Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Start-up Resource – NINDS Facioscapulohumeral Muscular Dystrophy CDE Recommendations

The National Institute of Neurological Disorders and Stroke (NINDS) and other Federal agencies and international organizations have the common mission of developing data standards for clinical research. Through the efforts of subject-specific working groups, topic-driven data elements have been created. The first set of Common Data Elements (CDEs) for Facioscapulohumeral Dystrophy was developed in 2014. The Core data elements to be used by an investigator when beginning a research study in this disease/disorder are listed in this resource document. All other recommendations are listed on the website and should be considered based on study type.

Each CDE or instrument could be classified according to the definitions below:

General Core: A data element that is required for all NINDS funded studies.

Disease Core: A data element that collects essential information applicable to any disease-specific study, including all therapeutic areas. The NINDS and its appointed working groups assign the disease "Core" classification based on the current clinical research best practices. In each case, the disease Core CDEs are a small subset of the available CDEs, where it is anticipated that investigators will need to collect the disease Core CDEs on any type of study. These are required for all disease-specific studies.

Disease Supplemental - Highly Recommended: A data element which is essential based on certain conditions or study types in clinical research studies. In most cases, these have been used and validated in the disease area. These data elements are strongly recommended for the specified disease condition, study type or design.

Disease Supplemental: A data element which is commonly collected in clinical research studies. Use depends upon the study design, protocol or type of research involved. These are recommended, but not required, for studies.

Disease Exploratory: A data element that requires further validation but may fill current gaps in the CDEs and/or substitute for an existing CDE once validation is complete. Such data elements show great promise but require further validation before they are ready for prime-time use in clinical research studies. They are reasonable to use with the understanding that it has limited validation in the target group.

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

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

Core CDEs for all NINDS Studies[1]:

Domain/Sub-Domain	CDE Name	CDE ID	Study Type
Participant Characteristics; Demographics	Birth date	C00007	All studies
Participant Characteristics; Demographics	Ethnicity USA category	C00020	All studies
Participant Characteristics; Demographics	Race USA category	C00030	All studies
Participant Characteristics; Demographics	Birth sex assigned type	C58676	All studies
Participant Characteristics; Demographics	Gender identity type	C58677	All studies
Participant History and Family History; General Health History	Medical history condition text	C00322	All studies
Participant History and Family History; General Health History	Medical history condition SNOMED CT code	C00313	All studies

[1] Note: Education year count C00015 is no longer a general Core CDE

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Supplemental – Highly Recommended CDEs for Facioscapulohumeral Muscular Dystrophy:

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Non-Imaging Diagnostics	ECG assessment data and time	C04501
Assessments and Examinations; Non-Imaging Diagnostics	ECG atrial arrhythmia type	C04500
Assessments and Examinations; Non-Imaging Diagnostics	ECG global result type	C04502
Assessments and Examinations; Non-Imaging Diagnostics	ECG heart rate	C04503
Assessments and Examinations; Non-Imaging Diagnostics	ECG heart rhythm result type	C04504
Assessments and Examinations; Non-Imaging Diagnostics	ECG left ventricular hypertrophy indicator	C04505
Assessments and Examinations; Non-Imaging Diagnostics	ECG previous myocardial infarction indicator	C04508
Assessments and Examinations; Non-Imaging Diagnostics	ECG QT interval	C04511
Assessments and Examinations; Non-Imaging Diagnostics	ECG QTc interval	C04512
Assessments and Examinations; Non-Imaging Diagnostics	ECG right ventricular hypertrophy indicator	C04506
Assessments and Examinations; Non-Imaging Diagnostics	ECG ST segment abnormality indicator	C04513
Assessments and Examinations; Non-Imaging Diagnostics	ECG T wave abnormality indicator	C04514
Assessments and Examinations; Non-Imaging Diagnostics	ECG ventricular arrhythmia type	C04515

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Non-Imaging Diagnostics	ECG heart rhythm result other text	C18777
Assessments and Examinations; Non-Imaging Diagnostics	ECG atrial arrhythmia other text	C18776
Assessments and Examinations; Non-Imaging Diagnostics	ECG ventricular arrhythmia other text	C18778
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab panel category	C01703
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab panel other text	C18729
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab specimen collection date and time	C01701
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Specimen accession number	C01700
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test name	C19887
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test LOINC code	C01720

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test performed indicator	C01708
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test result unit of measure UCUM code	C01713
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test abnormality significance type	C01707
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test abnormality significance type	C01707
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test name	C01705
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test other text	C18731
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test result status	C01709
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test result unit of measure	C01711

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Lab test result value	C01706
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant use indicator	C02002
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant name	C02014
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant RXNorm code	C02025
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant indication text	C02024
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant dose	C02006
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant dose other text	C18736
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant dose unit of measure	C02022
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant dose frequency	C02013
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant route type	C02015

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Domain; Sub-Domain	Data Element	CDE ID
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant route other text	C18737
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant ongoing indicator	C02016
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant end date and time	C02008
Treatment/Intervention Data; Prior and Concomitant Medications	Medication prior or concomitant start date and time	C02016
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Pregnancy test date and time	C01702
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Pregnancy test not applicable reason	C01714
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Pregnancy test performed indicator	C10896
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Pregnancy test qualitative result value	C01710

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Pregnancy test specimen type	C01704
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function complete exhalation indicator	C12310
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity cough exhalation indicator	C12314
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity early termination indicator	C12317
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity large value range indicator	C12316
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity leak indicator	C12315
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function index of lung function best trial measurement	C12318
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function index of lung function measurement	C12312

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function lung function measurement	C12516
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function maximal pressure trial difference indicator	C12313
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test date and time	C11098
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test equipment manufacturer name	C12303
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test equipment model name	C12304
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test equipment software program name	C12305
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test mouth apparatus type	C11101
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test not done reason	C12298

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test position type	C11100
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test not done indicator	C17874
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test type	C11099
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test not done text	C17876
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Ulna length measurement	C12306
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test bronchodilator indicator	C12308
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function index of lung function type	C12311
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Pulmonary function test seat position type	C12301
Outcomes and End Points; Muscle Strength Testing	Trial number	C10171

Summary of Core/Supplemental-Highly Recommended Recommendations: Facioscapulohumeral Muscular Dystrophy CDEs

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Pulmonary Function Testing/Respiratory Status	Ulna length measure tool name	C12307

Supplemental Highly-Recommended Instruments for Facioscapulohumeral Muscular Dystrophy:

1. 6 Minute Walk Test
2. Grip Strength*
3. Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale
4. Maximum Voluntary Isometric Contraction Testing (MVICT)

*Special circumstances apply.

For the complete list of NINDS CDE recommendations for FSHD, please see the NINDS CDE website.