A PHYSIATRIC APPROACH TO PATIENTS WITH FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY

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WHAT IS A PHYSIATRIST AND WHAT DO WE DO?

• Physical Medicine and Rehabilitation physicians (PMR)
• Physiatrist complete 4 years of medical school and a 4 year residency program
• Specialty of medicine focusing on the physical and functional manifestations of a person affected by a physical or cognitive impairment
GOALS OF PHYSIATRIC MEDICINE

• Maximize an individuals function
• Maintain a persons independence
• Prevent secondary medical comorbidities and complications
• Prevent or limit physical deformities
WHY SHOULD A PHYSIATRIST BE INVOLVED IN THE CARE OF SOMEONE WITH FSHD AND WHEN SHOULD A PATIENT BE REFERRED?

• Usually referred through a primary care physician or a neurologist

• Referral should be made early in the diagnosis (pediatric physiatrist when onset before 18)

• Important to focus on maintaining function and endurance and preventing secondary medical and musculoskeletal complications
THE PHYSIATRIC EVALUATION: WHAT TO EXPECT?

• Complete medical and functional history

• Functional history:
  – Level of assistance needed for activities of daily living (ADL’s) such as dressing, bathing, toileting, and feeding. Complex ADL’s (CADL) such as cleaning, cooking, community activities, driving or working
  – Level of assistance needed for mobility-Use of assistive devices
  – Frequency of falls
  – Use of orthotics
THE PHYSIATRIC EVALUATION CONT.

• Review of systems:
  - Level of endurance
  - Pain
  - Nutrition/swallowing difficult
  - Speech/articulation
  - Breathing/shortness of breath with activities
  - Mood /adjustment to the disability

• Physical exam:
  - Special focus on musculoskeletal and neurologic systems:
    ▪ UE and LE and trunk strength
    ▪ ROM
    ▪ Cranial nerves/Facial weakness
    ▪ Speech
    ▪ Gait[Pattern and stability]
THE PHYSIATRIC EXAM

Upon completion of the exam recommendations may include:

- Exercise prescription/recommendations
- Referral to PT, OT, and or speech therapy
- Referral to an Orthotist for bracing
- Recommendation to use an assistive device for mobility (cane, walker)
- Referral for a mobility device (wheelchair)
- Recommendations for treatment of pain

All recommendations are *individualized* to the patients functional and medical status and personal goals.
AN EXERCISE PRESCRIPTION FOR A PATIENT WITH FSHD

• Common questions asked:
  – Strength training VS. Aerobic training
  – Supervised (PT, OT personal trainer) or unsupervised
  – Will exercise make me stronger?
  – Will exercise make me worse or weaker?
LITERATURE REVIEW ON EXERCISE AND FSHD

• Paucity of literature looking at FSHD and exercise
• Cochrane database review (2013), reviewed only 2 studies pertaining to FSHD and strength training
  - Authors conclusion: “Moderate intensity strength training in patients with FSHD 1 (2 studies) appears to do no harm but there is insufficient evidence to conclude they offer benefit”
Anderson et al. (2015) investigated the effects of regular aerobic training (36 sessions, 30 min cycle ergometer) followed by ingesting a protein carbohydrate drink or placebo beverage.

- Results showed that all participants improved fitness, walking speed and self-assessed health and that the post-exercise protein drink did not add further benefit.
AEROBIC EXERCISE AND FSHD CONT.

- A 12 week study by Olsen et al. (2015) looked at low intensity aerobic exercise (cycle ergonometric at HR corresponding to work intensity of 65% VO2 max (a measure of cardiac fitness pertaining to the amount of oxygen your body is capable of utilizing in 1 minute) at 35 minute weekly sessions and increased to 5 times week in 4 weeks.
  - After 12 weeks, participants showed improved maximum oxygen uptake and work load (exercise performance) with no signs of muscle damage (measured blood plasma CK which is a marker for muscle breakdown)
• Bankole et al. (2016) evaluated the safety and efficiency of a 6 month home based exercise program (unsupervised) in 16 patients with FSHD
  – Patients were randomized to a control group with no HEP and a training group (cycle 3x a week for 35 minutes) for 24 weeks followed by the control group doing the HEP for 24 weeks
  – Found improvement in endurance, 6 minute walking test, and subjective improvement in fatigue with no detrimental effect on muscle tissue
A recent study by Anderson et al. (2017) looked at high intensity training (HIT) and patients with FSHD
- Number of participants: 13
- Patients with FSHD: 1
- HIT group (6 participants) underwent 8 weeks of supervised HIT (10 minute cycle ergometer, 3 times a week)
- Control group underwent 8 weeks of usual care (not well defined)
- Followed by all participants performed 8 weeks of unsupervised HIT
- Supervised and unsupervised HIT participants improved fitness (VO2 max)
- No training effect on muscle strength, 6 minute walk, 5 time sit to stand tests
- Plasma CK (evidence of muscle breakdown) and pain scales were unaffected
CONCLUSION

• More studies need to be done looking at role of exercise in patients with FSHD

• No evidence to suggest strength training is beneficial, however it does not appear to be harmful

• Moderate aerobic exercise (and possibly HIT) may be beneficial in improving fitness and overall well being

• Any exercise program should be individualized due to the heterogeneity of FSHD (patient have different muscle involvement and rates of disease progression)

• When initiating an exercise plan, it should be initially be under the supervision of a professional.
• Physical therapist will work on maximizing strength, gait, balance and ROM to maximize mobility

• May recommend an assistive device to aid in mobility/prevent falls

• Develop an **individualized goal oriented** home exercise program (HEP)-extremely important to follow-through to maintain gains

• Aqua therapy
  – No controlled studies looking at water therapy and FSHD
  – Theoretical Benefits
    ▪ Buoyance of water acts to assist mobility
    ▪ Many muscles can be worked simultaneously
The most common initial finding in patients with FSHD is weakness of the scapular stabilizers, making it difficult to performing reaching and overhead activities.

OT’s work on improving upper extremity mobility to aid with performing activities of daily living as well as community and work-related activities.

Also work on ROM to prevent contractures.

May recommend equipment to assist with ADL’s (reachers, sock aids).

May recommend bracing to improve function, prevent contractures.

Home Evaluation (PT and/or OT)
- Evaluate home environment to maximize safety and independence
- Need for/placement of grab bars
- Look at kitchen/bathroom set-up to make items more accessible
- Look at home obstacles (rugs, furniture) that may effect mobility
• Due to facial weakness, patients may have difficulty with articulation, and more rarely swallowing

• Speech therapist aid in maximizing communication and articulation

• Will evaluate swallowing via a bedside swallow(in the clinic) or with a Videoflouroscopic swallow study(more sensitive, performed in radiology dept.)
  – If indicated will provide strategies to maximize swallow ability and safety
  – May recommend diet changes to prevent aspiration.
ORTHOTICS AND FSHD

• May be recommended by your M.D. or therapist

• Often referred to a professional Orthotist to customize braces

• Common orthotics in FSHD:
  - AFO: Ankle foot orthosis
    ▪ Crosses the ankles and foot
    ▪ Used for foot drop due to peroneal muscle weakness
    ▪ Allows/aids in walking
    ▪ Prevents patient from catching the toes with walking
      ❖ Prevents falls
      ❖ Normalizes gait pattern
      ❖ Stabilizes knee to prevent hyperextension
      ❖ Prevent contractures
ORTHOTICS AND FSHD

• KAFO- Knee ankle foot orthosis
  – Crosses the knee joint
  – Used when quadriceps (thigh muscle) is compromised/weak and more support is needed to prevent knee from collapsing
    ▪ Stabilizes gait to prevent falls

Due to weight of this orthotics often not tolerated especially if there is substantial hip girdle weakness
ORTHOTICS AND FSHD CONT.

• Abdominal binders
  - Due to weak core muscles often have hyperlordosis of the spine which can lead to pain
  - Used to aid weak core to stabilize back

• Scapular Bracing
  - Sometimes used to stabilize scapular muscles
  - Often not tolerated or effective
ASSISTIVE DEVICES AND FSHD

• Common assistive devices
  – Cane (straight, quad), walker
  – Equipment for ADL’s (reachers, sock aids, raised toilet seat, etc.)
MOBILITY DEVICES AND FSHD

• 20% of patients will require a wheelchair for mobility after age 50

• Transport chair
  – Early in the disease, often used for longer community mobility, when patients are still walking but fatigue is an issue

• Power wheelchair
  – Due to upper extremity weakness, often difficult to use a manual wheelchair, power chairs are most commonly recommended
  – Referral should be made to specialized seating clinic
  – All wheelchairs should be customized to the individual
  – Support back, maintain postural alignment, minimize postural deformity
• Limited literature on this topic
• Up to 80% of individuals suffer from acute or chronic pain
• Most common areas are the neck, shoulders, low back and legs

• Reasons for Pain
  – Muscle imbalance between opposing muscle groups, leads to a sustain stretch of some muscles and prolong contractures of others resulting in inflammation
  – Weak muscles around a joint can not protect the joint, resulting in improper joint alignment and pain
  – Weakness of the shoulder muscles can result in stretch injury to the brachial plexus causing pain and weakness down the arm
  – Weak abdominal muscles can not protect the low back resulting in low back pain
TREATMENT OF PAIN

• Bracing for stability

• Modalities (Heat/Ice)

• Pain medication
  – NSAID’s, Acetaminophen, topical analgesic, nerve membrane stabilizers (lyrica, gabapentin)

• Rest if pain persists
  – Pain is the body informing us to stop doing an aggravating activity
CONCLUSION

• Integrated team approach to care
• Exercise can prevent fatigue and improve fitness
• Use of equipment/AD to maximize mobility and function
• The ultimate goal is to improve or maintain a person's independence, safety and

Disability