NURS 821 Alterations in the Musculoskeletal System

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Lecture 12

Part 4 Muscular Disorders

Muscular Dystrophy

- Definition: Most prevalent group of childhood muscle disorders; group of familial disorders leading to degeneration of skeletal muscle fibers
  - Characterized by progressive, symmetric skeletal muscle weakness and wasting
- Classification: Based on age at onset, progression rate, distribution of muscular involvement; inheritance patterns

MD

- **Etiology**: genetic defect, possible separate biochemical defect
- **Pathology**: genetic abnormality in intra-muscular fiber metabolism resulting in inability to absorb or metabolize a substance critical to muscular function; defect in creatinine metabolism and decreased levels of intra-muscular enzymes

Major Differences in Muscular Dystrophy Syndromes

Disease	Mode of inheritance	Age at clinical onset	Usual distribution	Rate of progression	Mental retardation
Duchenne dystrophy (DMD)	X-linked recessive	About 3 years	Hips and shoulders, quadriceps femoris, gastrocenemius (pseudohypetrophy)	Rapid	Frequent
Facioscapulohumeral (FSH) dystrophy	Autodomal dominant	In first or second decade	Shoulder girdle, neck, face, pelvic girdle (late)	Moderate	Occasional
Limb girdle (LG) dystrophy	Poorly defined or recessive	Variable	Pelvic and shoulder girdles	Variable	Variable
Myotonic dystrophy (MyD)	Autosomal dominant	Variable—birth to fifth decade	Distal extensor muscle, eyelids, face, neck, hands, pharynx	Slow, related to age at clinical onset, faster with younger patients	Frequent

Duchenne's MD

- Most common form, X linked inheritance
- **Etiology**: Deletion of DNA segment.
  Possible etiologies:
  - May have neurogenic basis due to abnormal motor neurons
  - Abnormal perfusion
  - Intrinsic muscle fiber abnormality

Manifestations of Duchennes

- Dx age 3 due to slow motor development
- Delayed sit, stand, walk, climbing
- Pelvic girdle weakness – waddling gait
- Hypertrophy of calf muscle
- Gower sign
- Toe Walking
- Muscle shoulder girdle involvement
- Muscle wasting and atrophy

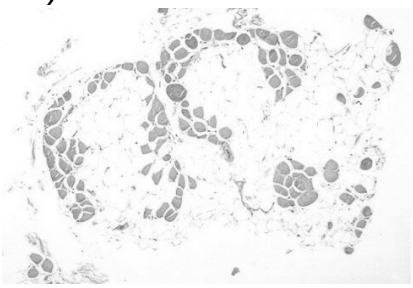
Duchenne's Pathology

- Muscle cells diminish, replaced by interstitial connective tissue and fat leading to increased muscle bulk
- Phagocytic necrosis of fibers and decreased number of fibers
- Disorganized tendinous insertions associated with fat infiltration
- Fibers may regenerate, but nonfunctional over time

MD Effects

- Muscle cells have increased numbers of nuclei forming chains and move centrally
- Muscle cells show necrosis and phagocytosis
- Muscle fibers may be swollen, indistinct, homogenous, with disturbed striation, no pattern to damage
- Fibers may be atrophic or hypertrophic
- Fibers replaced by fat and connective tissue

Fatty Infiltrates in Muscular Dystrophy (CDC, Ewing, 1972)



Duchennes MD

- **Complications:**
  - Kyphoscoliosis – altered version
  - Cardiac involvement
  - CHF
  - Mental retardation
  - Systemic smooth muscle dysfunction

Case Study

- 67 yo w F CC back pain unrelieved by rest, positioning, heat or cold, anti-inflammatories
- **PMH**-HTN 20 yrs controlled by mild diuretic
- 10 yrs ago-mouth cancer excised from mouth floor w skin graft. No add. TX
- 5 yrs ago-Br CA stage 1; lumpectomy w chemo and radiation
- Unremarkable menopause age 50, no HRT
- **Social**-married 45 yrs, 4 adult children, smokes 2 pk/day 20+ yrs, moderate to heavy drinker
- Physical exam-small stature (5 ft, 81 lbs), significant dowagers hump, thin skin, multiple ecchymoses, central obesity, abdominal striae, plethoric facies.

Case Study

- **Adopted 2 yo male. Parent CO decreased activity. Appears to avoid use of L leg and apparent limp. Child CO "knee pain". Hx-insignificant except for URI and GI illness (10 days ago).**
- Seen at ortho clinic for "toeing in" mostly on L. Brief use of brace, opted for conservative TX.
- Birth parent HX unknown.

Case Study

- 43 y.o. W M presents w acute L ankle inflammation. Job requres much walking. States broke L ankle as teen. No recent trauma.
- PMH-untx HTN
- **Appears moderately obese. High stress job. Has been fasting to lose wt. Aspirin taken for HA associated w fasting. No smoking HX. Alcohol consumed 3 days prior at wedding. Was on fishing trip 2 days ago.**

Case Study

- 4 y.o. male presents. Appearance shows bulging eyes. Jerky, flailing movements. Calve appear overdeveloped for age. Difficulty rising from floor. Uses hands to “climb” up his legs to standing position.