Neonatal Hypotonia

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KEYWORDS

• Hypotonia • Congenital • Weakness • Muscular dystrophy • Myotonia • Syndrome

KEY POINTS

- Determining the cause of hypotonia requires a detailed history and physical examination aided by diagnostic and laboratory evaluations.
- Medical genetic and neurologic consultations will help establish the diagnostic etiology of neonatal hypotonia.
- Advances in multidisciplinary medical care, home health care, and respiratory and nutritional support have led to longer lives at home for patients with severe hypotonia.

INTRODUCTION

Hypotonia, or low muscle tone, is defined by decreased resistance to passive movement, and may or may not be associated with decreased muscle strength or weakness. Recognition of hypotonia in the newborn may be straightforward, but determining the cause may be a challenge. The physical examination, including a detailed neurologic examination, is important in localizing the site of a defect within the nervous system (ie, central vs peripheral). History along with basic laboratory testing and imaging aids in the differential diagnosis. Identification of the cause is essential for determining the prognosis for the infant, associated morbidities, and the recurrence risk. Some disorders may have a specific treatment; however, the prevailing therapeutic modality comprises physical, occupational, speech/feeding, and respiratory therapy.[1–5]

CLINICAL PRESENTATION

A newborn term infant presents with poor respiratory effort and abnormal suck and swallow after a pregnancy complicated by decreased fetal movement. The baby

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was born by cesarean section because of breech presentation. Physical examination shows no dysmorphic features but significant hypotonia, with tongue fasciculations and absent deep tendon reflexes. The family history is negative for any muscle or neurologic disease, early infant deaths, or consanguinity. Examination of the mother is normal (specifically evaluating for signs of myotonia). What are the approaches to aid in the diagnosis and management of this infant? **Table 1** outlines the differential diagnosis and various evaluations that can be used in the diagnostic process.

DIAGNOSTIC ALGORITHM

Several approaches to a diagnostic algorithm for neonatal hypotonia have been proposed, consisting mainly of either a tree-structured approach based on the distinction between central and peripheral hypotonia[1,4–6] or a sequential method considering successively available tests according to their diagnostic yield.[7] Laugel and colleagues[8] proposed a combined approach involving the following:

1. History and physical examination
2. Neuroimaging/evaluation for congenital malformations
3. Medical genetic and neurogenetic evaluations (Oxford Medical Databases)
4. Karyotype, fluorescence in situ hybridization (FISH), microarray analysis
5. Biochemical evaluation (ammonia, lactate, amino acids, organic acids, very long chain fatty acids, carnitine/acylcarnitine profile, 7-dehydrocholesterol, transferrin isoelectric focusing, *N*- and *O*-glycan analysis, galactose-1-phosphate)
6. Muscle and nerve investigations (electromyography [EMG], nerve conduction velocity [NCV], muscle biopsy)
7. Other specific genetic testing

DIAGNOSTIC EVALUATION

Clinical evaluation of tone may be difficult in infants. Quantifiable measures, such as the Ashworth scales, may be useful in older children with serial measures, but reliability is questionable, especially in infants.[9] There are 4 standard maneuvers that can be used in assessing tone, particularly low tone, in infants. (1) "Pull to sit," whereby the infant is pulled by the arms from a supine to sitting position and a significant head lag is observed in the hypotonic infant. This reaction is also known as the traction response of the infant, whereby typically the infant attempts to counter this maneuver by flexion of the arms. (2) The "scarf sign," whereby an infant's arm is pulled across the chest to the opposite shoulder and there is minimal resistance, and the arm appears like a scarf across the hypotonic baby. (3) The hypotonic infant held under the arms requires significant support or the baby would slip through the evaluator's arms when held in "shoulder/axillary suspension." (4) "Vertical suspension," whereby the baby is suspended prone by the evaluator's hand under the abdomen/chest and the response of the baby's trunk, neck, and extremities are evaluated. Typically the baby will straighten the back, flex the limbs, and intermittently attempt to hold the head straight, whereas the hypotonic baby droops over the evaluator's palm loosely in the shape of a "U" with arms and legs dangling.[10]

Localization of hypotonia to central (ie, the brain and brainstem, either diffusely or focally) or peripheral (any component of the motor unit: anterior horn cell, peripheral nerve, neuromuscular junction, muscle itself) causes helps to further determine the cause of the hypotonia (**Table 2**).[11]