SickKids The hospital for SICK CHILDREN

Pediatric FSHD

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Why the importance to differentiate early onset FSHD Reduce the gap of knowledge

Natural history is different

Important to understand Clinical outcomes in preparation on clinical trials

Infantile FSHD

Defined as

- Presence of signs or symptoms of facial weakness before 5 years of age
- Or shoulder girdle weakness before 10 years
- ▶ 4% of the total FSHD population

More severe and rapidly progressive

Systemic involvement

Early onset

- Bimodal
- <2 years</p>
- ▶ 8-10
- The FSHD score ranges from 0 (no weakness) to 15 (severe weakness).



Disease Progression

High clinical variability

- Large contractions
 - ► → Earlier onset and more severe disease with faster progression
- Disease affecting shoulder and facial muscles first,
 - ▶ The 6-year risk of wheelchair \rightarrow 24.0%
 - Peak in the second decade associated with large D4Z4 contractions,
 - Followed by an age-related increase in risk



Muscle Nerve. 2014 Apr;49(4):520-7

Individual patients functional motor scores over time



Clinical Features of Childhood-onset FSHD Dora Steel et al.

Prevalence of complications by age at onset



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Clinical Features of Childhood-onset FSHD Dora Steel et al.



Challenges on facial weakness

- Facial weakness impacts patients' daily lives and social interactions.
- Patients often realize miscommunications post-event due to facial weakness.
- There is a demand for professional support in managing facial weakness.



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Non-Invasive Treatment Options for Altered Facial Expression

1. Physical Therapy

• Used in Bell's palsy, Parkinson's; techniques like Kabat therapy improve muscle strength/symmetry

- Orofacial exercises help with speech and mouth movement (esp. in Parkinson's)
- Application in FSHD remains uncertain due to different muscle pathology

2. Acupuncture

- Evidence in FSHD is limited and anecdotal
- 3. Speech Therapy & Communication Strategies
- Training in non-verbal strategies helps manage social interactions
- Improves overall patient communication and confidence

4. Surgical Options (Smile Restoration Surgery)

- Used in Moebius syndrome: techniques like free functional muscle transfer
- FSHD: minor improvements using dermal fillers with cog threads
- Requires careful selection due to progressive muscle involvement

5. Supportive & Psychosocial Interventions

- Social skills training, peer support, educational programs
- Reduce stigma, enhance coping and quality of life

Similar but not the same FSHD vs Sarcopenia



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B. TJ Barras, H. Royden Jones, Jr., M.M. Ryan & D.C. De Vivo (Eds): Neuromuscular Disorders of Infancy, Childhood and Adolescence, Second edition.

Asymmetric involvement leading to Pain



Balanced Alignment



Data on impact of excersice in pediatric FSHD lacking

Isometric exercise



Extramuscular Manifestations

- FSHD does not affect the cardiac muscle.
 - Mild, typically asymptomatic conduction abnormalities have been reported
 - Asymptomatic right bundle branch block
 - High-frequency hearing loss
 - 0.8% of patients develop an exudative retinopathy (Coats' syndrome)
 - To be check upt ot 4-5 years old

Symptoms and Signs

- Extraocular and swallowing muscles are usually not affected.
- ▶ Restrictive lung disease \rightarrow 10%
 - ▶ 1 to 8% requiring ventilatory support
- Sleep-disordered breathing and respiratory involvement with reduced forced vital capacities in 38%
 - 14% requiring noninvasive ventilation
- Patients with severe disease
 - Weak hip flexion or wheelchair use, and kyphoscoliosis
 - \rightarrow Higher risk for restrictive lung disease (often asymptomatic)

Conclusion



Gap on data and clinical trial preparedness

Thanks



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