Hypotonia

Introduction: Muscles and Muscle Tone

Our body contains over 650 **muscles**, which keep our internal organs working and let us move in many ways. Muscles are made of muscle cells that are bundled tightly together. **Muscle tone** **is the amount of tension in the muscles at rest or during passive movements**. We need proper muscle tone to hold our body upright.

When muscles receive signals from the nervous system, they contract. Muscle contraction changes the muscle tone and allows us to move. The amount of muscle tone determines the control, speed, and range of movement.

Hypotonia

Hypotonia means **low muscle tone**, or the muscles are "floppy". In a baby with hypotonia, the muscles show low or no resistance when the joints move. He/she may feel like a rag doll.

Hypotonia is usually detected soon after a baby is born. It may happen due to problems in the nerves or muscles, but it may have no known causes. It can be an indication that the baby has another underlying medical condition.

Symptoms of Hypotonia

Symptoms vary depending on the cause of hypotonia, but these are some of the common symptoms in babies with hypotonia:

- Feeling limp or floppy when being carried
- Muscle weakness
- Joint hyperflexibility (bending beyond the normal limits)
- Arms and legs that hang straight with no bend at elbow / knee
- Difficulty swallowing or sucking
- Unusual breathing (often more shallow)
- Struggling to reach motor-related developmental milestones, e.g. holding up the head without support, rolling over, sitting, crawling, walking etc.

Implications of Hypotonia in 22q11.2DS

- **Significant motor delays** (certain movements and postures happen at an older age than expected)
- **Coordination difficulties** (muscles do not work well together)
- **Pharyngeal hypotonia** (poor muscle tone in the throat)
  - Floppy muscles block the airway, making it difficult to breathe, especially when sleeping. See our sheets on Obstructive Sleep Apnea
  - Floppy muscles make it hard to swallow. See our sheets on Feeding Difficulties and Swallowing and Dysphagia

Hypotonia and 22q Differences

22q11.2 deletion syndrome (22q11.2DS)

- Studies on children and adolescents (aged 1 -19 years) who had 22q11.2DS found that 54 – 76% had hypotonia.
- Hypotonia is extremely rare in adults with 22q11.2DS
- Hypotonia was not found to correlate with age or IQ

22q11.2 duplication syndrome (22q11.2DupS)

- In a study in Belgium, 7/26 (27%) of children with 22q11.2DupS had hypotonia

Causes of Hypotonia

There are many causes of hypotonia. It is a symptom of multiple genetic disorders and metabolic syndromes (problems with how the body uses energy).

Hypotonia can happen due to:

- Damage in brain or spinal cord
- Problems with the formation of the brain or the spinal cord
- Problems in the nerves or in the way the nerves communicate with muscles
- Problems in the muscles
- Infections
- Lack of oxygen before the baby is born or right after

Hypotonia (continued)

Diagnosis of Hypotonia

- **Hypotonia is often noticeable within the first few months of life.**
- The doctor can check the baby's:
  - Balance and coordination
  - Motor skills (ability to grab things, roll over, sit up, etc.)
  - Reflexes (automatic, instantaneous, unlearned movements that the body makes in response to stimuli)
  - Response to sensory stimuli such as things they see, hear, or touch.
  - Brain activities (using MRI or CT)
  - Muscle and nerve functions (using Electromyography)

Treatment of Hypotonia

In some cases, hypotonia can be a longstanding condition, but muscle tone may improve through treatment:

- Management of conditions that led to hypotonia
- Physical therapy – to improve motor skills
- Speech-language therapy – to overcome problems in speaking, swallowing, and breathing
- Sensory stimulation therapy – to improve sensory functions
- Tube feeding – Insert a gastric tube ("G-tube") to the stomach or the intestines to deliver nutrients directly there, bypassing any hypotonia in the throat area.

Handling a Child with Hypotonia

Children with hypotonia are **fragile** and get **injured easily**.

- They may be awkward to carry.
- They may feel heavier than you would expect.
- They may not be able to control their head position
- They may not tuck their arms and legs when they are carried (so the arm/leg maybe left attended and prone to injury).
- Their bones may be fragile.
- Their joints may dislocate easily.
- They may not react quickly to something unexpected.
- Their muscle tone may change with their mood or the environment.

Handle the child with **extra care**:

- Always provide lots of **support**.
- Handle **carefully** and move **slowly**.
- Know where the child's **arms** and **legs** are.
- **Never pull a child up by the arms.**
- **Never pull a child's legs up by the feet.**
- Have the child help as much as he or she can.
- Make the child comfortable and aware.
- **Pay attention** to what you are doing.

This video on Caring for Children with Low Muscle Tone (Health Sciences Centre Winnipeg) provides useful information on the safe handling, positioning, range of motion, dressing, and lifting + transferring of children with hypotonia. Also see their Safe Handling Manual.

Resources

- Hypotonia in Babies – Cleveland Clinic
- Low Muscle Tone – The Royal Children's Hospital Melbourne
- Evaluation of the floppy infant – 2015
- Electromyography
- Caring for Children with Low Muscle Tone (video) and Safe Handling Manual (pdf) – Health Sciences Centre Winnipeg
- Updated clinical practice recommendations for managing [**children**][**adults**] with 22q11.2 deletion syndrome – 2023
- Disabilities and cognition in children and adolescents with 22q11 deletion syndrome – 2007
- Velo-cardio-facial syndrome: a distinctive behavioural phenotype – 2000
- Cognitive and behavior profile of preschool children with chromosome 22q11.2 deletion – 1999
- Cross-sectional and longitudinal findings in patients with proximal 22q11.2 **duplication**: A retrospective chart study – 2022
- ❖ Graphic of baby with hypotonia from Wikipedia

International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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