Presentation Objectives

1. Brief Melo Overview
2. Discuss common impairments
3. Discuss Physical Therapy’s Role
4. Surgical Options
5. Case Study
6. Exercise Suggestions
7. Questions
Melorheostosis Overview

• What is Melorheostosis
  – A rare benign but progressive disease that is characterized by hyperostosis (thickening) of cortical bone
  – Melo affects both bone and soft tissue (i.e. muscle, tendon, ligament) growth and development
  – Results in severe functional limitations, extensive pain, soft tissue contractures, limb/hand/foot deformity
  – Incidence 1/1,000,000

What is the cause

• Melorheostosis is due to a mutation of the LEMD3 gene (MAN1)
  – Mostly unknown
  – Encodes an inner nuclear membrane protein
  – This developmental error affects both intramembranous and endochondral bone formation
  – Commonly affecting the diaphysis/metaphysis epiphysis of adjacent bones and joints

*Azouz and Greenspan (2005)
Where does Melo Develop???

- **Most Common**
  - Arms, hands, legs, and feet
    - Can be unilateral or bilateral
    - Upper or lower quadrants or both
- **Less Common**
  - Pelvis, hips, sternum, ribs
- **Rare**
  - Spine and skull
Signs/Symptoms

- Irregular bone growth including cortical thickening and ‘candle wax’ appearance
- Limb length inequalities
- Joint swelling and fusion
- Soft tissue abnormalities including tendon and ligament shortening, absent or abnormal muscles, calcification, contractures resulting in malformed or immobilized joints
- Range of motion limitations
- Pain and stiffness
- Sensitivity to cold
- Hyper-pigmentation of skin
- Vascular abnormalities

Clinical Signs

- **Very Frequent**
  - Restricted joint mobility
  - Skeletal anomalies

- **Frequent**
  - Upper limb asymmetry
  - Lower limb asymmetry/hemihypertrophy

- **Occasional**
  - Thick Skin
  - Haemangioma-capillary
Physical Therapy’s Role

• What is Physical Therapy
• Model Definition-APTA
  – Physical Therapy includes:
    • Examining individuals with impairment, functional limitation, and disability or other health related conditions in order to determine a diagnosis, prognosis, and intervention.
    • Alleviating impairment and functional limitation by designing, implementing, and modifying therapeutic interventions.
    • Preventing injury, impairment, functional limitation, and disability, including the promotion and maintenance of fitness, health, and quality of life in people of all ages.
    • Engaging in consultation, education, and research.

*www.physicaltherapy.com
Physical Therapist’s Role cont…

• Physical Therapists specialize in the evaluation, diagnosis, prognosis, and treatment of any impairment of the human body in terms of movement and pain

• We don’t treat “diseases” such as melorheostosis but we do treat the signs and symptoms of melorheostosis
PT role continued...

• As per the 2006 Melo Newsletter
  – Melorheostosis’ symptoms include:
    • Limb length inequalities
      – Commonly affect the gait cycle as well as the joints of the lower back and lower extremities
      – PTs are trained in gait training, orthotic intervention, joint motion restoration, soft tissue mobilization, ankle, knee, hip and low back pain
    • Joint swelling and fusion
      – PTs can provide modality treatments, edema massage, joint stretching, joint manipulation, blood flow stimulation, positioning
PT role continued…

• Melorheostosis’ symptoms include:
  • Soft tissue abnormalities including tendon and ligament shortening, absent or abnormal muscles, calcification, contractures resulting in malformed or immobilized joints
    – PTs can provide joint manipulation, soft tissue manipulation, muscle stretching/strengthening, restoration of joint movement
  • Range of motion limitations
    – PTs specialize
PT role continued...

- Melorheostosis’ symptoms include:
  - Pain and stiffness
    - PTs can use modalities, massage techniques, pain education, and most importantly FIND WHERE THE PAIN COMES FROM
  - Vascular abnormalities
    - PTs can affect blood flow through positioning, soft tissue mobilization, and movement
Surgical Options

• Based on an article by Dr. Jeffrey King and Dr. James Dobyn
  – “Surgery to alleviate mechanical effects from melorheostosis in adults seems to be fairly effective”
  • Particularly true when these effects are due to asymmetric bone growth
  • Not as successful when soft tissue shortening is the cause
Dr. King and Dr. Dobyns go on to say...

- Surgery to relieve pain is rarely effective unless pain is derived from nerve pressure or irritation.
- Contracture releases are more effective in adults than in children.
- “Soft tissue releases alone in skeletally immature patients (children) have a 100% ‘failure’ rate in the literature.”
Surgery???

- Most important thing is to be educated
- SURGERIES CANNOT BE REVERSED
  - So research all of your options and also research your surgeon
    - Although most orthopedic surgeon can do joint and soft tissue releases-get someone that understands the disease and the research behind it (surgeons do surgery)
  - Speak with your Physical Therapist
    - Conservative treatment can always be tried first
    - All of these surgical options should be followed by physical therapy interventions to ensure successful tissue healing and return of motion
Melorheostosis
Case Study
Case Study

• Jennifer Gordy
  – Initial evaluation on 1/29/08
  – Prescription from PCP for eval and treat for L hip pain
  – She had been told to take advil and ice
  – NO RUNNING-stopped for 3 weeks
IE-Subjective

- 29 yo female
- Primary complaints L hip pain-onset early January
  - Pain is sharp and point specific to her L superior hip region
  - Pain increases with running, elliptical (mod), biking doesn’t affect, stationary positions
  - Unable to sleep on L side-preferred position
  - Occasional shooting pain and N/T in her LLE
IE-Prior Medical History

• Dx of Melorheostosis (1980-Age 2)
• Disease only affects her LEs
• Many corrective surgeries
  – 3-L Achilles lengthening procedures at 2yo, 6yo, 15yo
  – LLE surgical shortening procedure at 16yo
  – 2 Toe surgeries to correct contractures at 15yo, 20yo
IE-Objective

- **Gait Analysis**
  - Moderate R lateral gait deviation with L stance phase
  - Holds LLE in extended position throughout gait cycle

- **Standing evaluation**
  - Stands with LLE in slight hip extension and ER
  - All LLS bony landmarks (i.e. IC, ASIS, PSIS) are elevated
  - All RLS bony landmarks depressed
  - Shifts WBing to RLE after 30 sec
Objective continued…

- Hip, Knee, and Lumbar Spine evaluation
  - Hips
    - Supine and long sitting reveal leg length discrepancy (LLE long)
    - Mild L hip restrictions through medial/superior range
    - Decreased L hip IR PROM
  - Lumbar Spine (low back)
    - Good lumbar segmental mobility
    - Moderate lumbar musculature tone
  - Knee
    - Good B hamstring/quad flexibility
    - B knee AROM WNL-no pain
  - All B LE manual muscle tests grade 4/5
    - This is a “good” rating meaning she was able to move joints through available ROM and hold against moderate resistance
Objective continued…

- **Ankle Evaluation**
  - B ankle DF to 5 degrees
    - Normal Adult DF is 0-20 degrees
    - Need 10 degrees of DF for normal gait mechanics
  - All other B ankle AROM WNL
  - Single leg heel raise test in standing
    - RLE-25 reps
    - LLE-5 reps with more sway
  - Severe L calf atrophy (pt attributes to casting as child)
Problem List

- LLE leg length (long)
- Severe L calf atrophy and weakness
- L hip joint restrictions
- B ankle joint restrictions
- Altered gait mechanics
- Poor LLE balance
- Unable to run secondary to pain
- Unable to sit/stand > 30 min without pain
PT diagnosis

Pt presents to skilled Physical Therapy with signs and symptoms consistent with multi-level and multi-joint dysfunctions with an altered gait pattern
PT Goals

• STG-2-weeks
  – Compliance with HEP and lift application
  – Pt able to sit/stand > 60 min with min pain

• LTG-6 weeks
  – Pt able to sit/stand > 2hrs with min pain
  – L calf strength graded > 90% as compared to R
  – Pt is able to ambulate with normal gait mechanics
  – Pt is able to run 20 min with min pain at 6.5 mph on treadmill
Patient’s Goals

- Decrease Pain
- I want to get back to running before the spring so I can run Red Rocks again!!!

Too much to ask???
Plan

• Lift application
• Activity modification
• B calf and L hip stretching
• B ankle/foot and L hip joint mobilization
• L calf strengthening
• Gait Training
Treatment at IE

• Pt given 3mm lift to wear in R shoe 90% of day
  – Pt demo equal WBing with lift in standing
  – Gait analysis with lift in shoe
    • Equal WB w/ B lateral deviations through stance phase
• Given static stretches
  – L Hip IR/ER in supine
    • 2x/day for 30 sec hold ea
  – Standing gastoc/soleus stretches
    • 2x/day for 30 sec hold ea
    • Also given general calf stretch on stair
• Activity Limitation
  – No running or elliptical training for time being
2nd Visit

• Pt returned 1 week later
  – Pain decreased by 50%
  – No radiating LE symptoms since 1st visit
• Treatment-30 min
  – B hip mobilization/B hip stretching
  – Lumbar spine mobilization
  – Calf stretching
  – Calf strengthening
  – Finished with stationary bike-12min
  – Reviewed prior HEP
• New HEP
  – Calf stretch/strengthening on stair 2x/day
  – Hip flexor stretches 2x/day
3rd Visit

- Pt returned 3 days later
  - Pt hoped that she would replicate her initial improvements and be pain free
  - Still has L hip pain
  - No radiating LE symptoms, LBP minimal

- Treatment
  - Repeated 1st visits treatment but added
    - Hip strengthening exercise
    - Pt instructed in seated hip IR/ER stretching
4-5th Visits

• What changed?
  – Added B ankle and foot joint mobilizations
  – Dynamic Edge training
  – Pt started treadmill walking, stationary biking, and elliptical trainer outside of therapy

• Results after 5 visits
  – Pt is quite sore now after treatment sessions
  – After 24-48hrs grades herself at almost 100%
  – Can now do 10 min on elliptical trainer
    • 1st trial-severe soreness, 2nd trial-minimal pain
6-11th Visits

• What Changed?
  – Added SLS balance training on LLE
    • Pt to also perform as part of HEP
  – Concentration on unilateral calf strengthening/stretching for L side
  – Pt instructed to increase her elliptical time progressively-up to 20 min
  – Returned to recreational softball
  – Backed off to 1 visit/wk

• Results after 11 visits
  – Pt is finding fine line between doing too much and not doing enough
    • Doing more-24-48 hrs to recover
  – Improved L hip and B calf ROM/strength
  – Increased tolerance to exercise
12th Visit

- Patient returns and had successfully performed her first run at Red Rocks
  - Had B quad soreness
  - NO HIP PAIN

She felt great and had confidence in her ability to manage her pain!!!
13-15th Visits

• What Changed?
  – Pt backed off to 1 visit every 2-2 ½ weeks
  – Pt to progressively increase her exercise routine back to her PLOF

• Results
  – Pt discharged from physical therapy services to her own independent fitness routine
Physical Therapy Visits

• Pt was seen for 15 visits over a 16 week period
  – Week 1
    • Evaluation/Treatment
  – Week 2-3
    • 2x/wk
  – Weeks 4-12
    • 1x/wk
  – Weeks 12-16
    • F/u 1x every two weeks
Treatment results at D/C

- Pt has now returned to an even higher level for her fitness routine than prior level
  - Running 15 min every other day
  - Lifting 3x/week
  - Running Red Rocks every other week
  - Playing volleyball, basketball, softball
- Pain results
  - More muscle pains
  - Still gets joint pains with too much activity (ups/downs)

BUT...MY PAINS ARE VERY MANAGABLE
Case Overview

• Initial problems
  – L hip pain
  – Unable to sit/stand > 30 min
  – Unable to run due to pain
  – Diagnosis of Melorheostosis-1980
• Interventions
  – Joint mobilization, stretching, strengthening, activity modification, gait training, balance training, cardiovascular/endurance training, lift application
• Results
  – Pt able to return to all fitness activities of her choice with minimal pains

CONFIDENCE THAT SHE CAN LIVE HER LIFE ACTIVELY AND HEALTHY WITH MELORHEOSTOSIS
Exercise Suggestions

Disclaimer

All melorheostosis patients need to consult with their PCP or other medical professional to insure that they are healthy enough to start and continue a structured fitness program.
Stretching Program

• #1 mechanical impairment with melorheostosis that causes pains is due to muscle shortening and joint contractures
• All stretching should be done in a static manner
  – ACSM Recommendation (American College of Sports Medicine)
    • Frequency: 2-3 days/week
    • Intensity: To a position of mild discomfort
    • Duration: 10-30 sec
    • Repetitions: 3-4 for each stretch
  – Always stretch after your workouts
  – Static stretching is a battle between allowing the stretched muscle to relax and getting it to lengthen
    • If you stretch to aggressively all you are doing is playing tug-o-war with yourself
Muscles to Stretch

- Gastroc/soleus
- Hamstring/quadriceps
- Hip flexors
- Hip external and internal rotators
- Pectoralis Major/Lats
- Shoulder flexion/extension
- Shoulder rotators
- Wrist flexors/extensors
- Low back and Neck*
Strengthening Program

• Consult a Physical Therapist with a CSCS certification or a qualified personal trainer
  – ACSM Recommendations
    • 8-12 Repetitions per exercise
      – Choose exercises that train the major muscle groups
    • Minimum of 1 set/exercise
    • 2-3 days/week
    • Perform all exercises through full ROM
    • Perform all exercises in controlled manner
    • Maintain normal breathing pattern (Valsalva maneuver)
  – Full body resistance training
  – Multi-joint exercises
Time constraints

• Super-setting
  – Doing exercises back to back with no rest
    • Always alternate body parts or regions
      – Ex. Chest/Back, Lower body/Upper body
• Circuit training
  – Do full body workout with no rests between sets
Cardiovascular Training

• ACSM Benefits
  – What does it do for you?
    • Improves your heart’s ability to deliver oxygen to your muscles
    • Increases endurance performance

• ACSM Recommendations
  – Modes
    • The most improvement occurs when using large muscle groups over a prolonged period of time
      – Walking, hiking, running, cardio machines, swimming, cycling, rowing, dancing, skiing, skating, etc.
    • Make it an activity you love-compliance
  – Intensity
    • See a fitness professional
  – Duration
    • 20-60 min (see fitness professional)
  – Times per week
    • 3-5 times/week (progression)
Core Training

- The core is where your body derives its power from
- Its job is to allow you to interact with your environment with your arms and legs while stabilizing the spine
  - Muscles of the trunk
    - Transverse abdominis/Multifidus
    - Global Abs, Back Extensors, Lats, Hamstrings, Hip muscles, Glutes, Quads
- Always perform after your workouts
  - 3x/wk
Core Training Suggestion

- Supine/Prone exercises
- Exercise Ball
  - Stability exercises
  - crunches
  - Back stabilizers
- Balance training
- Unilateral resistance training
Presentation Conclusions

• Although the actual treatment of Melorheostosis lies in the medical field of Medical Doctors and geneticists

• Physical Therapists are the specialists in the treatment of pain that derives from the mechanical impairment of the human body
“Life is a series of obstacles, we may either choose to stop and admit defeat or choose to break through life’s obstacles and live free.”

-Ryan Winters 2008


