

# Managing FSHD

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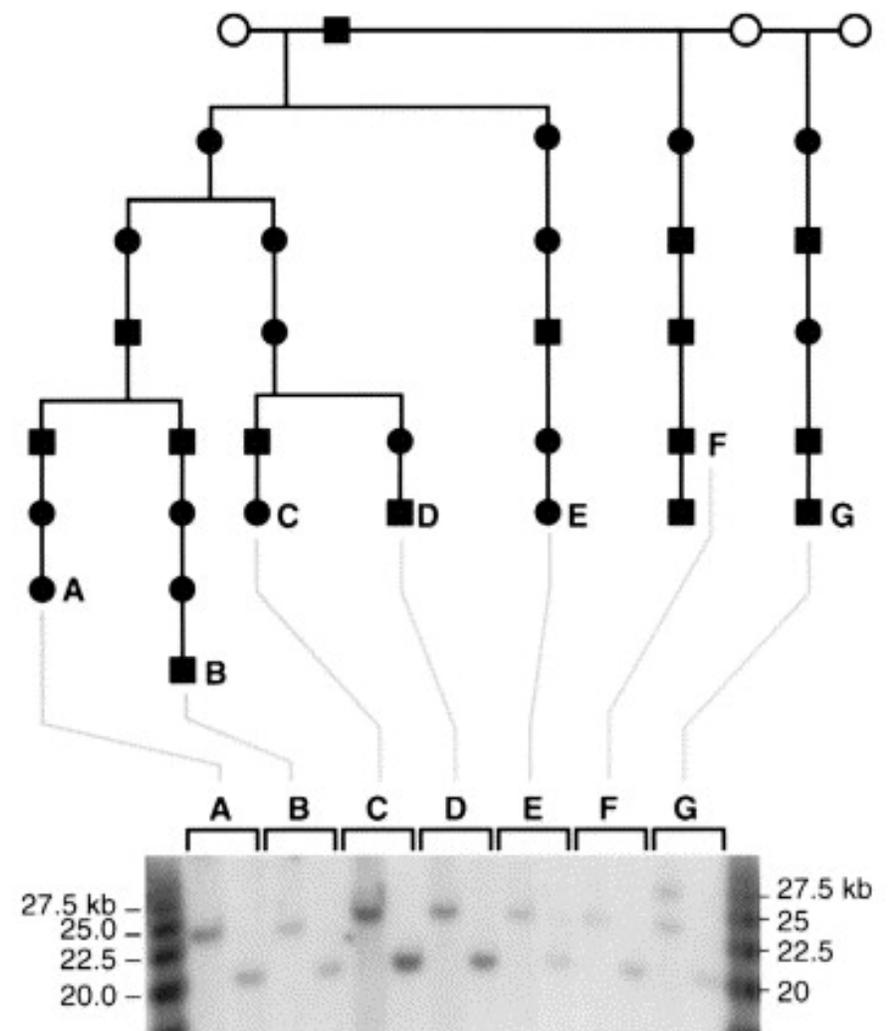
Virginia Commonwealth University

# Outline

- Diagnosis
- Clinical features
- Associated testing
- Treatment

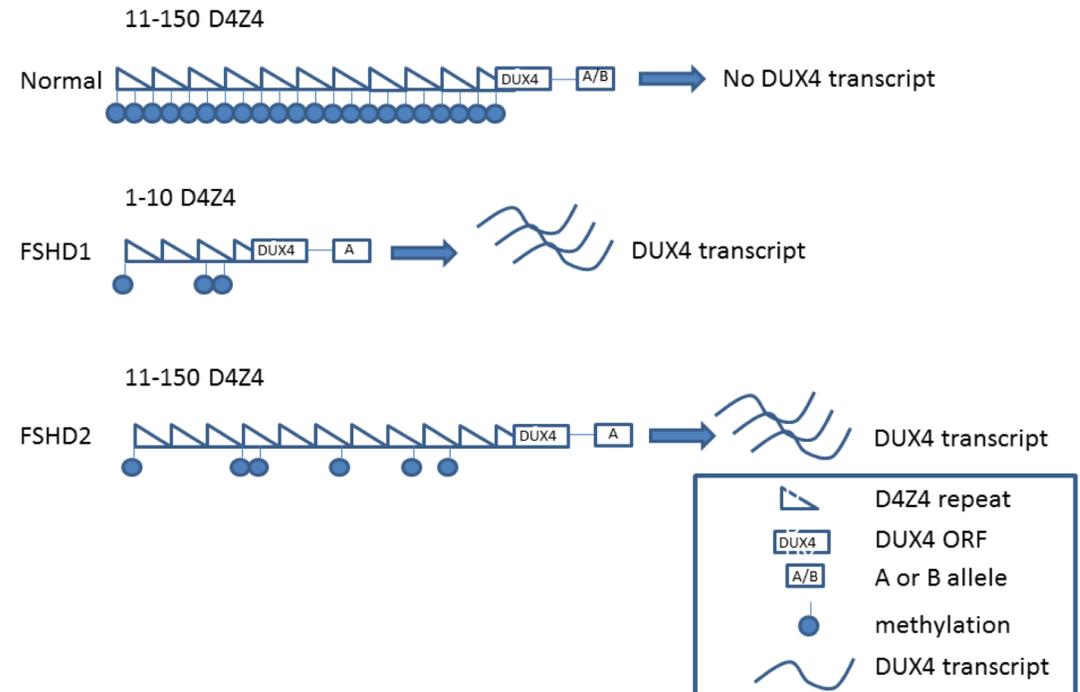
# Diagnosis of FSHD1

- Autosomal dominant condition
- Prevalence of 1:20,000
- Stable repeat length between generations



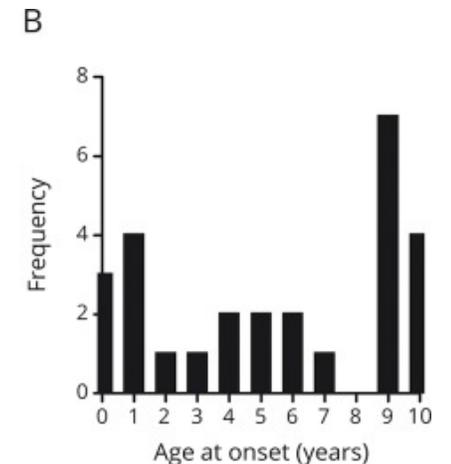
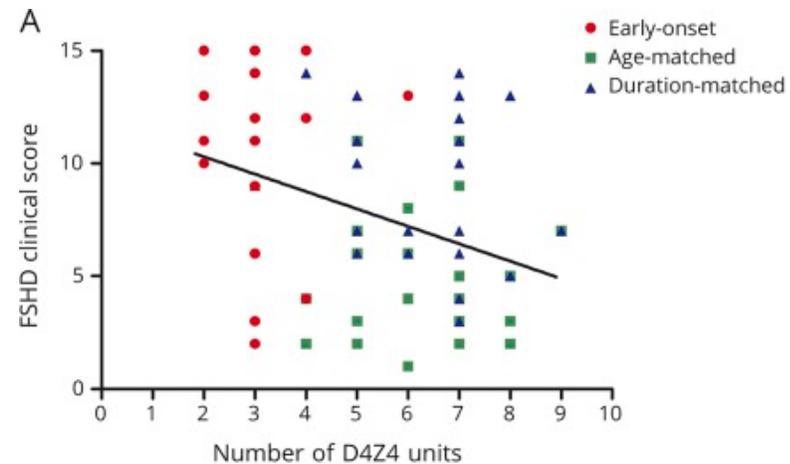
# How do you diagnose FSHD?

- Family members with clinical features
- Blood test for D4Z4 contraction
- SMCHD1 gene test
- Methylation assay



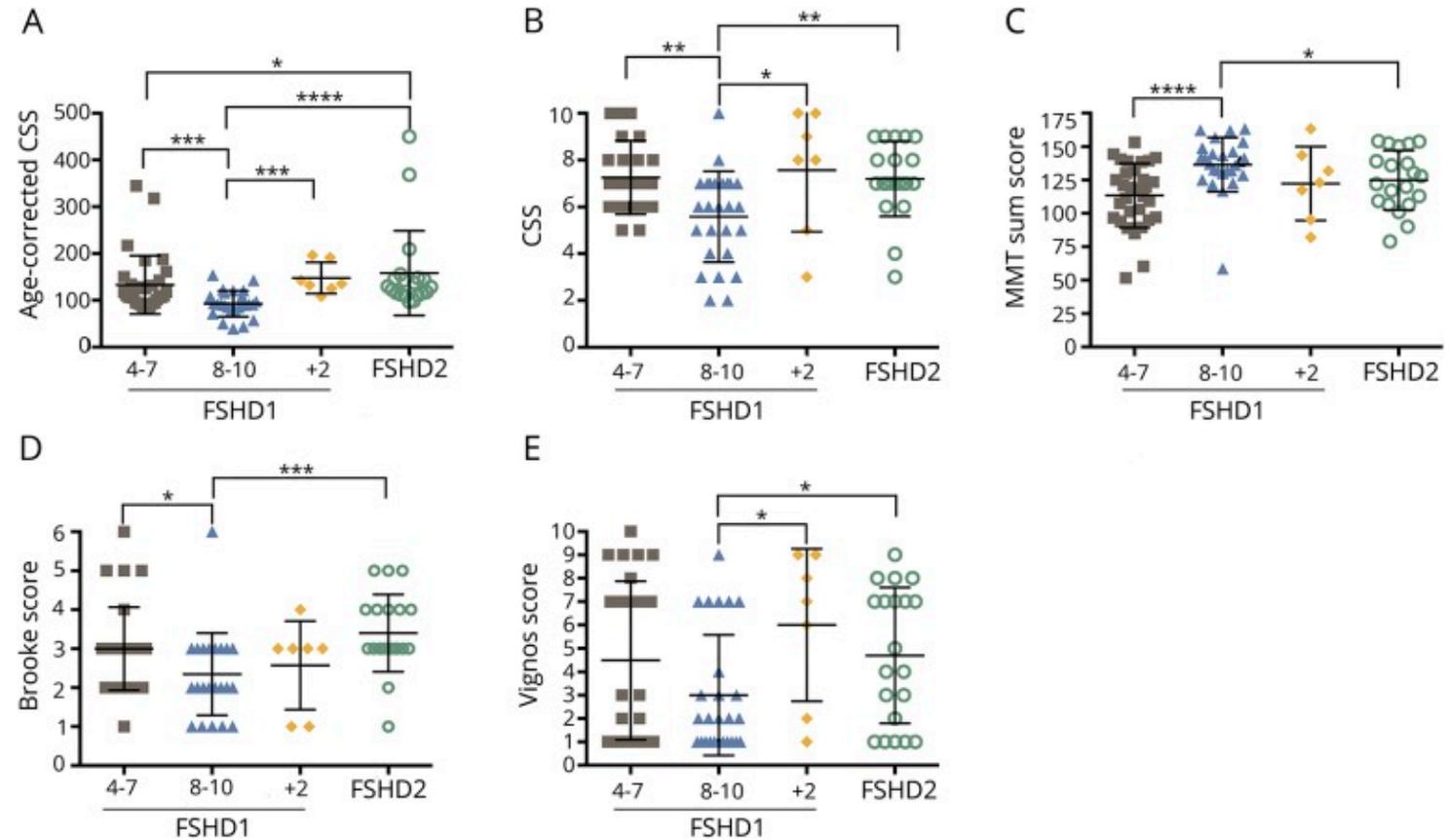
# How important is the repeat length in FSHD1?

- Likely predictive of a more severe phenotype (1-3 repeat units)
- Over 4 is less predictive
- May be affected by modifiers in methylation genes



# FSHD1 or 2: What is the difference?

- Genetically distinct
- Clinically no identified difference



# Phenotype

- Asymmetric, regional, and step-wise
- Face
- Scapular muscles
- Humeral muscles
- Eventually
  - Peroneal
  - Quadriceps
  - Abdominal muscles



# Predominantly a skeletal muscle disease

- Cross sectional survey of 328 participants with FSHD

**Table 4** Population impact score of symptomatic themes

Symptomatic themes	Population impact score <sup>a</sup>
Problems with shoulders or arms	2.59
Limitations with mobility or walking	2.49
Inability to do activities	2.36
Back, chest, or abdomen weakness	2.22
Changed body image due to disease	2.04
Fatigue	2.00
Pain	1.57
Problems with physical health	1.47
Decreased performance in social situations	1.29
Problems with hands or fingers	1.14
Decreased satisfaction in social situation	1.11
Emotional issues	0.97
Problems eating	0.48
Difficulty thinking	0.36
Communication difficulties	0.33

<sup>a</sup> Percentage of participants in whom an issue was experienced multiplied by the average life impact score of the issue.

# Other complications

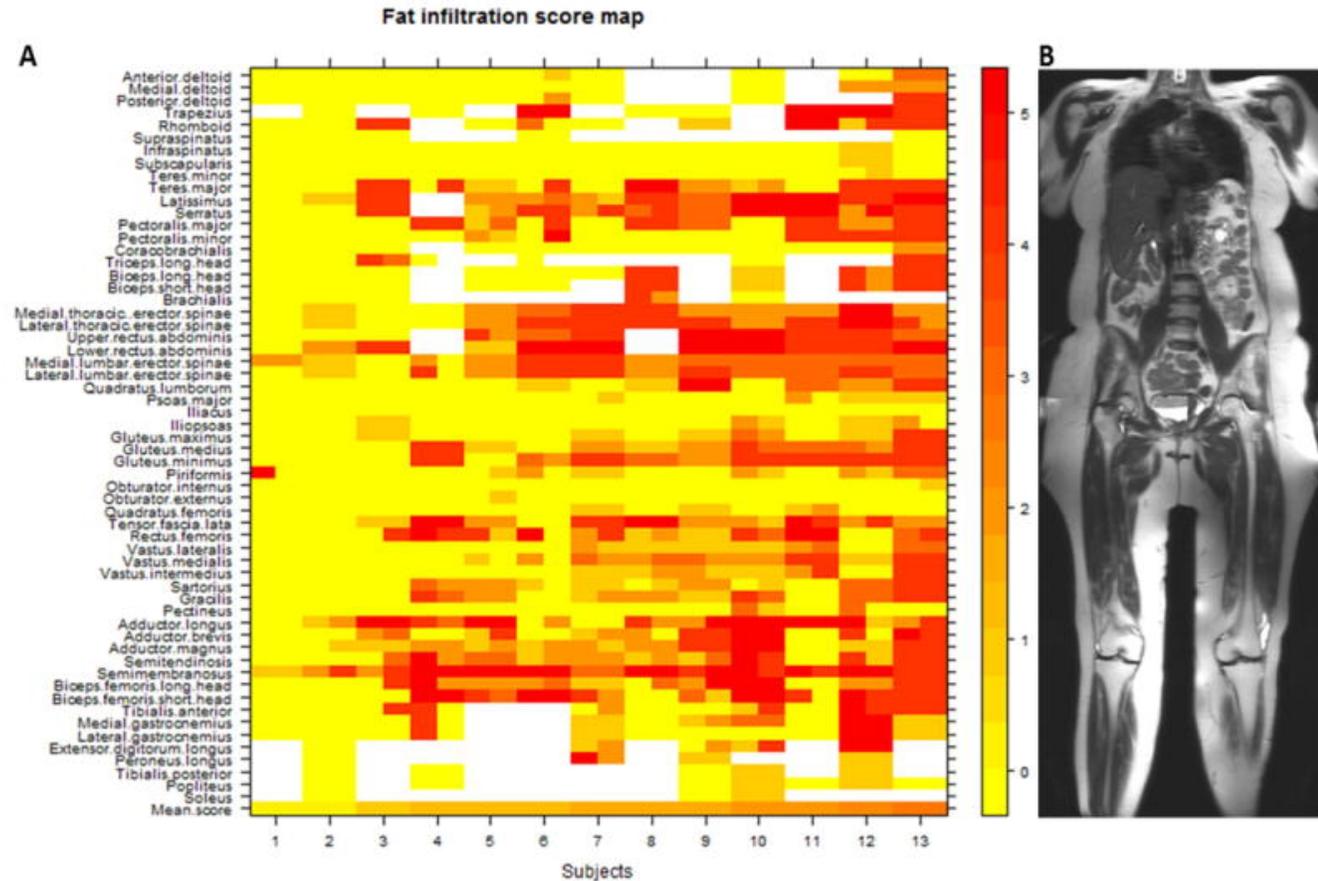
- Pain
- Hearing loss
- Less common:
  - Coat's disease
  - Respiratory failure

# Natural History of FSHD

- Slowly progressive
- Often periods of progression and plateaus

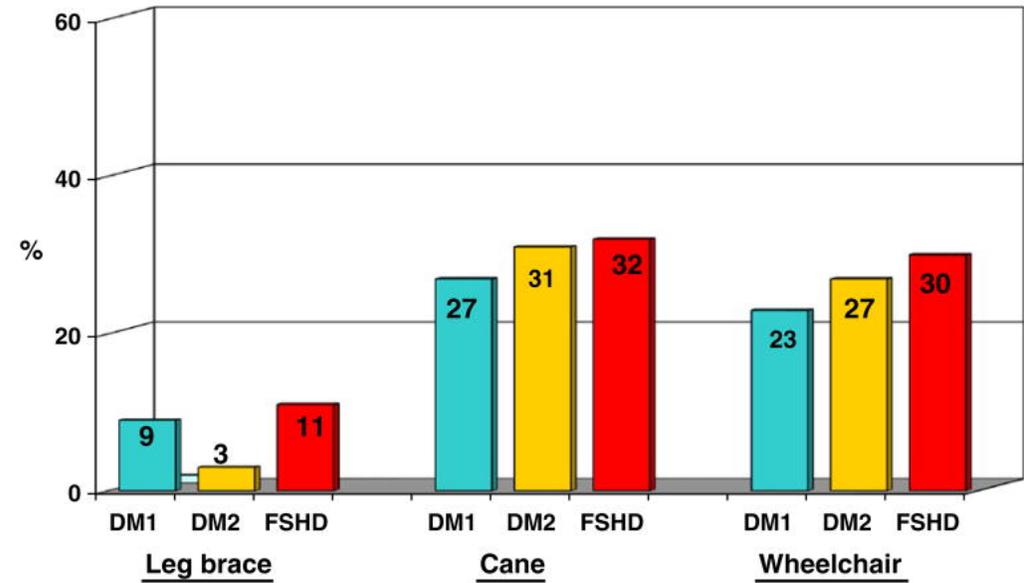
# Associated Testing

- Muscle MRI
- Muscle biopsy



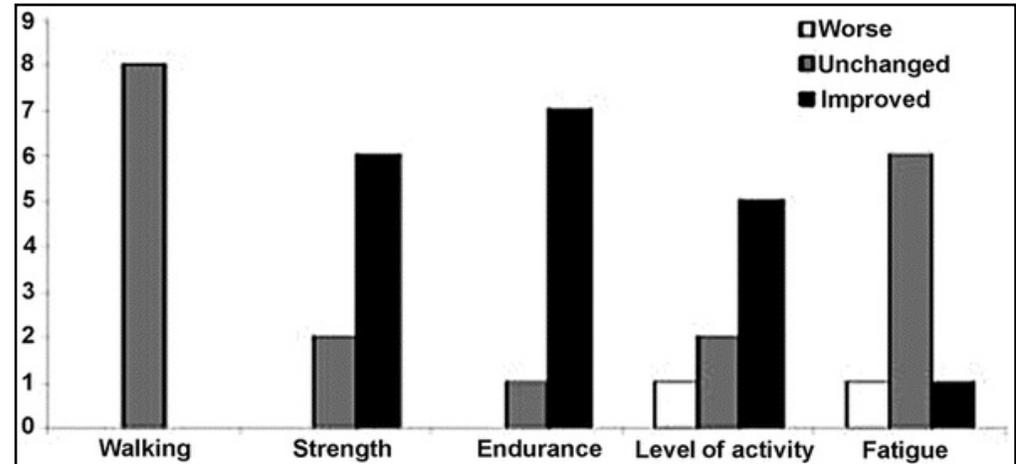
# Treatment

- Assistive devices
  - AFOs
  - Canes
  - Wheelchairs



# Exercise and FSHD

- Aerobic exercise has been shown to:
  - Improve endurance
  - Reduce fatigue
- Caution advised with weight bearing exercises



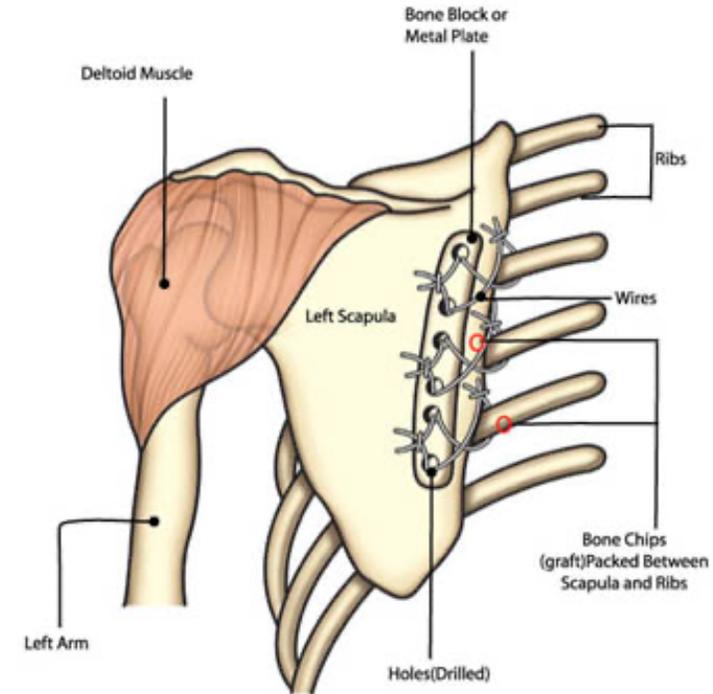
**Aerobic training improves exercise performance in facioscapulohumeral muscular dystrophy.**

Olsen, David; Orngreen, Mette; Vissing, John; MD, PhD

Neurology. 64(6):1064-1066, March 22, 2005.  
DOI: 10.1212/01.WNL.0000150584.45055.27

# Scapular fixation

- Minority of patients benefit
- May improve shoulder range of motion in appropriate patients
- Can assess benefit with manual fixation



# Medications

- No current therapy has shown benefit
- Studies have denied benefit of:
  - Albuterol
  - Corticosteroids
  - Myostatin inhibitors
- No studies support supplement use
- Treatment of pain with standard therapies, avoid opioids

# Annual monitoring

- Pulmonary function at baseline and with symptoms
- Retinal monitoring in appropriate patients
- Pain screen
- Hearing screen

# Summary

- Diagnosis by blood test and family history
- Repeat length may predict severity
- Aerobic exercise may be beneficial
- Current medications are not disease modifying