

FSHD Registry	
Facioscapulohumeral Muscular Dystrophy Module	
Data field	Entry guidelines and help tool
Age at clinical diagnosis	Enter the patient's age in years at clinical diagnosis.
Age at molecular diagnosis	Enter the patient's age in years at molecular diagnosis.
Was the patient the first person in their family to have the diagnosis of FSHD?	Answer (Yes/No) if known.
Which of the following has the patient received?	Tick any that apply.
Current best motor function	Select the option that best describes the patient's current best motor function from the drop-down list.
Wheelchair or mobility scooter use	Select the appropriate wheelchair/mobility scooter use if known.
Age when started using wheelchair/scooter	If part-time or full-time wheelchair/scooter use selected, enter age at first use here.
Heart condition	Has the patient been diagnosed with a heart condition? Select from drop-down list.
Non-invasive ventilation	Does the patient regularly use a non-invasive ventilation device?
Non-invasive ventilation start date	Select the date from which the patient started using a non-invasive ventilation device.
Invasive ventilation	Does the patient regularly use an invasive ventilation device?
Invasive ventilation start date	Select the date from which the patient started using any invasive ventilation device.
Positive family history	Select if any family members have also been diagnosed with Facioscapulohumeral Muscular Dystrophy.
Ethnic origin	How does the patient describe their ethnic origin? Select from the drop-down list.
Has the patient ever been pregnant?	Select answer (Yes/No) if known.
Number of pregnancies	Enter the number of pregnancies. Count the number of pregnancies even if they did not have the baby.
Number of childbirths	Enter the number of childbirths.

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Did the patient's condition deteriorate during or in the 6 months following their pregnancy?	Answer (Yes/No) if known.
Has the patient ever participated in a research study for FSHD?	Answer (Yes/No) if known.
Has the patient ever received experimental treatment for FSHD?	Answer (Yes/No) if known.
What was the treatment/clinical trial?	Enter the treatment or clinical trial name.
Does the patient take vitamins or supplements for their health?	Enter any vitamins or supplements taken by the patient.
Medications table	Identify all known medications and dosage.
Other registries	Other registries the individual is registered in.
Clinical features	Select 'Yes' for any that apply, or tick the box if there are no clinical features.
Age of onset of clinical features	If the patient has clinical features, enter the age at onset of these features in years.
Periscapular shoulder weakness	Select the option that best describes the patient's current best shoulder mobility from the drop-down list.
Hip girdle weakness: Chair mobility	Select the option that best describes the patient's current best chair mobility from the drop-down list.
Hip girdle weakness: Bed mobility	Select the option that best describes the patient's current best bed mobility from the drop-down list.
Retinal vascular disease attributable to FSHD	Has the patient been diagnosed with retinal vascular disease attributable to FSHD?
Retinal vascular disease age at diagnosis	If answered yes to having retinal vascular disease, what was the approximate age when diagnosed with retinal vascular disease?
Hearing loss	Has the patient been diagnosed with hearing loss?

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Hearing loss start age	If answered yes to having hearing loss, what was the approximate age when diagnosed with hearing loss?
Scapular fixation	Has the patient had scapular fixation?
Year of first scapular fixation surgery	If you answered yes to scapular fixation, enter year of first surgery here
Year of second scapular fixation surgery	If applicable, enter year of second surgery here
Pain	Does the patient have any pain? Select the area of pain as appropriate, or if no pain, select 'No I don't have any pain'.
Height	Enter the patient's height in centimetres.
Weight	Enter the patient's body weight in kilograms.
Socioeconomic factors	
What is your current occupation?	Select the employment status of the patient.
Comments	Enter any other comments about socioeconomic factors.
Has FSHD affected your employment?	Select from drop-down list.
Highest level of education completed.	Select the patient's highest level of education.
Genetic Module	
Detection of D4Z4 fragment	Result of testing for detection of a contracted D4Z4 fragment derived from chromosome 4q35. Select from drop-down list.
Other	Record other result if required.
D4Z4 Method of Testing	Select method of testing from drop-down list.
Other Method	Record other method if required.
Testing Site of D4Z4	Select site of testing.
Other Testing Site	Record other testing site if required.
Patient completed genetic test result	Select from drop-down list.