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 individual's function
• Maintain a person's 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 ergonometer) 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 ergonometer 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 over all 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 THERAPY AND FSHD

• 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.
SPEECH THERAPY AND FSHD

• 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
PAIN AND FSHD

• 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