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FSHD-SUM (Summarized Unified Measure) PROM – should we pursue this?

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Background

- FSHD is a rare, severe, autosomal dominant progressive neuromuscular degeneration.
- A validated, quick, and responsive PRO for easy use is missing.

Methods

Purpose & Design Principles

FSHD-SUM is a concise, multi-domain Patient-Reported Outcome Measure (PROM) that combines elements from FSHD RODS, FSHD-HI2, PROMIS, SF-36, UEFI, and FDI. It is designed to assess functional capacity and perceived impact across core domains relevant to individuals with FSHD.

Development Principles

- Multidomain coverage: Mobility, upper limb function, activities of daily living (ADLs), fatigue, pain, psychosocial impact, and body image. Balanced construct orientation: Combines objective function markers with subjective perceptions of health impact.
- Patient-centered design: A concise 15-item measure using a 0–5 Likert scale ensures minimal burden and clinical interpretability.
- Crosswalk-informed development: Each item was mapped to validated instruments and the International Classification of Functioning, Disability, and Health (ICF) domains.

Ultimate goal: Suitable for clinical trials and patient monitoring

Results

Domains and Example Items

Domain	FSHD RODS Items	FSHD-HI2 Constructs	ICF Codes
Upper Limb Function	Open door, Clip nails, Button shirt, Carry tray	Lifting objects, Opening jars, Hand/shoulder weakness, Dropping objects	d445, b730, d430
Self-care	Wash/dress lower body, Take a shower, Use toilet	Bathing, Dressing, Toilet use, Sexual function	d510–d540, d620
Mobility	Sit/stand, Walk, Run, Jump, Car transfers	Walking, Balance, Falls, Running, Stairs, Tripping	d450–d455, d410
Household Activities	Clean fridge, Mop floor, Move table	Cleaning, Yard work, Scrubbing, Using hand tools	d640, d630
Endurance & Load	Carry heavy item, Walk 1km, Dance, Kick ball	Fatigue, Recovery, Tired muscles, Exercise, Dancing	b455, d570
Balance & Stability	Stand on one leg, Squat	Standing, Tripping, Core weakness	d415, b755
Recreational Activity	Dance, Dunes walk, Kick ball	Sports difficulty, Limited recreation	d920

Domain	FSHD-HI2 Items	ICF Codes
Mental Health	Depression, Anxiety, Stress	b152
Social Participation	Social avoidance, Reliance on others	d750, d910
Fatigue & Energy	Sleep, Stamina, Recovery	b455
Pain	Muscle, Joint, Back pain	b280
Cognitive Function	Memory, Concentration	b140, b144
Communication	Speaking, Swallowing	b320, b510
Body Image	Posture, Walking, Self-perception	b180

Domain	Sample Item	Response Scale
Mobility	Walk 1 km	0–5: Not at all to Unable
Mobility	Lose balance walking	0–5: Never to Always
Upper Limb	Button shirt	0–5: Not at all to Unable
Upper Limb	Lift jug to shoulder height	0–5: Easily to Not at all
Self-Care	Showering independently	0–5: Not at all to Unable
Self-Care	Getting out of bed	0–5: Easily to Not at all
Fatigue	Muscles tire quickly	0–5: Not at all to Extremely quickly
Fatigue	Recovery from light exercise	0–5: No time to Long rest
Pain	Muscle or joint pain	0–5: Never to Always
Pain	Pain interferes with activity	0–5: Not at all to Completely
Social	Avoid social situations	0–5: Never to Always
Social	Confidence managing condition	0–5: Very to Not at all
Body Image	Affected by condition	0–5: Not at all to Extremely
Mental Health	Feel anxious/frustrated	0–5: Never to Always

Remapping FSHD-SUM 15-item Structure

Domain	Item Description	Original Source(s)	ICF Mapping
Mobility	1. Walk 1 km on level ground	RODS, SF-36, PROMIS Mobility	d450 (Walking)
	2. Navigate stairs without support	FSHD-HI2, SF-36	d455 (Moving around)
	3. Maintain balance while walking	FSHD-HI2, FDI	d410 (Changing position)
Upper Limb Function	4. Button a shirt or blouse	RODS, UEFI	d440 (Fine hand use)
	5. Lift a milk jug to shoulder height	FSHD-HI2, UEFI	d430 (Lifting and carrying)
	6. Open a sealed jar	UEFI	d445 (Hand and arm use)
Self-Care / ADLs	7. Shower or bathe independently	FSHD-HI2, SF-36	d510 (Washing oneself)
	8. Dress lower body independently	RODS, FSHD-HI2	d540 (Dressing)
	9. Get in/out of bed	FSHD-HI2, FDI	d410 (Lying to sitting)
Fatigue / Endurance	10. Muscle fatigue during daily activities	PROMIS Fatigue, FSHD-HI2	b455 (Exercise tolerance)
	11. Recovery time after exertion	FSHD-HI2	b455 (Stamina)
Pain	12. Frequency of muscle or joint pain	SF-36, FSHD-HI2	b280 (Pain sensation)
	13. Pain interference with physical activities	PROMIS Pain Interference, FSHD-HI2	b280, d850 (Life management)
Psychosocial Function	14. Confidence managing your health condition	PROMIS, FSHD-HI2	b130 (Energy & drive)
Body Image & Emotion	15. Embarrassment or distress due to visible symptoms (e.g., gait)	FSHD-HI2	b180 (Body image perception)

Scoring System

The total FSHD-SUM score is computed as the mean of all 15 item responses. This composite score provides a single numeric summary of perceived disease burden.

Proposed interpretive thresholds:

- Mild impact: 0.0–1.6, Moderate impact: 1.7–3.3
- Severe impact: 3.4–5.0

These thresholds serve as provisional guidance pending psychometric validation and longitudinal outcome analysis.

REDCap Integration

A fully deployable REDCap data dictionary has been constructed for FSHD-SUM. Each item is defined as a radio field with a standardized Likert response. Automatic scoring calculations for domain and total scores and field notes summarizing item rationale and source references are included. The tool is accessible via CSV format and supports single-site and multicenter research implementation.

FSHD-SUM RASCH-built

Instructions: Please indicate how difficult each activity has been for you in the past 7 days due to your muscle condition. Use the following scale for each item.

Response Options:

- 0 – No difficulty
- 1 – Slight difficulty
- 2 – Moderate difficulty
- 3 – Severe difficulty
- 4 – Extreme difficulty
- 5 – Unable to perform

1. Walk 1 kilometer on level ground Response: _____
2. Climb a flight of stairs without using handrails Response: _____
3. Maintain balance while walking on uneven surfaces Response: _____
4. Button a shirt or blouse Response: _____
5. Lift a full milk jug to shoulder height Response: _____
6. Open a tightly sealed jar Response: _____
7. Bathe or shower without help Response: _____
8. Dress your lower body without assistance Response: _____
9. Get in and out of bed independently Response: _____
10. Perform light household chores without excessive fatigue Response: _____
11. Recover from physical activity within a reasonable time Response: _____
12. Experience muscle or joint pain Response: _____
13. Manage your daily activities despite pain Response: _____
14. Feel confident managing your condition Response: _____
15. Feel self-conscious or embarrassed due to your physical limitations Response: _____

1. Sum all responses for the 15 items to obtain the raw score (range: 0 to 75).
2. Use the Rasch conversion table to map the raw score to a Rasch-scaled interval score (0–100).
   - The Rasch score reflects the individual's relative level of functional impairment on an interval scale.
   - Higher Rasch scores indicate a more significant disability or impact.

Example:
- Raw score = 30 → Rasch score ≈ 40.0
- Raw score = 60 → Rasch score ≈ 80.0

Summary

- Remapping of existing PROs to generate a FSHD-SUM PRO
- Covering 8 functional domains
- This RASCH-Build score needs to be in-depth validated
- Correlations to the existing FSHD PROs need to be performed

References of the original sources

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PaLaDIn
Patient Lifestyle & Disease Data Interactium

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https://www.project-paladin.eu

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