

# Melorheostosis

Living an Active and Healthy Lifestyle with Melo

## The Role of Physical Therapy

Presented by Dr. Ryan Winters PT, DPT, CSCS

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# 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

\* Melorheostosis Association Newsletter (2006)

# 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

# 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

\*Melorheostosis Association Newsletter (2006)

# 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.



# 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

# Surgery continued...

- 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-2weeks
  - 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

# 2<sup>nd</sup> Visit

- Pt returned 1 week later
  - Pain decreased by 50%
  - No radiating LE symptoms since 1<sup>st</sup> 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

# 3<sup>rd</sup> 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 1<sup>st</sup> visits treatment but added
    - Hip strengthening exercise
    - Pt instructed in seated hip IR/ER stretching

# 4-5<sup>th</sup> 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
    - 1<sup>st</sup> trial-severe soreness, 2<sup>nd</sup> trial-minimal pain

# 6-11<sup>th</sup> 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



# 12<sup>th</sup> 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!!!

# 13-15<sup>th</sup> 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 too 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

# Questions



Rocky Mountain Spine and Sport

[www.rockymountainpineandsport.com](http://www.rockymountainpineandsport.com)

9218 Kimmer Drive Suite 100

Lone Tree, CO 80124

303-792-7377

[winte403@regis.edu](mailto:winte403@regis.edu)



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