Therapeutic Intervention and Rett Syndrome across the Lifespan

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Role and Approaches

Potential Role for OT and PT across lifespan

1. Remedial: Assess, consult and treat re: increase motor skills, transitional skills, and prevent or reduce deformities, provide sensory programming/recommendations

2. Compensatory: Assess and provide support re: equipment provision, environmental adaptations, accessibility, act as an advocate and consultant for school or other community programs

Goals of Therapy:

1. Maintain or increase motor skills
2. Develop or maintain transitional skills
3. Prevent or reduce deformities

Things to think about

Considerations for therapy:

Apraxia is often the biggest challenge

- This is the inability to coordinate thoughts (what you want to do) and movement (of the muscles)

Spatial disorientation is also a challenge

- Many girls cannot correctly perceive proper upright position of the body
- In order to feel upright they may lean forward, back or to the side
- Sometimes overcorrection of positioning can help

Things to think about

Ataxia

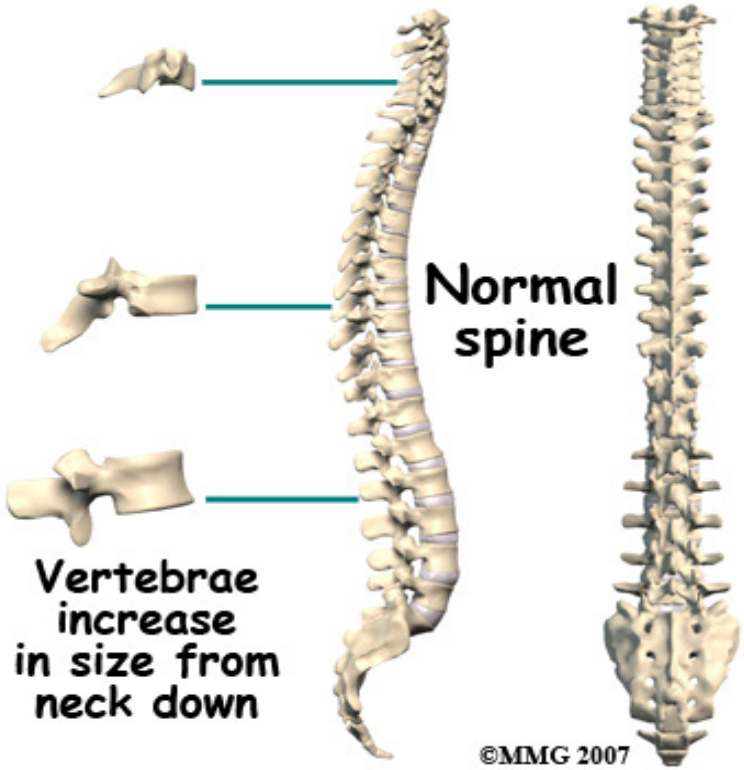
- If present, joints may become temporarily fixed or locked into a position
- Legs may be wide apart in standing because of this
- Weight shifting (in order to step) becomes difficult

To keep her balance the child with ataxia walks bent forward with feet wide apart. She takes irregular steps, like a sailor on a rough sea or someone who is drunk.

Orthopedic Concerns in Rett Syndrome

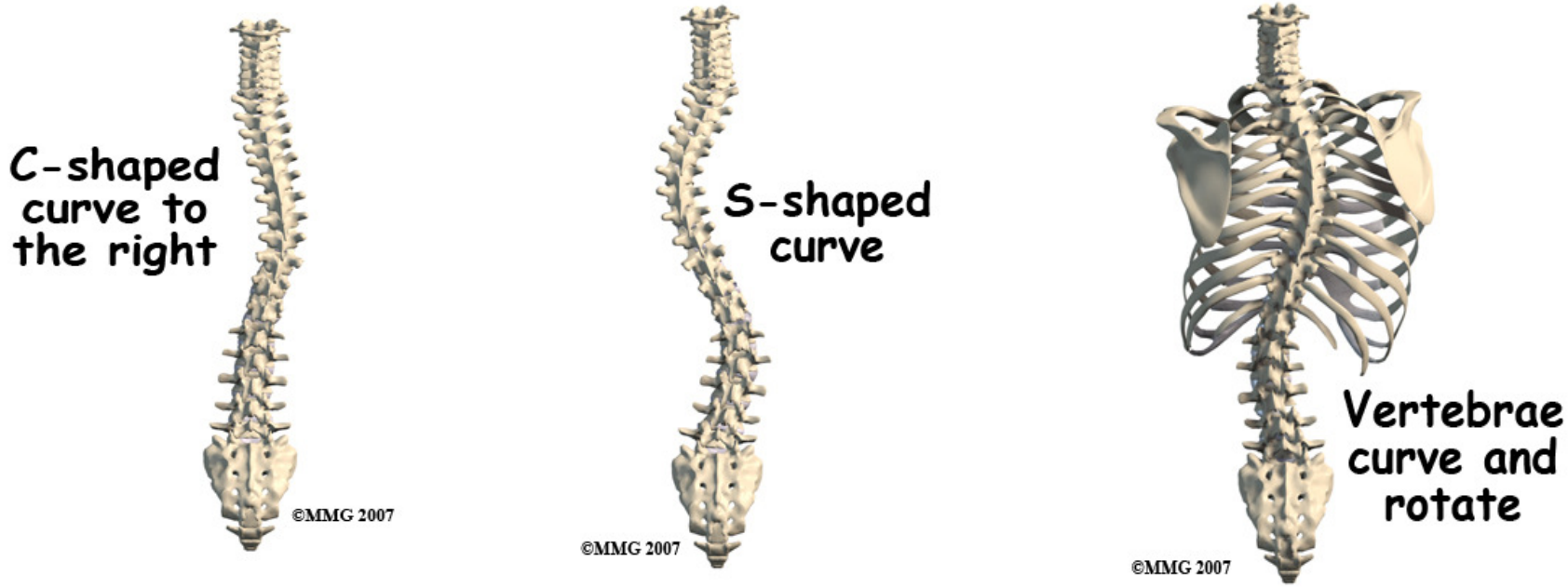
The Spine

- The spine is made of 24 bones (vertebrae) stacked on top of each other to provide upright support
- The spine makes a straight column with the head centered over top



Scoliosis:

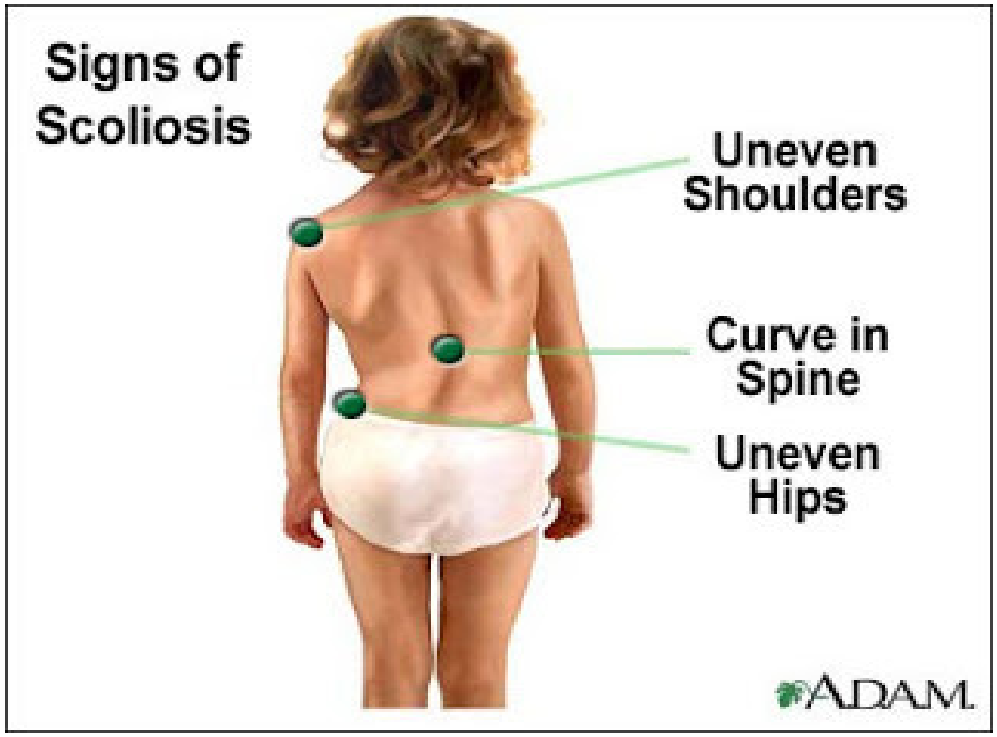
- Abnormal curve in the spine
- Excessive curving forward or backwards (kyphosis or lordosis) or sideways (scoliosis)
- It may include rotation of the spine

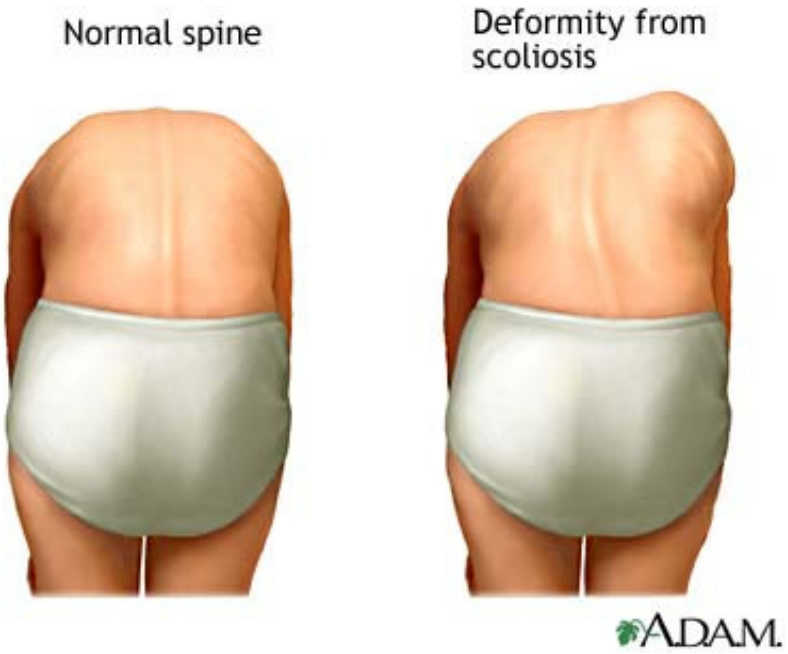


Rett Syndrome and Scoliosis:

- 80-85% will develop a scoliosis
- Progresses rapidly, especially in adolescence
- Prognosis is worse if early childhood hypotonia, in non-ambulators, scoliosis detected before age 5
- Caused by pull of orthopedic asymmetries over time
  - Lack of variety in positioning, decreased active and passive movement, inappropriate equipment for positioning
- Scoliosis can impact seating and positioning, pulmonary function and can cause discomfort

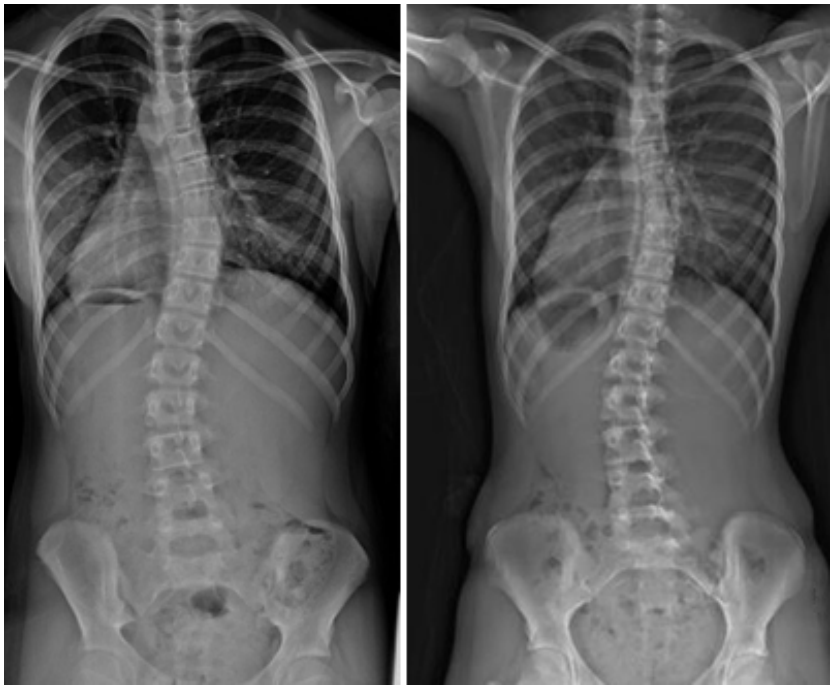
How to detect a scoliosis:





Monitoring of Scoliosis:

- Minimum yearly monitoring of spine
- Should be followed by an orthopedic surgeon 2x/year if a scoliosis is noted before the age of 5



What can we do for scoliosis in Rett Syndrome?

1. Standing program
2. Walking program
3. Stretching
4. Compression garments (for input not orthopaedic)
5. Surgery
6. 24 hour positioning

Scoliosis:
Standing Program

Benefits of a Standing Program:

- Maintain or improve range of motion
- Lessen/manage the progression of scoliosis
- Improve or maintain bone mineral density
- Strengthens the cardiovascular system and builds endurance
- Improves circulation
- Tone management in lower extremity
- Change of position
- Improved self esteem

Standing Program

Recommendations:

- Start with 5-10 minutes of standing at a time
- Gradually build standing tolerance up to 60 minutes/day
- Goal of standing for 60 minutes/day
- Wear orthotics to maintain foot and ankle alignment in standing

Other Tips:

- Knee immobilizers can be used if knees are tight
- A wedge can be added for ankle range of motion
- Try to find a stander that allows your child to be as active as possible in standing

Types of Standers

Types of standers:

- Upright
  - Less supportive, requires good head control
- Supine standers
  - More support, better control of trunk
- Sit to stand

Scoliosis:
Walking Program

Benefits of a Walking Program:

- Maintain or improve range of motion of hips, knees and ankles
- Lessen/manage the progression of scoliosis
- Improve or maintain bone mineral density
- Strengthens the cardiovascular system and builds endurance
- Strengthens legs and trunk
- Change of position
- Enhances the social experience

Walking Program

Recommendations:

- When possible, walking is an important activity to be included in the daily routine
- Walking programs can be completed with a walker, treadmill or support at hands
- Many girls do well with elbow immobilizers on or knee immobilizers (if needed)

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Walking Programs

KidWalk with Upper Body Support

Scoliosis:

Stretching and Compression garments

Stretching

Manual stretching to maintain mobility

- Trunk Rotation
  - Bend knees up
  - Lower knees to one side as far as they comfortably go. Repeat other side

Compression garments

Benefits:

- Increases core stability
- Provides sensory feedback
- Improves muscle control and tone
- Organizes sensory system
- Increases body awareness
- Improves respiratory mechanics

SPIO (Stabilizing Pressure Input Orthosis)
•www.spioworks.com

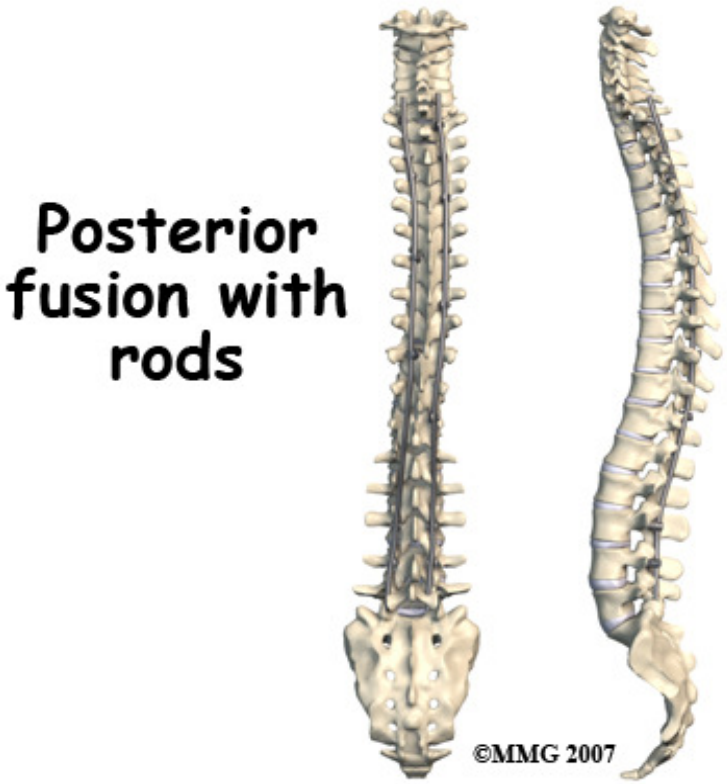
DMO (Dynamic Movement Orthosis)
•www.dmorthotics.com

Scoliosis: Surgery

Spinal Fusion Surgery:

- Goal is to straighten the spine as much as possible

- Spine is straightened using metal rods along the vertebrae with bone grafts to help the spine fuse solidly



Surgery

- Benefits may include:
  - Improved sitting balance
  - Ability to move arms more freely (instead of needing to use them for support)
  - Prevent lung compression
  - May minimize pneumonia risk

- The surgery does take away the flexibility of the spine and might interfere with ability to rotate in bed

24 Hour Positioning Solutions

24 Hour Positioning Overview

- Mobility
- Sleeping
- Transportation
- Feeding
- Bathing/ Toileting
- Play

24 Hour Positioning Overview

- Individual considerations for each child means individualized assessment and approach to interventions (overseen by OT and PT)

- Equipment provision and appropriate choices are different for each child- important to consult with therapists

Sleeping

- Side lying position with supports
  -towels and rolls
  -side rails for safety
  -bumper pads on rails or buckwheat rolls/noodles

- Sleep system (*to be assessed with therapist*-
  consider hospital bed for transfers/bed mobility and positioning as well as surface selection)

Transportation

- Pediatric MTO guidelines re: forward and rear facing
  - rear-facing car seat until the child weighs at least 9 kg (20 lb.).
  - Some rear-facing car seats are made for children that weigh up to 20 kg (45 lb.).
  - best to keep your child in a rear-facing car seat until they reach the manufacturer´s weight or height limits

- Fit of car seat:
  - Harness straps should sit at or slightly below the child's shoulders.
  - You should not be able to fit more than one finger underneath the harness straps at the child's collarbone.
  - The chest clip should be flat against the child's chest at armpit level

Transportation Positioning Options

- **Positioning for younger children under 30 lbs**
  - Snuggin Go
- **Adapted Car seat for positioning (MTO approved- met crash testing standards) **recent change**
  - Convaid Carrot

- **Older child/Adult safe transportation**
  - Accessible transportation (in mobility device)
  - Transfer to seat with harness

Feeding

- **Lateral supports to maintain upright functional feeding posture**
  - Home base option for adapted mobility (strollers)
  - Snuggin Go
  - Other inserts
  - Rolls/pool noodles
- **Adult Feeding**
  - In mobility Device
  - In standard chair

Play

- **Variety of positions for development**
  - Prone
  - Sidelying
  - Sitting
  - Standing

Potential adaptations or equipment

- Home base
- Standers
- Adapted seats/activity chairs
- Floor systems (squiggles)

Bathing

- Support to maintain safe, upright position in bath tub or accessible shower or toilet
  - Adjustable bath seat
    - Minnow
    - Otter/Manatee/Leckey
    - *bath slider system
  - Shower Commode
    - Raz
    - Rifton
    - Invacare (Ocean)

Mobility

- Support for upright functional positioning and orthopedic concerns
- Assessment for ADP approved devices by therapist
- Consideration of each unique individual needs and personal factors (positioning, tolerance, skin integrity, Orthopedic needs, transportation, accessibility, family use and age appropriateness)

Mobility

- Manual wheelchair vs adapted stroller and types (tilt)

- Positioning considerations and focus
  - Laterals
  - Head positioning
  - Pelvic support
  - Tilt

- Associated funding for devices (ADP/ACSD/ODSP)

Mobility: Seating Considerations

Custom seating or modular?

- Age and growth considerations
- Orthopedic concerns (scoliosis/hips planned surgical intervention)
- Tone management
- Tolerance and fit
- Importance of thorough MAT assessment and trial with therapist for each client

Transfers

- Independent
- Assisted
- Dependent (mechanical and person assistance)
  - If lift- PSW criteria for equipment (over 40 lbs)
  - Hoyer, ceiling track, pressure fit lift systems

Sling Selection

- Different styles depending on their use
  - Universal, Hygiene, Walking, Hammock
- Different Materials depending on need
  - Mesh, Parachute, Spacer
- To be discussed and prescribed by your OT

Summary – Equipment Selection

- Therapist assessment and support for equipment prescription including funding process
  - Assessment for different goals/priorities and abilities
  - Importance to monitor and reassess ongoing for growth and changes and orthopedic needs

- Variety of options which may or may not be appropriate for each client at different times (or available in your location)
  - Example- equipment in USA may not be eligible funding wise or approved MTO wise for transit

Other orthopedic Deformities: Ankles and Hips

Joint Deformities – Why is this a problem?

- Many girls with Rett syndrome have abnormal muscle tone with spasticity (the muscle has resistance to fast movements)
- Muscles can be overactive and pull joints out of balance
- This can lead to contractures (permanent shortening of the muscle)
- The **ankle** and the **hip** are the most common affected

Ankles:

- Overactive calf muscles tend to pull the ankle into plantar flexion (toes point down). This can result in foot deformities

- Sometimes the foot will turn inwards as well

Ankles

What can we do?

- **Goal is to prevent or minimize development of deformity**

1. Standing and walking programs
2. Orthotics
3. Serial casting
4. Stretching

1. Standing and walking

- Standing programs are important to prevent foot deformities
- Use a wedge in the stander to increase the stretch

Ankles

2. Orthotics

- **AFOs for stretching:**
  - With a hinge at the ankle joint and stretching “pull” straps
  - Worn overnight, sometimes with a knee immobilizer

- **AFOs to maintain foot alignment:**
  - Worn throughout the day

- **AFOs for standing and walking:**
  - Used in stander and walker for support

Important Information:

- Always watch for red marks or blisters that last for more than 20 minutes when the orthotic is removed
  - Contact your orthotist if you notice this
  - Stop wearing the orthotics until the problem is resolved (otherwise you risk skin breakdown and pressure sores)

- Orthotics will be needed to be adjusted for growth

- Shoes
  - Take out the insole to help the orthotic fit better
  - Look for a shoe with a wide toe box (front ½ of the shoe)
  - Shoes typically need to be about 1 size larger

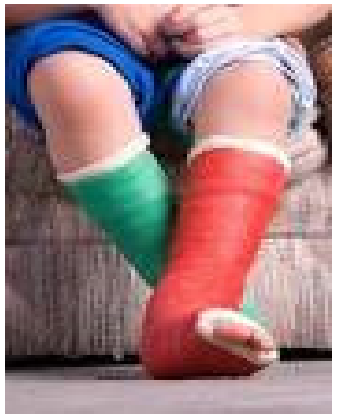
Ankles

3. Serial Casting

- A gradual stretch the muscles in the lower legs by using a series of fiberglass casts (what is used if you had a broken bone)

- The cast is left on for about a week.
  It is removed and a new one is put on

- This is completed for a series of weeks to slowly stretch the muscles

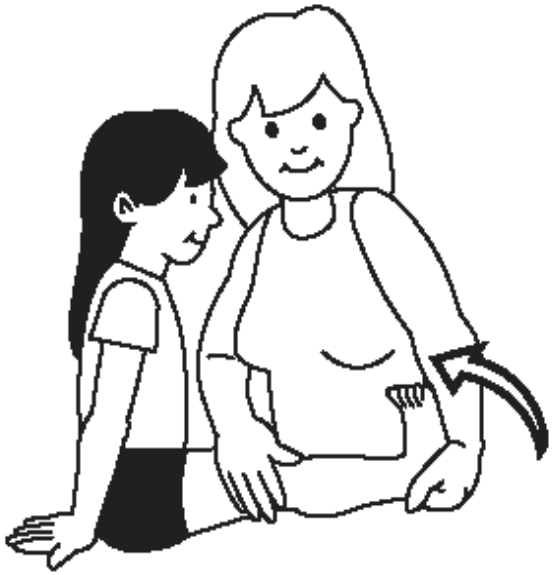


Ankles

4. Stretching:

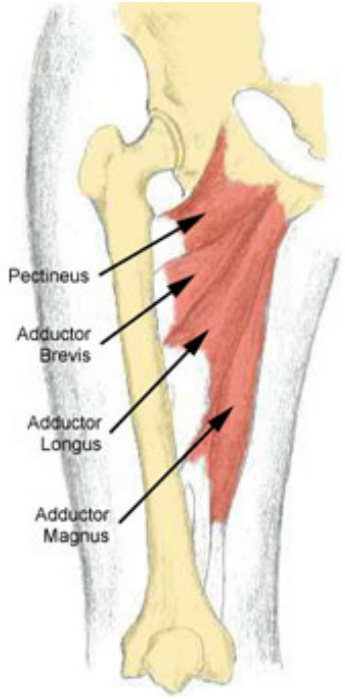
Ankle Dorsiflexion (toes up)

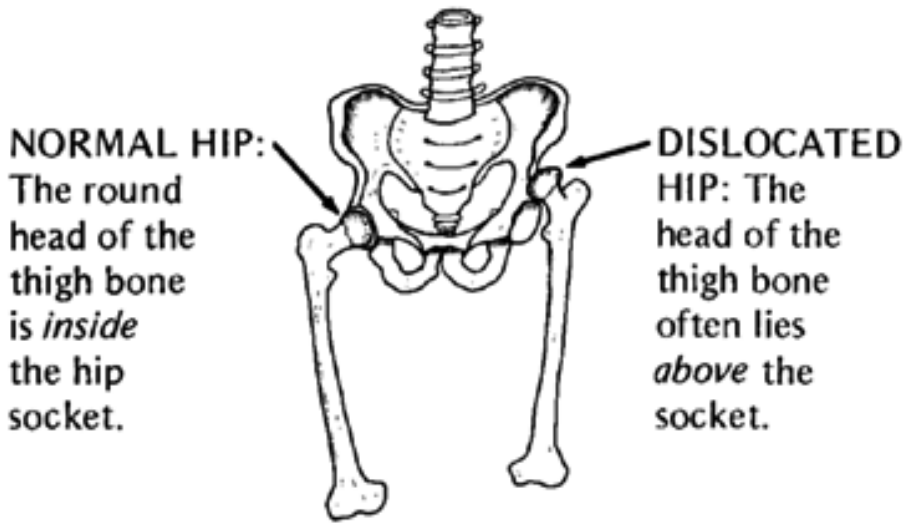
- Hold the heel of the foot firmly in your hand
- Support the rest of the foot with your forearm
- Slowly push back the foot for a firm stretch
- Hold for 30-60 seconds, 2-3x/leg



Hips:

- Overactive hip adductors (pull legs together) or hip flexors (bend hip) can lead to hip dislocation





What can we do?

- If you notice tightness in the hip adductor or hip flexor muscles request a referral to an orthopedic surgeon to monitor changes

1. Positioning with a wedge

- To stretch hip adductors overnight

Hips

2. Sitting in hip abduction (legs apart)

- Can be done over your legs
- Can use a bolster

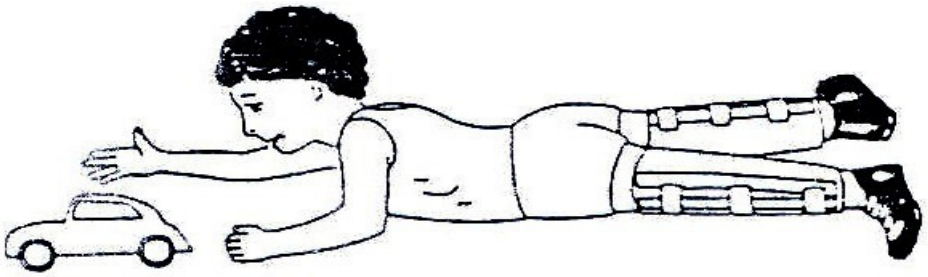
3. Stretching

Hip Extension (stretch front of hip)

- Start in side lying
- Stand behind and hold the hip stable
- Gently bring the leg back to stretch the front of the hip joint

AND/OR

- Spend time laying on stomach



Upper Extremity Splinting and Stretching

Splinting and Stretching

- **Purpose:**
  - Maintain ROM at elbow joint
  - Maintain skin integrity
  - Function

- Home use- positioning and focus (ADLs and communication device access)
  - Therapy session and play (weight bearing and support)

Elbow Extension splint

- **Use and Application** (Custom vs prefabricated)

- Assessment completed by therapist re: type to best address individual child
- Considerations after assessment (ROM, level of support and skin)
    - Cost
    - Care (washing and wearing)
    - Sizing
    - Application

Elbow Extension splint

- **Potential Type and Associated Cost**
  - Custom
  - Bamboo splint (brand of prefab)

- **Funding:**
  - May be covered by insurance
  - Other funding sources (hospital support funding)

Sensory Considerations

OT home programming

- Individualized assessment and set up and consult
  - Individual child presentation
  - Individual child response to environmental stimuli
  - Changes and recommendations (calming vs alerting)
    - Alerting- To start gross motor program
    - Calming- Sleep Hygiene, respiratory

OT home programming

- Collaborative approach to introducing sensory program with family and caregivers and school
- Importance for consistency and routine
- Monitoring routine for changes and adaptations (sensory needs change over time)
- May be used in different manners (example: prep for gross motor)

Sensory Intervention

- Home programs/School programs and education
- Snoezelen Room
- Pressure garments
- Auditory input
- Vestibular input/therapy swings

Other Therapy Options

Hippotherapy

Hippotherapy:

- Horseback riding therapy
- Develop body awareness, improved posture and balance
- Stretches hip adductors when sitting on horse
- Experience the rhythm of walking

Hydrotherapy

- Therapy in the pool
- The water provides good tactile stimulation
- Can be used to increase range of motion, relax, increase strength

Bike

- Adapted bikes are good for range of motion and cardiovascular fitness
- Not many funding agencies, can be expensive

Accessibility: Home and Vehicle

Accessibility

➢ Who is involved…..

Occupational Therapists

- Current and future needs of your child and family
- Support with potential funding sources (Social Workers)

Accessibility Design Consultants

- Costs, selection of potential homes, advice

Contractors/Vendors

- Structural changes/construction

Families

- Previous experiences, advice and suggestions

Accessibility

- **Renovations**
- Access into home (porch lift, ramp, stair lift)
- Interlevel access (main floor set up, stair lift, elevator, telecab)
- Bathroom (wheelchair accessible shower, no threshold)

- Codes, Permits and Expertise

- **Planning/Funding**
- Income dependent (Ontario March of Dimes (HVMP- home), Presidents Choice Childrens Charity, Easter Seals)

- Non income dependent resources

- CRA submission options

Accessibility

- Van Modification/transportation
  - Type of modification: rear entry vs side entry or transfer seat (valet or curbside)
  - Accessible parking permits

- Planning/Funding
  - Income dependent (Ontario March of Dimes (HVMP-vehicle), Presidents Choice Childrens Charity, Easter Seals)
  - Non income dependent resources
  - CRA submission options

Pediatric vs Adult Considerations

Pediatric vs. Adult

- **Girls are getting Bigger- what does this mean?**

- May now require lift/transfer equipment

- May require larger mobility device

- Spine may have progressed OR spine surgery has been completed

- Need for PSW support (ensure lift systems in place if your daughter is unable to assist with transfers, address bathing equipment or potential bed positioning/equipment)

Pediatric vs. Adult

- Goal would be to obtain most equipment, access available funding before your daughter turns 19.
- Continue with stretching/orthotics/positioning/equipment/ home programming recommendations
- Girls eligible to remain in school until 21 years of age.
- Ensure transition planning occurs early (15-16 years) so that you are better prepared.
- Transition to adult services (OT/PT support now serviced through the CCAC, Seating through adult seating clinic or CCAC therapist, continue with current orthotics services, adult ortho surgeon referral).

Pediatric vs. Adult

Funding

- Funding opportunities NO longer available
  - Ex: Presidents Choice Children's Charity, Easter Seals, any children's hospital family fund, Jennifer Ashleigh foundation

- Funding opportunities NOW available
  - Ex: ODSP, March of Dimes Assistive Devices Program

- Other options: local service clubs (Lions, Rotary, Kiwanis), Ceridian Cares

Pediatric vs. Adult

- **Assistive Devices Program (ADP)**
  - Continues to cover manual wheelchair, manual wheelchair with tilt.
  - *Will only fund a stander IF your daughter had one as a child*

- **ODSP**
  - Access for funding for bathing/toileting equipment, hospital beds, 25% coverage of a mobility device.

- **March of Dimes - HVMP**
  - Access to apply to MOD (after 18 years of age independent from family income)- May open up the opportunity to obtain van conversion and/or home renovation assistance.

Take Home Messages/Tips

Tips

1. Ensure OT/PT assessment before proceeding with equipment and therapy at home

2. Obtaining a stander before 19- if appropriate (accessing ADP).

3. Proactive planning/access of funding to optimize resources

4. Start transition process early from your children's treatment center

Questions?

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