

FSHD 360

FSHD Clinical Aspects and Care

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Disclosures

- None relevant to today's talk
- Advisory board for Akcea, Sanofi Genzyme, Takeda, Alexion, and speaker fees from Sanofi Genzyme and Alexion



Objectives

- To describe the clinical features of adults with FSHD
- To discuss management and clinical care of adults with FSHD
- To touch on the care landscape for neuromuscular diseases in Toronto



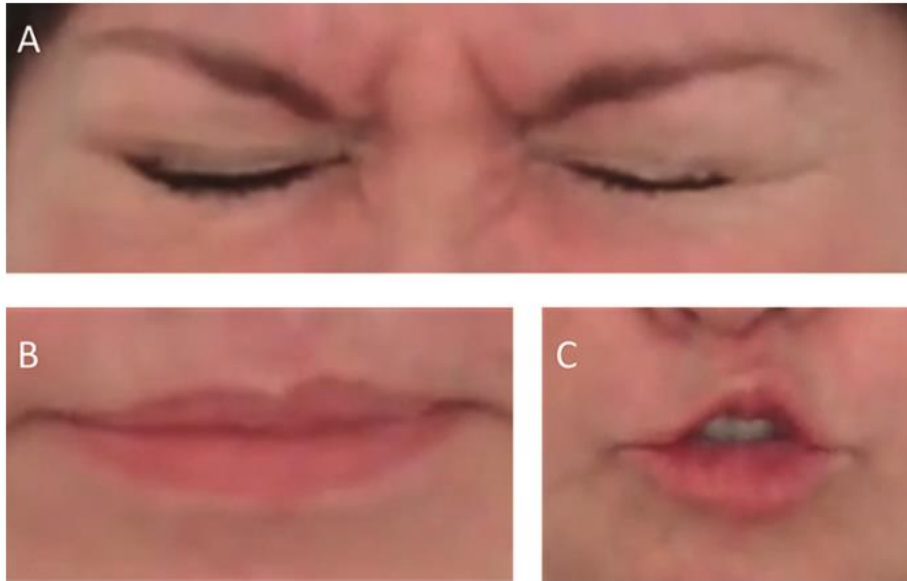
Facioscapulohumeral dystrophy (FSHD)

- One of the most common genetic muscle diseases
- ~5-12 per 100,000 persons
- Wide age range: From childhood, to early adult to late adulthood



Facioscapulohumeral dystrophy (FSHD)

- What's in a name?
- Facial weakness: Incomplete eye closure, horizontal smile
- Scapular muscle atrophy, “winged scapula”
 - Relative hump
- Humeral: humerus, triceps



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Facioscapulohumeral dystrophy (FSHD)

- What's in a name?
- Facial weakness: Incomplete eye closure, horizontal smile
- Scapular weakness: Winging, pectoral atrophy, “axillary fold”
 - Relative sparing of deltoid: Triple hump sign
- Humeral: Weakness of biceps and triceps



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What's not in the name?

- Anterior leg compartment weakness: Foot drop
- Trunk muscle weakness:
Abdominal muscle weakness preferentially involving lower abdominals
- Weakness can progress to involve other muscles over time



FSH Muscular Dystrophy

- **Muscular dystrophy**
 - Progressive disorders of the muscle
 - Muscle damage over time leads to degeneration of muscle fibers
 - Replacement of muscle by connective tissue and fat
 - Muscle biopsy/Pathology: No longer needed



FSHD – Clinical

- Presentation can be variable
- Atypical presentations
- Diagnostic delays



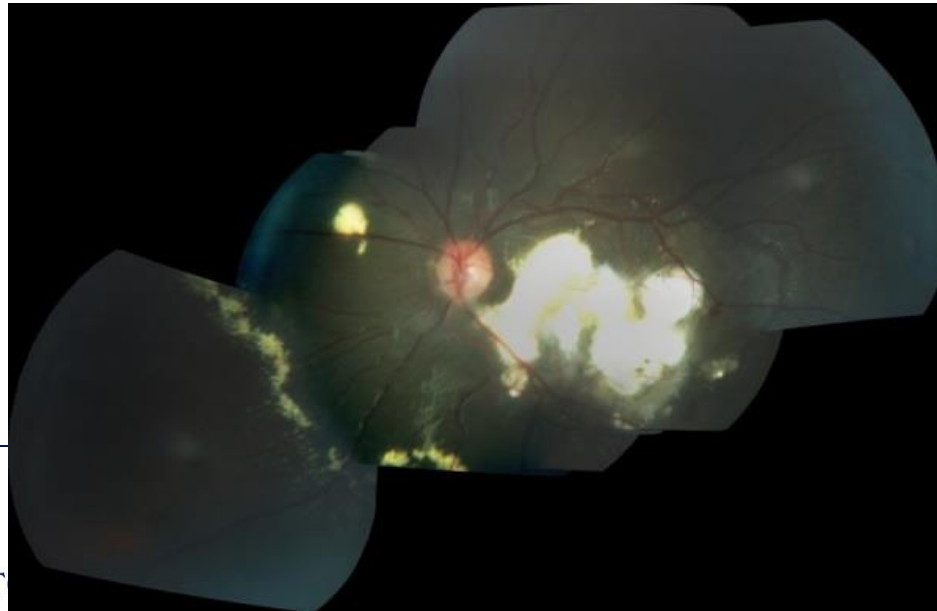
Progression

- Can be very asymmetric
- Weakness slowly progresses
- May be periods of more rapid worsening and periods of relative stabilization
- Life expectancy not reduced



“Extra-muscular manifestations”

- Retinal vasculopathy
- Hearing loss
- Pectus excavatum
- Restrictive lung disease
- Heart rarely involved



Current management and care

- Monitoring
 - Respiratory function testing
 - Ophthalmological evaluation
 - Hearing screening



Current management and care

- Exercise and Rehabilitation
 - Low to moderate aerobic training improves strength and endurance
 - May improve chronic fatigue
 - Walking, biking, swimming
 - Resistance training may help to maintain and improve function
 - Physiotherapy assessment



Current management and care

- Exercise and Rehabilitation
 - Examples:
 - 10-30 minutes of cycling 3-4 times per week
 - Resistance: Step-ups, wall-sit, squats



Current management and care

- Supportive
 - Occupational therapy
 - Braces: Ankle-foot orthosis for foot drop
 - Assistive devices



Lived experience in FSHD

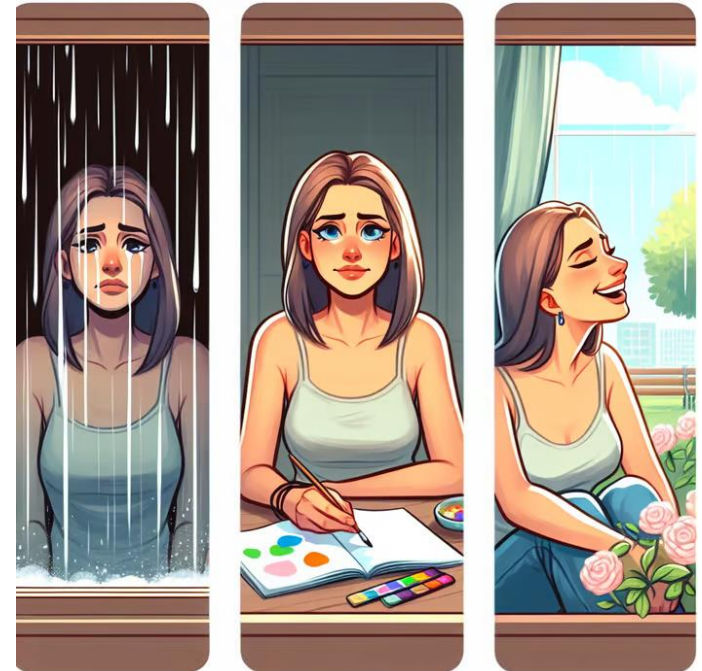


- Life as symptoms progress
 - Fatigue, facial weakness, muscle weakness
 - Active problem-solvers
 - Good QoL was possible



Lived experience article

- The emotional journey
 - Impact on mood
 - Sense of relief at having a diagnosis
 - Stress about impact



Lived experience article

- Family burden
 - Impact on family
 - Caregivers
- Social connection and disconnection
 - Non-verbal communication with facial weakness
 - Fatigue and social engagements
 - Social isolation



Lived experience article

- Visibility vs. Invisibility
 - To disclose or not
 - Stigma, bias
 - Rare condition (even among HCPs)



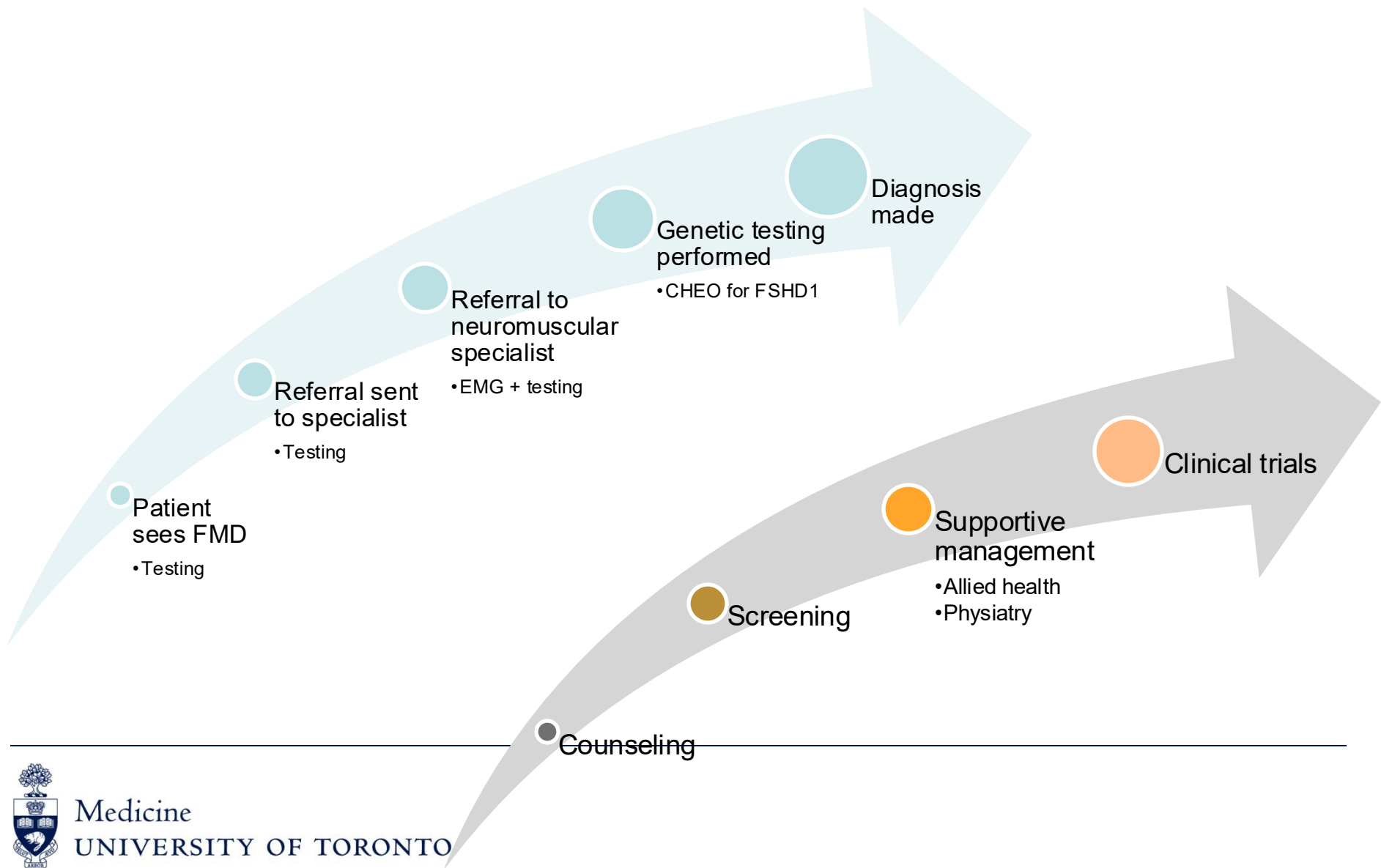
Neuromuscular care in Toronto

- Multiple academic and community neurologists and physiatrists
- Physiotherapy and occupational therapy
- Specialists as needed

- Lack of a comprehensive multidisciplinary clinic or care model



Neuromuscular care in Toronto



Take-Home Points

- FSHD has characteristic patterns of weakness
- Asymmetry and varied presentation can make diagnosis challenging
- Management includes exercise, supportive care, and appreciation of the multifaceted impact of this condition on the patients and their families



Thank you!

