The United Kingdom Facioscapulohumeral Dystrophy Patient Registry

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Data source | Human | Disease registry

Administrative details

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Data source ID

37237

Data source acronym

The UK FSHD Patient Registry

Data holder

John Walton Muscular Dystrophy Research Centre, Newcastle University

Data source type

Disease registry

Main financial support

Funds from patients organisations, charity and foundations

Care setting

Hospital outpatient care

Other

Data source qualification

If the data source has successfully undergone a formal qualification process (e.g., from the EMA, ISO or other certifications), this should be described.

No

Data source website

https://www.fshd-registry.org.uk/

Contact details

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Data source regions and languages

Data source countries

Ireland

United Kingdom

Data source languages

English

Data source establishment

Data source established

10/05/2013

Data source time span

First collection: 10/05/2013

The date when data started to be collected or extracted.

Publications

Data source publications

Evangelista T, Wood L, Fernandez-Torron R, Williams M, Smith D, Lunt P, Hudson J, Norwood F, Orrell R, Willis T, Hilton-Jones D. Design, set-up and utility of the UK facioscapulohumeral muscular dystrophy patient registry. Journal of neurology. 2016 Jul 1;263(7):1401-8.

Website for full list of registry publications

Data elements collected

The data source contains the following information

Disease information

Does the data source collect information with a focus on a specific disease? This might be a patient registry or other similar initiatives.

Yes

Disease details

Facioscapulohumeral muscular dystrophy

Rare diseases

Are rare diseases captured? In the European Union a rare disease is one that affects no more than 5 people in 10,000.

Yes

Pregnancy and/or neonates

Does the data source collect information on pregnant women and/or neonatal subpopulation (under 28 days of age)?

Yes

Hospital admission and/or discharge

No

ICU admission

Is information on intensive care unit admission available?

No

Cause of death

Not Captured

Prescriptions of medicines

Not Captured

Dispensing of medicines

Not Captured

Advanced therapy medicinal products (ATMP)

Is information on advanced therapy medicinal products included? A medicinal product for human use that is either a gene therapy medicinal product, a somatic cell therapy product or a tissue engineered products as defined in Regulation (EC) No 1394/2007 [Reg (EC) No 1394/2007 Art 1(1)].

No

Contraception

Is information on the use of any type of contraception (oral, injectable, devices etc.) available?

No

Indication for use

Does the data source capture information on the therapeutic indication for the use of medicinal products?

Captured

Indication vocabulary

Not coded (Free text)

Medical devices

Is information on medicinal devices (e.g., pens, syringes, inhalers) available?

Yes

Administration of vaccines

No

Procedures

Does the data source capture information on procedures (e.g., diagnostic tests, therapeutic, surgical interventions)?

Captured

Procedures vocabulary

Not coded (Free text)

Healthcare provider

Is information on the person providing healthcare (e.g., physician, pharmacist, specialist) available? The healthcare provider refers to individual health professionals or a health facility organisation licensed to provide health care diagnosis and treatment services including medication, surgery and medical devices.

Yes

Clinical measurements

Is information on clinical measurements (e.g., BMI, blood pressure, height) available?

Yes

Genetic data

Are data related to genotyping, genome sequencing available?

Captured

Genetic data vocabulary

Other

Genetic data vocabulary, other

Not coded (free text)

Biomarker data

Does the data source capture biomarker information? The term “biomarker” refers to a broad subcategory of medical signs (objective indications of medical state observed from outside the patient), which can be measured accurately and reproducibly. For example, haematological assays, infectious disease markers or metabolomic biomarkers.

Not Captured

Patient-reported outcomes

Is information on patient-reported outcomes (e.g., quality of life) available?

No

Patient-generated data

Is patient-generated information (e.g., from wearable devices) available?

Yes

Units of healthcare utilisation

Are units of healthcare utilisation (e.g., number of visits to GP per year, number of hospital days) available or can they be derived? Units of healthcare utilisation refer to the quantification of the use of services for the purpose of preventing or curing health problems.

No

Unique identifier for persons

Are patients uniquely identified in the data source?

Yes

Diagnostic codes

Captured

Diagnosis / medical event vocabulary

Other

Diagnosis / medical event vocabulary, other

Diagnosis uses the standard definition of mild, moderate, severe based on symptom onset

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Medicinal product information

Captured

Medicinal product information collected

Brand name

Dosage regime

Dose

Medicinal product vocabulary

Not coded (Free text)

Quality of life measurements

Captured

Quality of life measurements vocabulary

other

SF-36

Quality of life measurements, other

SF-MPQ, INQOL

Lifestyle factors

Not Captured

Sociodemographic information

Captured

Sociodemographic information collected

Age

Ethnicity

Gender

Quantitative descriptors

Population Qualitative Data

Population age groups

Paediatric Population (< 18 years)

Preterm newborn infants (0 – 27 days)

Term newborn infants (0 – 27 days)

Infants and toddlers (28 days – 23 months)

Children (2 to < 12 years)

Adolescents (12 to < 18 years)

Adults (18 to < 46 years)

Adults (46 to < 65 years)

Elderly (≥ 65 years)

Adults (65 to < 75 years)

Adults (75 to < 85 years)

Adults (85 years and over)

Estimated percentage of the population covered by the data source in the catchment area

Disease prevelance is uncertain but estimated at 1 in 8,000 - registry currently has 906 patients so around 11% of expected UK FSHD population

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Description of the population covered by the data source in the catchment area whose data are not collected (e.g., people who are registered only for private care)

Any patients who have chosen not to self-register, or who are undiagnosed or unaware of the registry

Population

Population size

1133

Active population size

906

Population by age group

Age group	Population size	Active population size
Paediatric Population (< 18 years)	24	18
Preterm newborn infants (0 – 27 days)	0	0
Term newborn infants (0 – 27 days)	0	0
Infants and toddlers (28 days – 23 months)	0	0
Children (2 to < 12 years)	3	2
Adolescents (12 to < 18 years)	21	16
Adults (18 to < 46 years)	408	317
Adults (46 to < 65 years)	434	345
Elderly (≥ 65 years)	267	226
Adults (65 to < 75 years)	163	140
Adults (75 to < 85 years)	95	81
Adults (85 years and over)	9	5

Median observation time

Median time (years) between first and last available records for unique individuals captured in the data source

51.00

Median time (years) between first and last available records for unique active individuals (alive and currently registered) capt

52.00

Data flows and management

Access and validation

Governance details

Documents or webpages that describe the overall governance of the data source and processes and procedures for data capture and management, data quality check and validation results (governing data access or utilisation for research purposes).

https://www.fshd-registry.org.uk/

Biospecimen access

Are biospecimens available in the data source (e.g., tissue samples)?

Yes

Biospecimen access conditions

Not directly, but potential for local linkage with Newcastle biobank specimens if relevant consents in place.

Access to subject details

Can individual patients/practitioners/practices included in the data source be contacted?

Yes

Description of data collection

Online patient-entered questionnaires. Annual reminders sent by email, alongside newsletters and other registry correspondence (e.g. clinical trial information). Genetic test information can only be inputted by nominated professional (e.g. consultant, genetic counsellor, registry manager) but patients can upload PDF of their report for analysis and data entry via a secure upload portal.

Event triggering registration

Event triggering registration of a person in the data source

Other

Event triggering registration of a person in the data source, other

User self-registers via registry website

Event triggering de-registration of a person in the data source

Death

Emigration

Loss to follow up

Other

Event triggering de-registration of a person in the data source, other

Patient or user records logged as inactive if deceased, non-UK resident, LTFU with no active contact details, or if patients requests to be withdrawn.

Event triggering creation of a record in the data source

Registry user updates patient information. Automatic annual reminders are sent to patients and clinicians via email, one year after their last login.

Data source linkage

Linkage

Is the data source described created by the linkage of other data sources (prelinked data source) and/or can the data source be linked to other data source on an ad-hoc basis?

No

Linkage description, possible linkage

Not linked directly, but data can be compared with other registries collecting TREAT-NMD Core Dataset for FSHD.

Data management specifications that apply for the data source

Informed consent for use of data for research

Other

Possibility of data validation

Can validity of the data in the data source be verified (e.g., access to original medical charts)?

Yes

Data source preservation

Are records preserved in the data source indefinitely?

Yes

Approval for publication

Is an approval needed for publishing the results of a study using the data source?

Yes

Informed consent, other

Patients consent to their de-identified data being used for research. The registry steering committee evaluate requests for data reports or trial recruitment support.

Common Data Model (CDM) mapping

CDM mapping

Has the data source been converted (ETL-ed) to a common data model?

No