

PediatricsⁱⁿReview[®]

Hypotonia in Infants

Laurence Stiefel

Pediatrics in Review 1996;17;104

DOI: 10.1542/pir.17-3-104

The online version of this article, along with updated information and services, is located on the World Wide Web at:

<http://pedsinreview.aappublications.org/content/17/3/104>

Pediatrics in Review is the official journal of the American Academy of Pediatrics. A monthly publication, it has been published continuously since 1979. Pediatrics in Review is owned, published, and trademarked by the American Academy of Pediatrics, 141 Northwest Point Boulevard, Elk Grove Village, Illinois, 60007. Copyright © 1996 by the American Academy of Pediatrics. All rights reserved. Print ISSN: 0191-9601.

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™



apy for relief of acute symptoms. Although beta-agonists may themselves prove to be harmful (in particular the safety of fenoterol has been questioned), a more likely explanation is that the ongoing inhalation of a beta₂-adrenergic agent may result in

underuse of appropriate anti-inflammatory drugs, such as cromolyn or a corticosteroid. Perhaps the best use of inhaled beta₂-agonists may be for treatment of acute exacerbations and for prophylaxis against exercise-induced asthma, not for long-term,

regularly scheduled administration. The first-line choice for ongoing maintenance therapy probably should be an anti-inflammatory agent.

Henry M. Adam, MD
Associate Editor, *In Brief*

IN BRIEF

Hypotonia in Infants

Clinical Evaluation of the Floppy Infant.

Crawford TO. *Pediatr Ann.* 1992;21:6:348-353

Floppy Infant. Ment LR. In: Oski FA, DeAngelis CD, Feigin RD, Warshaw JB, eds. *Principles and Practice of Pediatrics*. 1st ed. Philadelphia, Penn: JB Lippincott; 1990:326-329

Neonatal Hypotonia. Miller VS, Delgado M, Iannaccone ST. *Semin Neurol.* 1993;13:73-83

Neonatal Peripheral Hypotonia: Clinical and Electromyographic Characteristics. Parano E, Lovelace RE. *Child's Nervous System.* 1993;9:166-171

Neonatal hypotonia can have many different etiologies. Floppiness in an infant can be caused at various levels of the nervous system from disorders of the brain to spinal cord lesions, neuropathies, neuromuscular junction disorders, and myopathies. A variety of diagnostic tools are available for defining the source of the hypotonia, but before any serum values, muscle biopsies, electromyograms (EMGs), or nerve conduction studies are ordered, a thorough neurologic examination is essential for determining the diagnosis.

The first goal in diagnosing the source of neonatal hypotonia is to ascertain if it is central or peripheral. Infants who have central hypotonia from a brain source usually have other central deficits. If the child is alert, responds to surroundings, and has normal sleep-wake cycles, the hypotonia is likely to be peripheral in origin. On the other hand, if the child lacks visual tracking, appears lethargic, and has seizures or delayed milestones, it most likely is caused by a central source.

Central Hypotonia

The most common forms of neonatal floppiness are central in origin. Disorders of the developing brain that cause hypotonia include hypoxic en-

cephalopathy, intracranial hemorrhage, infection, metabolic disorders, perinatal trauma, hypoglycemia, hypothyroidism, and chromosomal abnormalities. Infants who have central brain disorders present with a decrease in active motor strength compared with passive tone, which actually can be increased markedly. A typical infant would have poor head control and hip/shoulder weakness, but with spastic extremities. Reflexes are either normal or increased, as opposed to most peripheral causes, where the reflexes are decreased significantly.

Hypotonia can be caused by brain dysgenesis. In such cases, the patients often have dysgenetic features, such as malformed facies, fingers, and ears. Recognizable syndromes such as trisomy 21 and Prader-Willi have hypotonia as a prominent feature.

Cervical Spinal Cord

Cervical spinal cord trauma can present with hypotonia. The affected babies begin by exhibiting hyporeflexia, but days to weeks later they develop hyperreflexia and spasticity. The patient also develops other neurologic symptoms, including respiratory difficulty, bladder dysfunction, decreased rectal tone, and vasomotor instability, depending on the level of cord transection. Cervical spinal cord transection most commonly is a complication of the delivery of breech or cervical presentations.

Anterior Horn Cell Disorders

Infantile spinal muscular atrophy, also known as Werdnig-Hoffman syndrome, is an autosomal recessive disease that causes anterior horn cell degeneration. One third of patients

who have this condition present in the neonatal period. These patients present first with decreased fetal movements and often have had a breech presentation. Also secondary to decreased fetal movement is the presence of congenital contractures and hip dislocations. At birth the infants present with hypotonia, generalized weakness, absent reflexes, and poor suck and swallow. The infants assume a frog leg posture soon after birth. However, they have alert, inquisitive facies, in contrast to those who have central, cortical types of hypotonia. Patients who have infantile spinal muscular atrophy have the classic tongue fasciculations that are noted in this disorder; often, finger and biceps fasciculations are noted as well. Definitive diagnosis is made by muscle biopsy, which demonstrates perinatal denervation.

Glycogen storage disease type 2, also known as Pompe disease, is an autosomal recessive condition that causes an acid maltase deficiency leading to glycogen deposits in anterior horn cells as well as in liver and brain tissue. The patients present with weakness, hypotonia, cardiomyopathy, and hepatosplenomegaly. These patients also have tongue fasciculations and muscles that feel rubbery to palpation. Diagnosis is made by a muscle biopsy demonstrating vacuolar myopathy.

Although presently a rare infection, poliomyelitis can cause neonatal hypotonia. The poliovirus infects anterior horn cells. Neonatal cases can present in the first few weeks of life with the sudden onset of asymmetric weakness. The patients also can have encephalitic and bulbar symptoms. Diagnosis usually is made by detection of slowed nerve conduction velocities, but the poliovirus also can be isolated from stool cultures.

Neuromuscular Junction

Transient neonatal myasthenia gravis (MG) develops in 10% of children whose mothers have MG. Infants present with facial palsies, impaired suck and swallow, a weak cry, generalized weakness, and hypotonia. Respiratory assistance may be needed until the condition resolves spontaneously. In transient neonatal MG, maternal antibodies to the acetylcholine receptor cross the placenta and affect the newborn. The diagnosis should be suspected strongly in patients whose mothers are known to have MG and can be confirmed by an edrophonium or neostigmine challenge. The transient neonatal variety of MG usually resolves in 6 weeks.

Congenital MG is a much less common disorder and usually does not present in the immediate neonatal period. When it does develop, congenital MG first presents more commonly with ptosis and ophthalmoplegia, and hypotonia does not develop until later.

Infantile botulism, caused by the exotoxin of *Clostridium botulinum*, usually presents between the ages of 6 weeks and 12 months. The toxin prevents nerve terminals from releasing acetylcholine. Infants classically present first with constipation, but soon afterward develop hypotonia and weakness, with a weak cry, poor feeding, facial weakness, ptosis, and decreased eye movements. The patients usually are areflexic with unreactive pupils. Apneic spells are common, and ventilatory support is indicated in those cases. Symptoms develop several hours to 1 week after ingestion of contaminated food. Cases also have occurred after wound infections and from ingestion of soil, household dust, and honey. The diagnosis is established by identifying the *C botulinum* organism or its toxin in a stool sample or from toxin in the

serum in the case of a wound infection.

Drugs have been found to interrupt function at the neuromuscular junction and cause hypotonia. Hypermagnesemia can occur secondary to the treatment of severe eclampsia with magnesium sulfate. The high levels of magnesium block the release of acetylcholine from the nerves and cause weakness, hyporeflexia, and increased risk of respiratory failure in the infant. The diagnosis is established by checking serum magnesium levels. Treatment is supportive.

Aminoglycoside antibiotics such as gentamicin, neomycin, and streptomycin can disrupt the presynaptic release of acetylcholine and cause hypotonia and areflexia. Management of affected infants is supportive.

Congenital Myopathies

Congenital myotonic dystrophy, also known as Steinert disease, is an autosomal dominant disorder most commonly inherited from the mother. Mothers who also have symptoms from the condition may develop uterine dystocia, which can lead to complications during delivery. These babies are born with severe hypotonia, bilateral facial weakness, poor suck, areflexia, and respiratory distress. They have a characteristic facial appearance, with tenting of the upper lip, thin cheeks, and wasting of the temporalis muscles. They also tend to have dislocated hips and arthrogryposis—most particularly, club feet—secondary to decreased fetal movement. Respiratory failure and the increased risk of aspiration often leads to early death, but if the infants survive the first 3 weeks of life, motor function may improve significantly, although facial diplegia persists.

The clinical course of these patients depends on how much muscle weakness improves over the long term. The patients tend to have intellectual deficits, but it is unclear whether they are secondary to central nervous system dysfunction or perinatal asphyxia. Patients who have myotonic dystrophy also have other abnormalities, including gonadal atrophy and cataracts. Diagnosis is supported by an EMG demonstrating myotonic discharges, but this finding may not be detectable in infancy. In many cases the diagnosis is confirmed by noting myotonic symptoms or a characteristic EMG in the infant's mother.

A wide range of congenital myopathies exists; many present clinically in the neonatal period with hypotonia, hyporeflexia, facial paralysis, ptosis, and a high-arched palate. The etiologies include nemaline myopathy, myotubular myopathy, and mitochondrial dysfunction, and more may be isolated as neuropathologic techniques continue to advance. Diagnosis is made by muscle biopsy.

Benign Congenital Hypotonia

Benign congenital hypotonia is a diagnosis of exclusion. In these cases, all laboratory studies are normal. Hypotonia is present, but weakness and developmental delay usually are absent, and intelligence is normal. Reflexes are normal or mildly hypoactive. The hypotonia usually persists into adult life. One complication is that joint hypermobility in these patients may lead to frequent dislocations in their adult years.

*Laurence Stiefel, MD
Elmhurst Hospital/
Mount Sinai Medical Center
New York, NY*

Hypotonia in Infants
Laurence Stiefel
Pediatrics in Review 1996;17;104
DOI: 10.1542/pir.17-3-104

Updated Information & Services

including high resolution figures, can be found at:
<http://pedsinreview.aappublications.org/content/17/3/104>

Permissions & Licensing

Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:
</site/misc/Permissions.xhtml>

Reprints

Information about ordering reprints can be found online:
</site/misc/reprints.xhtml>

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™

