

Facioscapulohumeral Muscular Dystrophy CDE Highlight Summary Document

NIH Resources

The NINDS also strongly encourages researchers to use these NIH developed materials for NINDS-sponsored research, when appropriate. Utilization of these resources will enable greater consistency for NINDS-sponsored research studies. These tools are free of charge.

- National Institutes of Health (NIH) Toolbox
- Quality of Life in Neurological Disorders (Neuro-QOL)
- Patient-Reported Outcomes Measurement Information System (PROMIS)

Suicidal Ideation

Investigators should review the FDA's "[Guidance for Industry: Suicidal Ideation and Behavior: Prospective Assessment of Occurrence in Clinical Trials](#)" for the most up-to-date information about suicidal ideation and behavior. One scale that FDA suggests is the Columbia Suicide Severity Rating Scale (C-SSRS) (available at <http://www.cssrs.columbia.edu>)

Disease/Domain	Recommendations
Facioscapulohumeral muscular dystrophy	<p>These instruments and elements are recommended for use in FSHD studies:</p> <p>Core elements:</p> <p>See Start-Up Resources Listing document (FSHD Start-Up Resource Listing)</p> <p>Supplemental – Highly Recommended instruments: 6 Minute Walk Test, Grip Strengthⁱ, Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale, Maximum Voluntary Isometric Contraction Testing (MVICT)</p>
Participant / Subject Characteristics; Demographics	<p>Core: General Core, Demographicsⁱⁱ</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Demographics</p> <p>Exploratory: None</p>
Participant / Subject Characteristics; Social Status	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Social Status</p> <p>Exploratory: None</p>

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Disease/Domain	Recommendations
Participant/Subject History and Family History; Epidemiology/ Environmental History	Core: None Supplemental – Highly Recommended: None Supplemental: None Exploratory: World Health Organization Disability Assessment Schedule (WHODAS)
Participant/Subject History and Family History; General Health History	Core: None Supplemental – Highly Recommended: None Supplemental: Family History, Medical History, Pregnancy Outcome ⁱⁱⁱ Exploratory: None
Assessments and Examinations; Imaging Diagnostics	Core: None Supplemental – Highly Recommended: None Supplemental: Dual X-Ray Absorptiometry (DXA), Magnetic Resonance Imaging (MRI) Exploratory: None
Assessments and Examinations; Laboratory Tests and Biospecimens/ Biomarkers	Core: None Supplemental – Highly Recommended: Laboratory Tests and Tracking Supplemental: Mutation Analysis Exploratory: None
Assessments and Examinations; Non-Imaging Diagnostics	Core: None Supplemental – Highly Recommended: Electrocardiogram (ECG) Supplemental: Electrocardiogram (ECG) Exploratory: None
Assessments and Examinations; Physical/Neurological Examination	Core: None Supplemental – Highly Recommended: None Supplemental: Polysomnography Exploratory: Electrical Impedance Myography (EIM)
Treatment/ Intervention Data; Drugs	Core: None Supplemental – Highly Recommended: Prior and Concomitant Medications Supplemental: None Exploratory: None



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Disease/Domain	Recommendations
Treatment/ Intervention Data; Therapies	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Respiratory Interventions</p> <p>Exploratory: None</p>
Outcomes and Endpoints; Functional Status	<p>Core: None</p> <p>Supplemental – Highly Recommended: 6 Minute Walk Test</p> <p>Supplemental: Goniometry, Stair Climb, Timed Up and Go (TUG)</p> <p>Exploratory: 2 Minute Walk Test</p>
Outcomes and Endpoints; Muscle Strength Testing	<p>Core: None</p> <p>Supplemental – Highly Recommended: Grip Strength, Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale, Maximum Voluntary Isometric Contraction Testing (MVICT)</p> <p>Supplemental: Hand Held Dynamometry, Pinch Strength</p> <p>Exploratory: None</p>
Outcomes and Endpoints; Pain	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Brief Pain Inventory (Short Form), McGill Pain Questionnaire</p> <p>Exploratory: None</p>
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	<p>Core: None</p> <p>Supplemental – Highly Recommended: Pulmonary Function Testing^{iv}</p> <p>Supplemental: Measures of Gas Exchange</p> <p>Exploratory: None</p>
Outcomes and Endpoints; Quality of Life	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Individualized Neuromuscular Quality of Life Questionnaire (INQoL), Short Form 36-Item Health Survey-36 (SF-36)</p> <p>Exploratory: Quality of Life in Neurological Disorders (NeuroQoL)</p>

ⁱ Supplemental – Highly Recommended for Facioscapulohumeral Muscular Dystrophy (FSHD) studies involving muscle strength testing.

ⁱⁱ Contains Core CDEs

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ⁱⁱⁱ To be completed by female participants/subjects only.

^{iv} The most useful measure of pulmonary function for FSHD is sitting and supine Forced Vital Capacity. Pulmonary Function Testing (PFT) is likely to have the greatest yield in clinical studies in which subjects are non-ambulatory and have severe weakness or early (“infantile”) onset of disease. PFT should be performed with a face mask in FSHD due to the inability to form a tight seal with a mouthpiece.