Living with FSHD

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FSH SOCIETY
FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY

Kennedy Krieger Institute

- Activity
- Nutrition
- Bone Health
- Hormone replacement
- Other issues

Activity

- A few studies suggest benefits of exercise
- Bankole et al., Medicine 2016
  - Cycling 35 minutes 3 times a week for 24 weeks
  - Increased in peak oxygen uptake
  - Increased 6-minute walk distance
  - Increased strength of quadriceps
  - Decreased fatigue

Activity

- Andersen et al., Neurology 2015
- Cycling 30 minute, three times weekly for 12 weeks
- Increase in fitness

and walking speed

Activity

- Voet et al., Neurology 2014
- Both aerobic exercise and cognitive-behavioral therapy reduced chronic fatigue
- Cycling 30 minutes, 3 times a week for 16 weeks
- Goal 50-65% of heart rate reserve (HRR)
  - HRR=maximal heart rate – resting heart rate
  - Maximal heart rate is 220-age
  - For a 60 year old with a resting heart rate of 75
    - (220-60)-75=85
  - 50% of HRR: 85 x 0.5=42.5 + 75(resting HR)=117.5
  - 65% of HRR: 85 x 0.65=55.25 + 75 (resting HR) =130.25
  - Therefore goal would be HR 117 to 130 bpm

Dieting

- Rapid weight loss can rob from muscle as well as fat
- Center for Disease Control and Prevention
  - No more than 3,500-7,000 calorie deficit a week
  - 4-8 pounds a month
- In FSHD
  - May not be able to burn calories
  - A sedentary woman of 50 burns on average 1,600 calories a day
  - Can't trim 500 calories a day without being nutritionally deficient
  - A safer goal would be to trim 175 to 250 calories a day
    - 1.5-2 pounds a month

Nutrition

- Protein
  - Meat/Fish
  - Beans/Nuts/dairy/eggs
  - To determine amount multiply weight in pounds by 0.36
  - E.g. 160 lb person would need 160 X 0.36= 58 grams
  - E.g. A cup of yogurt for breakfast, a peanut butter and jelly sandwich for lunch, nuts for snack and a salmon for dinner (60 grams)

Nutrition

- Calcium
  - Dairy
  - Fortified products (orange juice, cereal)
  - Vegetables
  - TUMS (500-1000mg)
  - RDA 1 -1.2 g a day

Nutrition

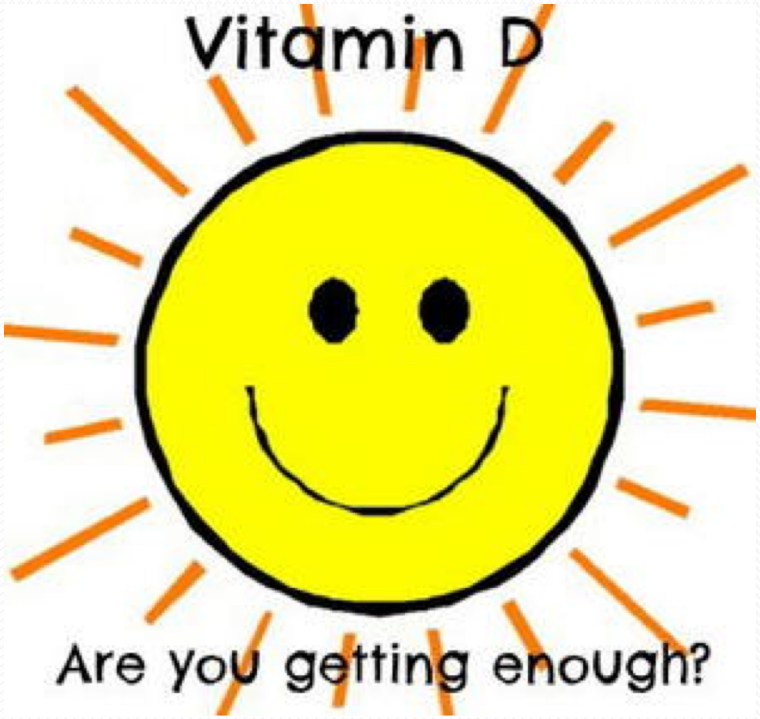
- Coenzyme Q10
  - Bioenergetics: Electron carrier intermediate in mitochondrial respiratory chain
  - Potent antioxidant
  - Multiple studies in trained athletes: enhancing power and decreasing fatigue
  - Dose 300 mg a day

Vitamin D

- Needed for calcium absorption
- Requires direct exposure to sun without sunblock
  - 10-15 minutes in summertime
- Fatty fish (salmon, tuna)
- Eggs
- Fortified products

(orange juice, cereal)

- OTC supplements



Bone Health in FSHD

- Chagarlamudi et al., Muscle Nerve 2017
- 100 individuals with FSHD
- Nearly 1/3 of subjects were deficient in VitD3
- 36% had a history of fracture
- Bone mineral density was associated with strength and function

Lifestyle

Physical activity "Use it or lose it"

- Weight-bearing activities and mechanical stress promote ↑ bone mineral density
- Athletes in high-impact sports had greater bone density than those involved in low-impact sports

- Prolonged bed rest and immobilization
- Reduction of mechanical stress

→inhibits osteoblast bone formation
→accelerates osteoclast resorption

Results in bone loss

Who is likely to have a fracture?

- Age > 65 both men and women
- Postmenopausal women: sharp decline in estrogen
- Ethnicity: Caucasian and Asian

- Family history of fracture
- Previous history of fracture
- Low body weight <127 pounds
- Smoking
- Alcohol >3 /day
- Certain drugs (steroids)

Bone Mineral Density: Best predictor for fracture

*DEXA

- Imaging technique of choice for measuring BMD
- Easy
- Minimal radiation

Two X-ray beams are aimed at the bones
Soft tissue is subtracted out

➢ BMD test: determines whether you have osteoporosis

*DEXA = *Dual-energy x-ray absorptiometry*

Results of the DEXA scan

- Normal
- Osteopenia
- **Osteoporosis**

Values of Bone Mineral Density are in the form of T and Z-scores.
"The Z-score at the left hip -3.8 …"

- The T-score compares the patients' BMD to the average for young adults at the time of peak bone mass
- The Z-score compares the BMD to persons of the same age.

Treatment for low BMD: Bisphosphonates

- alendronate (Fosamax) oral weekly
- risedronate (Actonel) oral daily, weekly or monthly
- ibandronate (Boniva) oral
- zolendronic acid (Reclast) IV once per year
- pamidronate (Aredia) every 3 months

- Oral agents are easier to take
- Must be upright for at least 30 minutes to reduce the risk of esophagitis
- Intravenous agents taken once every several months
- Flu like reaction with first intravenous infusion

Hormones

- Estrogen
  - Protective effect on bone
  - Prevents osteoclast formation and shortens the life span so there is less resorption
  - With menopause, there is a decline in estrogen levels which leads to bone loss in women

- Testosterone
  - Low levels are associated with accelerated bone turnover and increased fracture risk
  - Inhibits bone resorption and maintains bone mass

Testosterone replacement

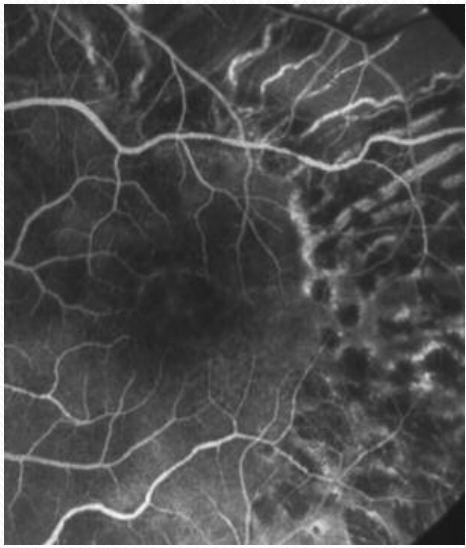
- Levels should be checked in the morning
- Replacement of low testosterone
  - Increase energy
  - Increase libido
  - Increased bone density
  - Increase muscle mass and possibly strength
- Risks of testosterone replacement
  - Blood clots
  - Increased risk of heart attack and stroke
  - Growth of prostate/testicular cancer

Study of testosterone and rHGH in FSHD (STARFISH)

- University of Rochester
- Open label study of daily human growth hormone (Genotropin 5.0 ug/kg sq) and testosterone (testosterone enanthante 140 mg IM every 2 weeks)

Ophthalmologic Care

- Association of FSHD with retinal vascular disease
- Similar to Coat's syndrome
- Recommendation: Biannual retinal exam by ophthalmologist
- Fluorescein angiography if abnormal retinal exam or family history of Coat's syndrome in FSHD
- Prevention of corneal abrasions
  - Lacrilube at night, artificial tears



Hearing Loss

- Two recent studies showed no increased incidence of hearing loss in typical FSHD
- However, high frequency hearing loss associated with infantile onset FSHD
- Recommendation: Hearing test for children with FSHD

Scapular fixation

- Anticipated benefits: reduced pain, improved arm abduction/extension, less “winging”
- Risks: surgical risks, infection, pain, decreased pulmonary function, long rehab
- No randomized controlled trials
- Important to have a surgeon familiar

with FSHD and the procedure

Pain

- Wood et al., Muscle Nerve 2018
  - 88.6% reported current pain
  - 30.4% reported chronic severe pain
  - Most frequently shoulders and low back
- Medications
  - Nonsteroidal antiinflammatory (NSAIDS)
    - Meloxicam
  - Cymbalta (duloxetine)
- Physical Therapy
- Abdominal binder
- Scapulofixation

Fatigue

- Kalkman et al., J Neurol Neursurg Psych, 2005
  - 61% reported severe fatigue
- Aerobic exercise
- Cognitive behavioral therapy
- Testosterone replacement
- Stimulants

Questions?

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