A diagnostic approach to hypotonia in infancy

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Arrest of normal development of muscle tone during gestation and infancy from many causes leads to hypotonia. This may result from malfunction of neuromuscular system, a central nervous system disorder, a metabolic defect or be incidental in other diseases. Some of these conditions are relatively benign whereas others progress relentlessly. A distinction between these is usually possible and a combination of clinical electromyographic and pathological features is sometimes necessary for accurate diagnosis and prognosis.

Hypotonia is the most frequent abnormality of muscle tone observed in infancy. Its causes are diverse. Some of these hypotonic children have marked weakness whereas others do not have any significant disease of neuromuscular system. Evaluation of these children is obviously directed towards establishing a firm diagnosis and thus giving a prognosis.

The main clinical features associated with hypotonia are (1) bizarre and unusual postures; (2) diminished resistance of the joints to passive movements and (3) increased range of joint movements. These children thus present with paucity of active movements in newborn period and delayed motor achievement in the toddler age group. In later childhood previously attained motor ability may be lost.

Hypotonia may be seen in general neurological disorders with damage to upper motor neurones, be associated with severe mental retardation, be a manifestation of a connective tissue disorder, or a feature of various metabolic disorders. No attempt is made here to give a comprehensive list of various diseases (see Florence et al, and Dubowitz, 1) or discuss the neurophysiological basis of various symptoms and signs (see Myers 3).

The clinical evaluation of muscle tone is hampered by lack of objective tests and the examination though fairly reliable is mainly subjective, depending largely on clinician's experience. In general it is useful to follow the usual norms of any clinical examination i.e. history taking, physical examination followed by laboratory tests.

Clinical examination

In children below the age of two the preceding history is usually brief. There may have been decreased fetal movements, suggestion of prenatal or natal insults, or suggestion of hypoxic damage. Normal, motor milestones may or may not have been attained. Failure is generally associated with some degree of muscle weakness.
Family history often gives unexpected clues and it must be remembered that parents may be affected and yet unsymptomatic.

Examination of a child with hypotonia greatly depends upon the age. Resting posture and spontaneous movements need to be observed in an infant with limited ability to co-operate. A child with cerebral hypotonia frequently shows good spontaneous movements against gravity. Ventral suspension and traction on hands in supine position are the two most important positions for assessment of hypotonia irrespective of its cause. The distribution of hypotonia is essential to determine. Distal hypotonia (limb muscles) generally results from an interruption of the peripheral reflex arc. In contrast, axial hypotonia (trunk and limb girdle) suggests a lesion of central nervous system which is established when hypotonia is combined with brisk tendon reflexes. A fluctuation in muscle tone from time to time is occasionally seen in these children. This difference between distal and axial hypotonia is particularly important in older children. Hypotonia limited to a single limb is usually due to a plexus lesion and that affecting all muscles to an equal degree likely to be secondary to metabolic lesions or interference with neuromuscular transmission.

Additional clinical features may help in localizing the site of pathology. Fasciculations in muscles, especially in tongue (not to be confused with normal tremulous movements in an infant) are seen in anterior horn cell disorders and may suggest Werdnig-Hoffmann's disease. Long thin face, transverse smile and high arched palate are seen in congenital myopathies. Wasting of muscles is most likely to be due to a neuromuscular disorders. Changes in deep tendon jerks often reflect the site of lesion.

It is essential to separate muscular weakness from muscular hypotonia. Strength may be normal in cerebellar disease where tone may be markedly diminished. The formal examination of muscle strength however, is not always possible and an uncooperative infant's strength has to be inferred from its functional ability.

Preliminary history taking and examination sometimes result in an established diagnosis eg. muscular dystrophy. In other cases one can at best attempt to locate the anatomic site of maximal pathology. It is easy enough to diagnose a central affection in presence of mental retardation, brisk tendon jerks and focal neurological deficits but localization of disease even to various levels of the motor unit is often not possible at first visit. However, it is helpful to subdivide cases into various broad groups at first instance.

Recent advances have blurred the strict differenciation between a neurogenic and a myopathic illness but some generalizations are available. Few myopathies produce distal weakness and major brunt of a neuopathic illness is borne by distal muscles. One must remember however, that spinal muscular atrophies particularly juvenile varieties are proximal in distribution and myotonic and scapuloperoneal dystrophies sometimes distal. Neurogenic illnesses cause early wasting, fasciculations especially when abundant suggest denervation and presence of contractures is more likely to be due to a myopathic illness. If the degree of weakness is out of proportion to the degree of wasting, the illness is probably myopathic. Both