Rehabilitation Interventions in MECP2 Duplication Syndrome

MECP2 Duplication Conference 2020

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MECP2 Duplication Syndrome

- Intellectual disability and speech delay
- Fine motor and gross motor delays
- Dysphagia
- Tonal abnormalities
  - Hypotonia with associated joint laxity
  - Spasticity
  - Dyskinesias
- Stereotypies
Rehabilitation Interventions

- Therapies
  - Physical/Occupational
  - Speech
  - Hippotherapy/aquatherapy
  - Music therapy
  - Massage
  - Complementary/alternative = CAM
Rehabilitation Interventions

• Orthoses (bracing/splinting)
• Assistive technology
• Durable medical equipment
• Guidance on spasticity/tone reduction interventions
  – Focal versus systemic
Physical Therapy

- Focus is usually gross motor/lower limbs and trunk
- Flexibility/range of motion
- PREVENT CONTRACTURES
- Strengthening
- Coordination and postural control
- Mobility
  - Transfers
  - Transitional movements/gait training
- Equipment
- Family training
Occupational Therapy

- Focus is usually upper limbs and activities of daily living
- Strengthening/control of posture
- Apraxia/Dyspraxia
- Fine motor/dexterity
- Access to assistive technology/augmentative communication
- Splinting/adaptive aids
Speech Therapy

• Communication
  – Verbal and non-verbal
  – Low tech and high tech

• Swallow skills
  – Oromotor strengthening and coordination
  – Positioning strategies
  – May overlap with OT
Communication

“Not being able to talk is not the same as not having anything to say”
Rosemary Crossley

• Ask for therapist with experience in augmentative communication
• School can bring in “Assistive Technology” consultant
Hippotherapy

- Rhythmic motion of horse simulates gait
- Helpful for trunk strengthening and balance
- Benefits are more than physical
  - Socialization
  - Emotional
  - Cognitive/Motivation
Aquatherapy

- Start program with therapist then practice
- Buoyancy helps with mobility
- Watsu techniques assist with relaxation
- May need light weights for strengthening program and to give additional proprioceptive feedback
Alternative therapies

• Music and massage improve social interactions, environmental awareness and help with relaxation
  – Music in particular serves as a motivator and can help to arouse or calm

• Neurophysiological Responses to Music and Vibroacoustic Stimuli in Rett Research in Developmental Disabilities 35(1) June 2014
Goals Should be Individualized

• Every child is different…no standard path
• Wide variation, course is not predictable and prognosis may be unknown
• Some children have greater deficits in strength or tonal abnormalities than others
• Some have medical issues that will impact therapy participation
  – Anxiety/pain, seizures, infections
Therapy Approaches

- One therapy approach may work for one child and not for another – be flexible
- Therapists should be willing to seek out suggestions and collaborate with each other (same facility or other)
- While there may be similarities in approaches to children with CP, children with MECP2 duplication syndrome are very different
Family Participation

• Practice, practice, practice = Repetition is key

• Ideal therapist provides feedback about technique/positioning, alternate strategies, and is willing to change directions/approaches

• Family/caregiver involvement is a must so that there is a team approach
  – If goals are not within reach then this may jeopardize continued approval of services

• ROM to be effective must be done daily!

• Many insurers are now looking at documentation that family is performing a home exercise program
Therapy Intensity and Location

• Outpatient therapy most common
• Adds to school based programs which are only required by law to provide “educationally based” therapy
• Therapy goals and progress determine frequency and duration
• May do a “burst” program and reassess at regular intervals
• Clinic setting provides greater access to equipment
• Home health for more medically fragile children however expertise may vary significantly
Musculoskeletal Changes

• Architecture of muscles and joints change with time and growth

• Goals are to preserve
  – Upright posture
    • Social interaction
    • Safe swallow
    • Functional use of upper limbs
    • Medical benefits – GI/respiratory
  – Full joint motion
  – Weight bearing ability if able
Orthopedic Issues Can Impact Posture and Seating

- Hamstring contractures contribute to posterior pelvic tilt
- Scoliosis can affect functional use of upper limbs
Orthoses

• Support/stabilize joints
  – Improve alignment
  – Impact proximal segment
  – Decrease hypermobility

• Prevent/limit deformity
  – Severe spasticity or early contractures may require stiffer plastic and inner boots for AFOs.
  – Scoliosis bracing is also influenced by severity of curve and degree of truncal weakness/hypotonia

• Don’t be afraid of bracing – be proactive.
Supramalleolar Orthosis

- Plastic extends above malleoli (ankle bones)
- Primary effect is control over pronation or “collapsed arch”
- Uses:
  - Pes planovalgus (from hypotonia with excessive motion or mild hypertonia with restricted motion)
  - Mild control of toe walking due to plantarflexor hypertonia
Articulated AFO
Hinged ankle foot orthosis

- Articulated allows more fluid motion
- Hinge will add width to brace
- Add straps or stops to control crouch
- Inner boot of softer material assists to pad bony prominences

*Shown with molded inner boot. Optional.*
Lever Arm Dysfunction

• May be seen in low or high tone children
• Essentially limbs are not ideally aligned for muscles to function as they should
• Changes foot progression angle usually with far rotation outwards
• Lose lever arm for push off with crouch and decreased knee extension
• Secondary knee and hip flexion contractures
Trunk Orthoses

• Compression garments
  – Proximal stability
  – May improve arm function
  – Proprioceptive input
  – Caution if cardiac involvement

• Scoliosis
  – Flexible more for postural support
  – Rigid for true scoliosis
Upper Limb Orthoses

- Optimize function
- Prevent deformity and limit self-injurious behaviors
- Benik brand products popular as neoprene less likely to scratch if hands brought to face

Custom polypropylene hinged brace with stops
Pediatric Equipment

- Treating therapist(s) input important
- Vendor should have pediatric experience and assure growth potential
- Postural support important for both function and to aid in limiting deformity
- Needs change over time
- Where will equipment be used?
- Standers and gait trainers have different functions
Pediatric Equipment

- Typical items covered by payors with supporting documentation/necessary criteria met include: wheelchairs, gait trainers, standers, bath equipment, specialty beds and carseats

- Difficult to obtain items include devices used by able bodied population such as strollers, exercise equipment and items without supporting literature for efficacy or benefit.
Gait training

Kidwalk
www.primeengineering.com

Lite gait
www.litegait.com
Adaptive Trikes

- Supports are customized
- Feeling of security in seated position
- Promotes dissociated movements and strengthening

Not usually covered by insurance but charity/philanthropic
Goals of Hypertonia Management

• Decrease spasticity or dystonia if interfering with volitional movement
• Improve comfort
• Prevent or decrease progressive deformity
  • Contractures
  • Bony changes
    • Hip subluxation, rotational changes
• Help with caregiving and positioning
  • Dressing, hygiene, diapering
Traditional Step-Wise Approach to Treat Hypertonia

- Neurosurgical and orthopedic procedures
- Medications for hypertonia
- Rehab Therapies/Orthoses
- Assess for medical reason for change in tone and/or remove noxious stimuli
Oral Medications for Hypertonia

• May be recommended for generalized hypertonia
• Take into consideration potential side effects
• Decreased postural control
  – Increased drooling
  – Constipation and GI upset
  – Interaction with other medications
• In severe cases, usually meds help some but not enough and maximum doses are reached due to side effects
Botulinum Toxin A Injections

- Works at interface between nerve ending and muscle = neuromuscular junction
- Provides focal tone reduction without systemic side effects
- Duration of effect 3-6 months
- Dose limited by weight
- Covered by most insurers
- Can be performed with sedation or topical anesthesia
Serial Inhibitive Casting

- Used alone or along with botox/phenol procedures for lengthening of muscle and tendon unit gradually
- Different protocols but generally one cast change per week for average of six weeks
- Custom footplate built into cast to ensure that arch is supported properly
Intrathecal Baclofen Pump Therapy

- Used for global tone reduction in patients with severe spasticity or dystonia
- Catheter placement will impact degree of tone reduction
- Implanted system directs concentrated baclofen to spinal fluid
New Developments - Lokomat
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