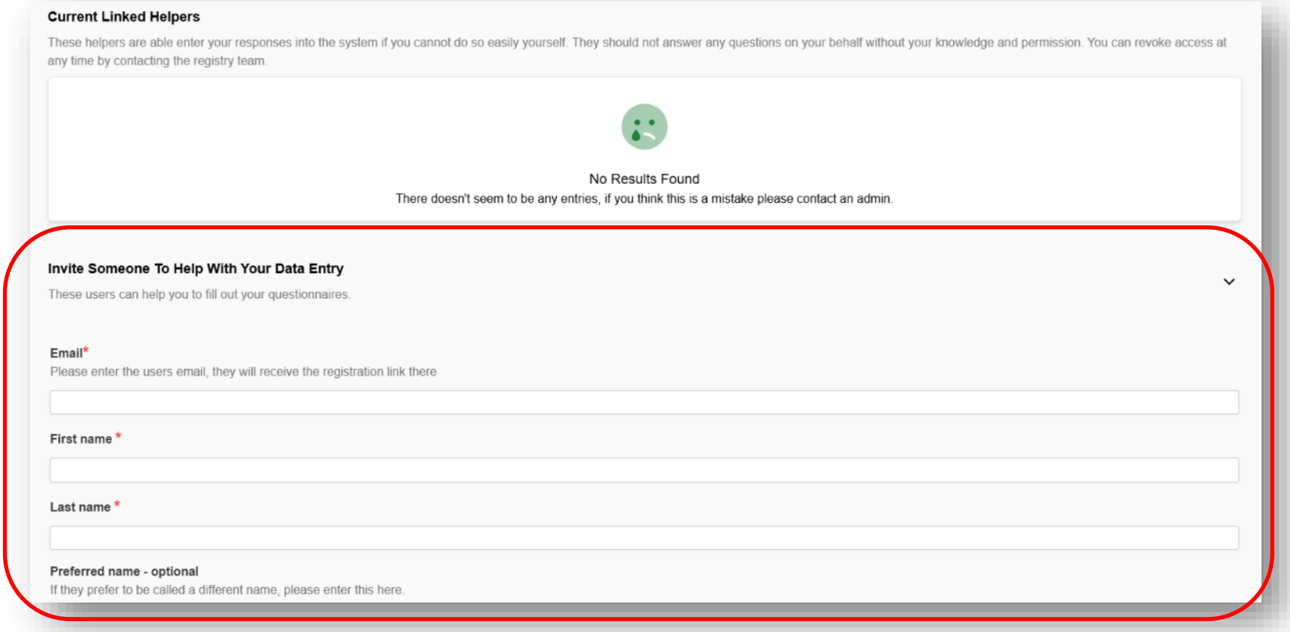
No Results Found

There doesn't seem to be any entries, if you think this is a mistake please contact an admin.

Invite Someone To Help With Your Data Entry

These users can help you to fill out your questionnaires.

- To invite a new person to help with your registry updates, click on the arrow icon circled above to open the invitation form.



Current Linked Helpers

These helpers are able enter your responses into the system if you cannot do so easily yourself. They should not answer any questions on your behalf without your knowledge and permission. You can revoke access at any time by contacting the registry team.

No Results Found

There doesn't seem to be any entries, if you think this is a mistake please contact an admin.

Invite Someone To Help With Your Data Entry

These users can help you to fill out your questionnaires.

Email*

Please enter the users email, they will receive the registration link there

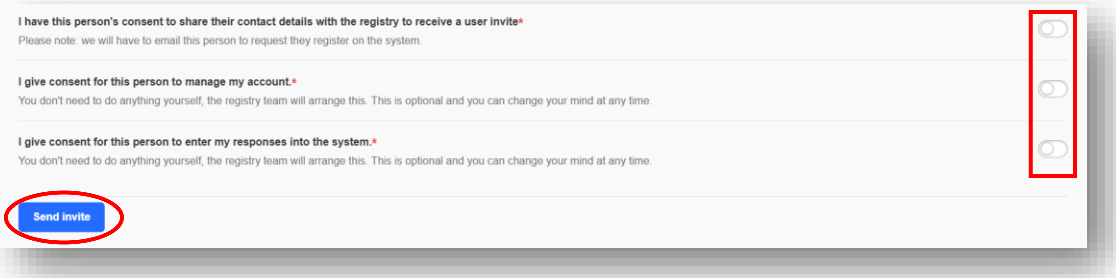
First name *

Last name *

Preferred name - optional

If they prefer to be called a different name, please enter this here.

- Complete the details required in the form then scroll to the bottom of the page.
- Confirm that you have permission to share your helper's contact details, then accept the consent statements and click 'Send invite'.



I have this person's consent to share their contact details with the registry to receive a user invite*

Please note: we will have to email this person to request they register on the system.

I give consent for this person to manage my account.*

You don't need to do anything yourself, the registry team will arrange this. This is optional and you can change your mind at any time.

I give consent for this person to enter my responses into the system.*

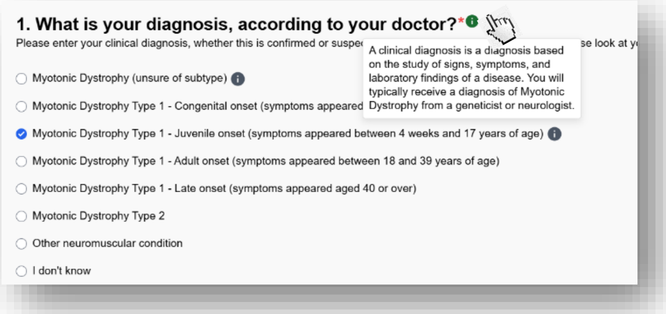
You don't need to do anything yourself, the registry team will arrange this. This is optional and you can change your mind at any time.

Tips and tricks – How to complete different questions

- You may notice some differences in the new registry platform. To help you complete your patient questionnaire, here are some tips on how to answer the questions.

Information Icons

- If you do not understand what a question is asking, there may be an information icon to help you.
- To access the extra information, you can hover your curser over the icon, and the information box will appear.
- Some information boxes have links to external websites with further details.



1. What is your diagnosis, according to your doctor?*

Please enter your clinical diagnosis, whether this is confirmed or suspe

se look at y

A clinical diagnosis is a diagnosis based on the study of signs, symptoms, and laboratory findings of a disease. You will typically receive a diagnosis of Myotonic Dystrophy from a geneticist or neurologist.

○ Myotonic Dystrophy (unsure of subtype)

○ Myotonic Dystrophy Type 1 - Congenital onset (symptoms appeared

● Myotonic Dystrophy Type 1 - Juvenile onset (symptoms appeared between 4 weeks and 17 years of age)

○ Myotonic Dystrophy Type 1 - Adult onset (symptoms appeared between 18 and 39 years of age)

○ Myotonic Dystrophy Type 1 - Late onset (symptoms appeared aged 40 or over)

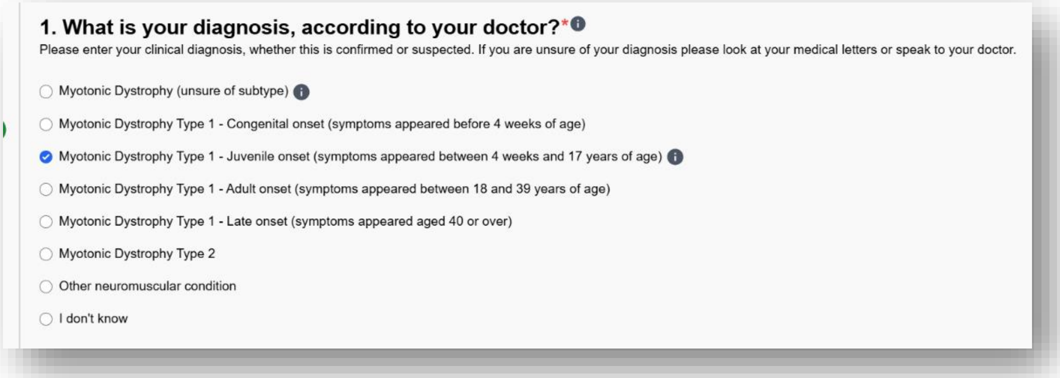
○ Myotonic Dystrophy Type 2

○ Other neuromuscular condition

○ I don't know

Single Choice Questions

- For this question type, you select a single option from a list of answers.



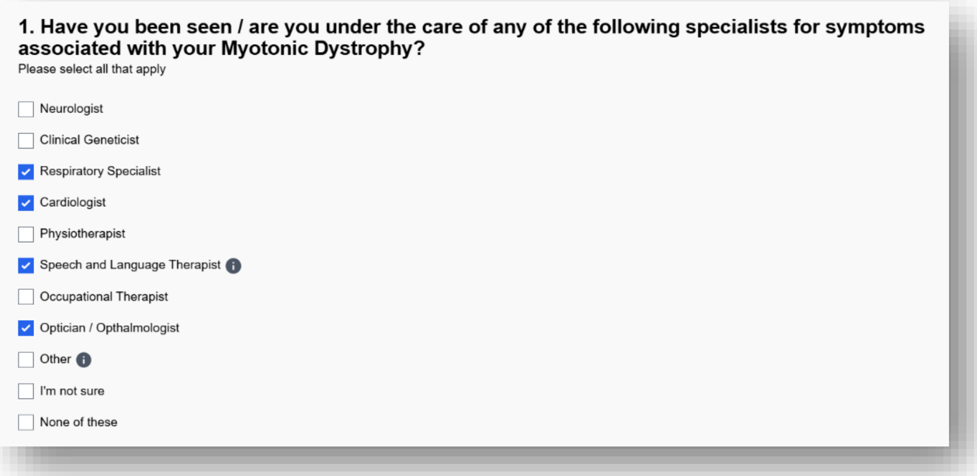
1. What is your diagnosis, according to your doctor?*

Please enter your clinical diagnosis, whether this is confirmed or suspected. If you are unsure of your diagnosis please look at your medical letters or speak to your doctor.

○ Myotonic Dystrophy (unsure of subtype)
○ Myotonic Dystrophy Type 1 - Congenital onset (symptoms appeared before 4 weeks of age)
● Myotonic Dystrophy Type 1 - Juvenile onset (symptoms appeared between 4 weeks and 17 years of age)
○ Myotonic Dystrophy Type 1 - Adult onset (symptoms appeared between 18 and 39 years of age)
○ Myotonic Dystrophy Type 1 - Late onset (symptoms appeared aged 40 or over)
○ Myotonic Dystrophy Type 2
○ Other neuromuscular condition
○ I don't know

Multiple Choice Questions

- For this question type you can select any applicable answers from the list.



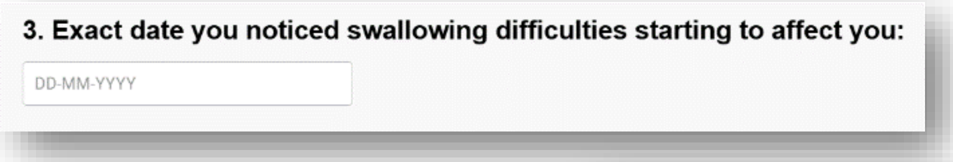
1. Have you been seen / are you under the care of any of the following specialists for symptoms associated with your Myotonic Dystrophy?

Please select all that apply

☐ Neurologist
☐ Clinical Geneticist
☑ Respiratory Specialist
☑ Cardiologist
☐ Physiotherapist
☑ Speech and Language Therapist
☐ Occupational Therapist
☑ Optician / Opthalmologist
☐ Other
☐ I'm not sure
☐ None of these

Date Questions

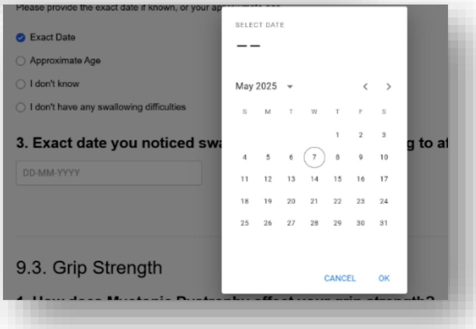
- Some questions may ask you to provide an exact date (e.g. of symptom onset) if known.



3. Exact date you noticed swallowing difficulties starting to affect you:

DD-MM-YYYY

- When you complete the question, you can type the date, or use the calendar pop-up to find your chosen date.



Please provide the exact date if known, or your ap

● Exact Date
○ Approximate Age
○ I don't know
○ I don't have any swallowing difficulties

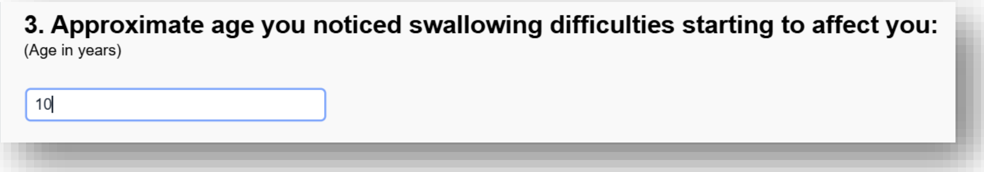
3. Exact date you noticed swa

DD-MM-YYYY

9.3. Grip Strength

Number Questions

- Some questions may ask you to provide an age or number.
- You need to type your answers in number form, rather than written as a word (i.e. ‘10’ not ‘ten’).

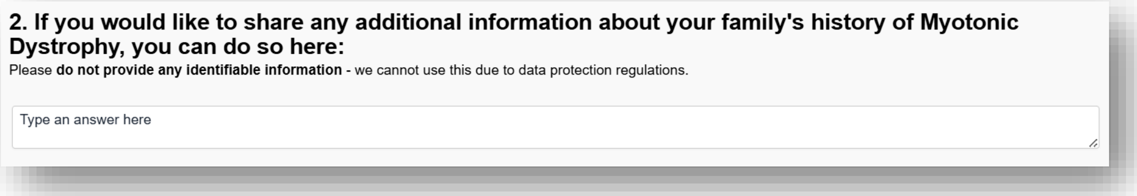


3. Approximate age you noticed swallowing difficulties starting to affect you:
(Age in years)

10

Open Answer Questions

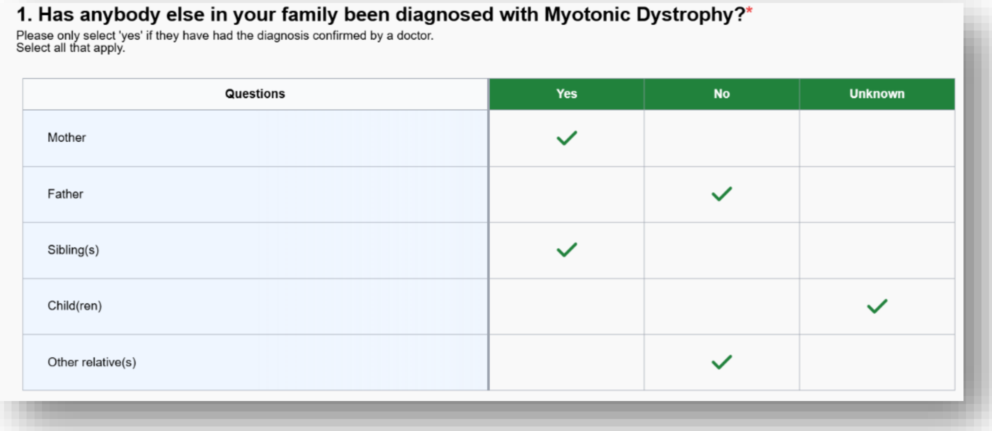
- This question type lets you type your answer out in free text.



2. If you would like to share any additional information about your family's history of Myotonic Dystrophy, you can do so here:
Please **do not provide any identifiable information** - we cannot use this due to data protection regulations.

Matrix Questions

- These questions have questions and answers in columns.
- You can only select one answer option per question.

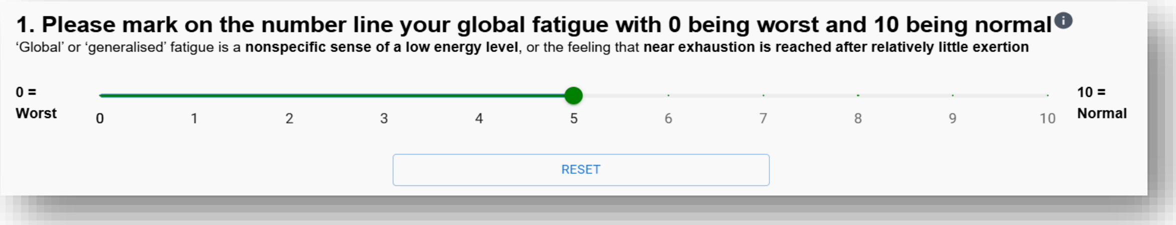


1. Has anybody else in your family been diagnosed with Myotonic Dystrophy?*
Please only select 'yes' if they have had the diagnosis confirmed by a doctor.
Select all that apply.

Questions	Yes	No	Unknown
Mother	✓		
Father		✓	
Sibling(s)	✓		
Child(ren)			✓
Other relative(s)		✓	

Scale Questions

- These questions ask you to place a value on a scale that accurately represents your condition.
- You can slide the value up and down the scale.

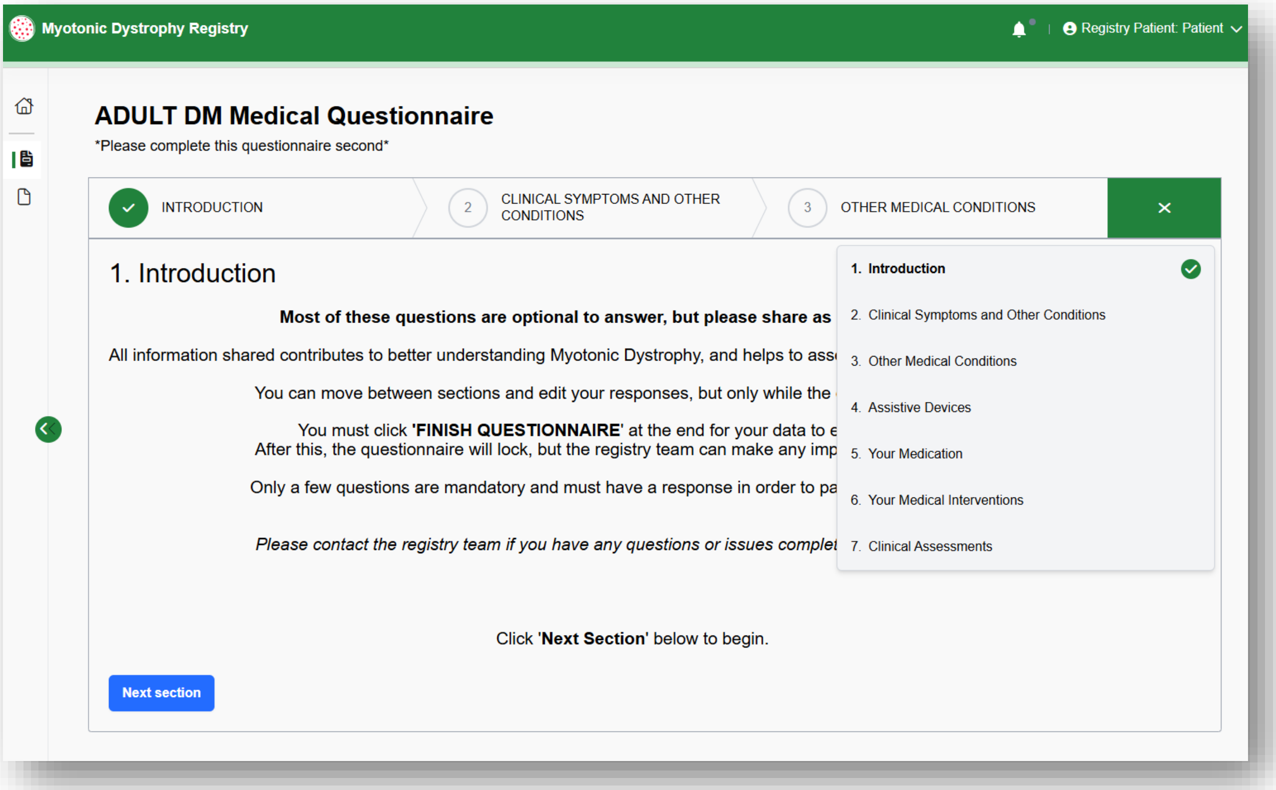


1. Please mark on the number line your global fatigue with 0 being worst and 10 being normal
‘Global’ or ‘generalised’ fatigue is a **nonspecific sense of a low energy level**, or the feeling that **near exhaustion is reached after relatively little exertion**

Step 11 – Adding additional information

Medical Questionnaire

- A new questionnaire has been added to the registry to allow both patients and clinicians to add their medical information.
- You will find the questionnaire in your patient questionnaires tab.
- **The questionnaire is designed to capture clinical information that you may find in your doctor's notes or letters. If you do complete this questionnaire, we kindly ask that you also upload evidence of this information where possible.**



ADULT DM Medical Questionnaire

Please complete this questionnaire second

1. Introduction

Most of these questions are optional to answer, but please share as

All information shared contributes to better understanding Myotonic Dystrophy, and helps to ass

You can move between sections and edit your responses, but only while the

You must click '**FINISH QUESTIONNAIRE**' at the end for your data to e
After this, the questionnaire will lock, but the registry team can make any imp

Only a few questions are mandatory and must have a response in order to pa

Please contact the registry team if you have any questions or issues complet

Click '**Next Section**' below to begin.

Troubleshooting

- If you see an error message at any point, please try to take a screenshot or a photograph of the screen if possible and make a note of the time. This can help our IT team identify what caused the problem.

- You can try refreshing the webpage which may resolve the issue without the need for any further steps. The refresh button usually looks like a circular arrow and will be somewhere near the website address (example circled below). It might look slightly different or be in a different position at the top of your screen depending on which web browser you are using. Clicking this button will re-load the page you were on so you may need to enter a small amount of information again.

- Please contact the registry team if you have any problems using the system or are unsure what to do. You can click here to send us an email, or contact us by telephone on 0191 241 8640. We can guide you through the process step by step, or can even complete your registration for you over the phone if you are not confident using the website.

Problems logging in?

- If you have any problems with logging in to the registry, **please do not try to register again** as this can lead to duplicate records. Please contact the registry team who can help identify and resolve the issue.

- If you are unsure if you have registered previously or not, please contact us to check if you have any historical or partially completed registrations.

If you have any questions, comments, or feedback, or would like more information about anything to do with the UK Myotonic Dystrophy Patient Registry please get in touch with the registry team who are on hand to help.