Shirley Ryan Abilitylab®

A PHYSIATRIC APPROACH TO PATIENTS WITH FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY

SUSAN KEESHIN M.D.
MEDICAL DIRECTOR DAY REHAB
THE SHIRLEY RYAN ABILITY LAB (FORMERLY KNOWN AS RIC)

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"

AEROBIC EXERCISE AND FSHD

- 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)

AEROBIC EXERCISE AND FSHD CONT.

- 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

OCCUPATIONAL THERAPY AND FSHD

- 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

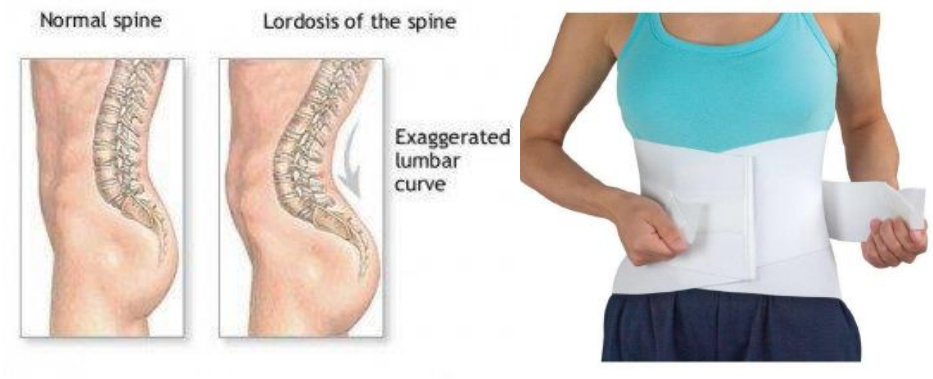
ORTHOTICS AND FSHD

- 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

Shirley Ryan Abilitylab

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 persons **independence**, **safety** and

Shirley Ryan Abilitylab