Facioscapulohumeral Muscular Dystrophy and Physical Therapy Implications

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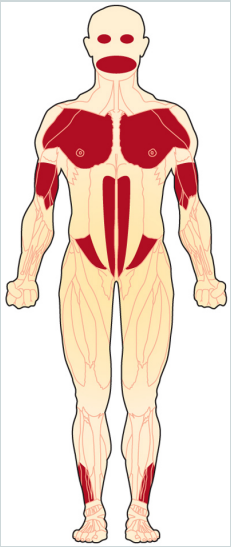
What is **F**acio**S**capulo**H**umeral Muscular **D**ystrophy (FSHD)?

- Inherited progressive neurodegenerative disease that initially primarily affects muscles in the proximal upper extremities and facial region.[5]

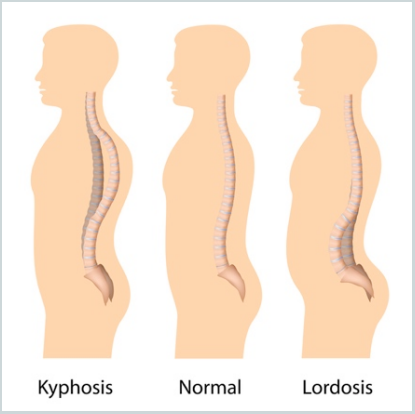


- What does it stand for?
- F = Facio = Face
- S = Scapulo = shoulder blades or Scapulae
- H = Humeral = shoulder girdle or upper arm
- D = Dystrophy = tissue of the body wastes away

- Other muscles are affected as well and this progression is different in each individual

Clinical Manifestation

- Varies from person to person
  - Scapular winging
  - Protruding abdomen and/or lumbar lordosis
  - Muscular atrophy surrounding shoulder girdle
  - Facial muscle weakness
- Primary initial complaints of:
  - Difficulty reaching overhead
  - Cervical pain
  - Difficulty climbing steps and/or standing from a chair
  - Tripping over uneven surfaces or when ambulating in community
  - Fatigue [1]



Physical Therapy: What is it?

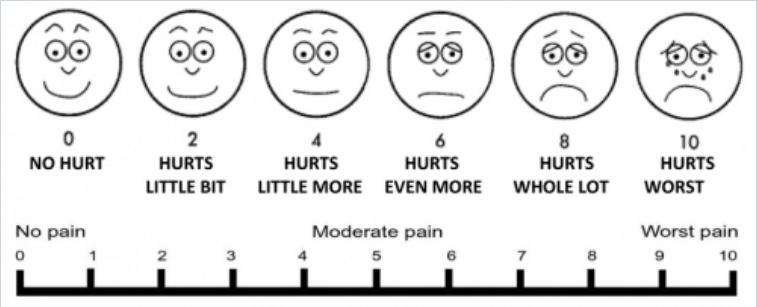
- A type of physical medicine that focuses on correcting physical impairments and disabilities.
- Treatment goals and outcomes are geared toward restoring movement in the human body as it relates to function.
- Improve efficiency of movement
- Improve quality of life

Physical Therapy and FSHD

- Not a cure
- Maintain current muscle function through an appropriate and patient specific exercise program
- Addresses pain with use of modalities, stretching and ROM program
- Evaluate need for adaptive equipment to improve overall functioning
- Improve home safety
- Educate patient and family

PT Evaluation

- Subjective Report:
  - New onset symptoms
    - Pain
    - Progressive muscle weakness
    - Falls
    - Difficulty reaching overhead
    - Difficulty transferring from various surfaces
    - Fatigue with ambulation
    - Difficulty ambulating
    - Difficulty with ADL performance



PT Evaluation

- Objective Report:
  - ROM
    - Are contractures developing?
  - MMT's
    - Assess for weakened musculature (0-5)
  - Balance:
    - Rhomberg EO / EC, SLS, Tinetti, TUG, Berg
  - Gait Analysis
    - Foot slap
    - Foot drop
    - Steppage gait
  - FIM testing

Goals and Plan of Care

- Improve joint ROM to prevent contractures and manage tone
- Decrease pain
- Improve quality of life
- Educate patient and family
- Assess need for adaptive equipment
- Avoid strength goals with your therapist and focus on function

Range of Motion

- Changes in muscle tone brought on by loss of muscle function may cause stiffness and/or pain in joints
- Makes walking and transitional movements difficult and increases risk for falls
- Stretching exercises are issued to specific muscle groups based on therapists assessment
- GOAL: Prevent contractures, maintain good postural alignment, reduce pain

PT interventions

- Restorative
- Compensatory
- Preventative

Bed Mobility

- May have difficulty rolling or getting in and out of bed

Transfers

- Difficulty transitioning from sitting to standing
  - Raise surface height
    - Lift chairs, power w/c, raised toilet seat, grab bars in bathroom

Gait Training

- Goal is to normalize gait pattern
- Noted abnormalities:[3]
  - Lack of adequate toe clearance (foot drop)
  - Excessive hip and/or knee flexion (steppage gait)
  - Lack of heel strike on initial contact
- Assistive devices:
  - Standard cane
  - Rolling walker
  - Platform rolling walker
  - 4 wheeled RW vs. 3 wheeled
  - AFOs [2,3]
  - LSO

Is it safe for me to walk?

- Take into account strength assessment, balance screening, pt and family testimony, fall history.
- Buckling of knees or tripping, excessive back pain?
- Foot drop
- Poor trunk control
- Decreased endurance

Power Mobility Options

- Coordinate with local Neurologist, PT and power mobility company to decide which wheelchair would be best for you and which options are appropriate.
- Goal is INDEPENDENCE

Home Safety

- Grab bars in bathrooms (make sure there is 1.5” between wall and handrail)
- Lift all throw rugs
- Offset hinges to widen doorways or remove doors
- Clear pathways
- Make sure doorways are at least 32” wide
- Replacing doorknobs with lever handles
- Tub bench
- Raised Commode

Energy Conservation: Why is it so important?

- Fatigue occurs due to decreased muscle strength
- Working too hard and too fast can result in overcompensation, when muscles with normal strength take over for weaker muscles.
- Treatment for fatigue is energy conservation:
  - Balance rest with activity
  - Do not push through fatigue
  - Accept help
  - Establish a regular sleeping pattern
  - Use adaptive equipment such as a tub bench, raised commode, etc.
  - Plan ahead
  - Avoid stressful situations
  - Paced breathing and diaphragmatic breathing
- Monitor your patterns and fatigue levels

Research

- 1. Voet N, Bleijenberg, G, DeGroot I et al. **Both Aerobic exercise training and cognitive behaviour therapy reduce chronic fatigue in patients with facioscapulohumeral muscular dystrophy: A randomized controlled trial** *Ann Phys Rehabil Med*. 2014;57(1):e96
  - Objective: investigate effect of aerobic exercise training (AET) and cognitive-behavioral therapy (CBT) on chronic fatigue in patients with FSHD.
  - Methods: 57 patients with FSHD type 1 with reported severe chronic fatigue and divided into 3 groups, AET, CBT, usual care. Assessed outcomes before treatment, after 16 weeks of intervention and after a 12 week follow up
  - Results: Both AET and CBT group had reported less fatigue
  - Conclusion: AET and CBT can reduce subjective complaints of chronic fatigue in patients with FSHD

Research

- 2. Bankole, L, Millet G, Temesi J. et al. **Safety and efficacy of a 6 month home-based exercise program in patients with facioscapulohumeral muscular dystrophy: A randomized controlled trial** *Medicine(Baltimore)*. 2016;95(31): e4497
  - Objective: to invesitgate the safety and efficacy of a longer exercise program on fitness, muscle and motor function in patients with FSHD
  - Methods:16 FSHD patients randomly assigned to training group (TG) or control group (CG). TG = cycling for 35 minutes 3x a week for 24 weeks with combination of strength, high-intensity interval and low intensity aerobic.
  - Results: Significant improvements were seen in VO2 peak (muscle was more efficient with oxygen uptake), MAP (oxygen exchange). Muscle endurance, MVC and 6 minute-walk distance. Dystrophic pathophysiolgic patterns were not exacerbated.
  - Conclusion: Patients with FSHD can safely perform combined training without compromise to muscle tissue in order to attain functional gains.

Benefits of an exercise program

- Will help you stay active
- Keep blood moving through muscles to bring nutrients to muscles and lubricate joints
- Boost energy levels
- Remove damaged proteins
- Reduce inflammation
- Strengthen neuromuscular junctions
- Boosts neurotrophins (substances that assist with protecting motor neurons in the brain)
- More beneficial than harmful

Beneficial exercise

- Individualized
- Initial rehab consultation is important
- Aerobic exercise [1,3,6] (15-30 min 3x)
- Balance training with and without orthotics

Common Myths

- “PT can’t help me”
- “Once I get a power device I will never walk again”
- “These braces will just make my muscles atrophy faster”
- “I was told that if I exercise I will damage my muscles more”

Educating your clinician

- Work together to set goals
- Ensure clinician knows your daily routine, responsibilities, lifestyle
- Be open and honest about how you are responding to regimen

Thank

References

- 1. N.Voet, Bleijenberg, G., DeGroot I et al. Both Aerobic exercise training and cognitive behaviour therapy reduce chronic fatigue in patients with facioscapulohumeral muscular dystrophy: A randomized controlled trials *Ann Phys Rehabil Med*. 2014;57(1):e96
- 2. Aprile, I. Bordieri C. , Gilardi, A. et al. Balance and walking involvement in facioscapulohumeral dystrophy: a pilot study on the effects of custom lower limb orthoses. *Eur J Phys Rehab Med. 2015;49(2):169-78*
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- 5. Pandya S, King WM, Tawil R. Facioscapulohumeral dystrophy. *Phys Ther*. 2008;88(1):105-13.
- 6. Voet, N., Van der Kool E., Riphagen I. et al. Strength training and aerobic exercise training for muscle disease. *Cochrane Database Syst Rev*. 2013; Jul 9;(7):CD003907.