Evaluation of the floppy infant

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Abstract

This review outlines a clinical approach to the evaluation of the floppy infant. Attention is drawn to the varied manner in which the condition can present, and emphasis is placed upon a detailed assessment of characteristic clinical findings. A distinction is drawn between central and peripheral causes for hypotonia. Guidance is given regarding the importance of evaluating the child for signs of weakness, which is an important marker of neuromuscular pathology. Reference is made to situations where peripheral pathology may mimic central disorders. A diagnostic algorithm is outlined for the investigation of neuromuscular disorders, and reference is made to the discrepancy in findings that often exists between electromyography and muscle biopsy findings. Attention is drawn to available newer diagnostic and therapeutic options, as well as the importance of addressing ethical issues, which become of particular importance once a diagnosis is reached. In practice points, the emphasis has been given to first line investigations based on clinical clues.

Keywords hypotonia; neuromuscular; neuropathy

What's new?

- Recently, array CGH (Comparative Genomic Hybridization) has come up as a powerful diagnostic tool having detection rate of 5—17% in cases of normal karyotype.
- Exon sequencing is promising in detecting causal genetic variants of rare monogenic disorders.
- Newborn screening using tandem mass spectrometry can be helpful for metabolic disorders.
- Recent advances in genetics have uncovered new conditions causing hypotonia and weakness such as congenital myasthenic syndromes and spinal muscular atrophy variants.
- Some of these advances have allowed for specific therapeutic interventions, e.g. use of acetylcholinesterase inhibitors, 3,4-diaminopyridine and ephedrine,salbutamol or fluoxetine in some congenital myasthenic syndromes.
- Antisense oligonucleotide therapy has been under trial for Duchenne muscular dystrophy.
- Enzyme replacement therapy (myozyme) for infantile Pompe disease.

Introduction and definition

The word 'floppy' can be used to mean:

- decrease in muscle tone (hypotonia);
- decrease in muscle power (weakness);
- ligamentous laxity and increased range of joint mobility.

Strictly speaking, the term 'floppy' should be used to describe hypotonia. The interconnection between tone, muscle strength and joint mobility can be appreciated through a consideration of the definition of tone — the resistance to passive movement around a joint. Phasic tone is assessed by the response of the muscle to a rapid stretch, illustrated classically by a tendon reflex, whilst postural tone is measured by the response of the muscle to a sustained low-intensity stretch, as illustrated by the body's ability to maintain posture against the force of gravity.

Clinical appearance

Some features are common to all floppy infants regardless of the aetiology and location of the abnormality. A child is generally said to be floppy if he/she assumes a frog-like posture, is unable to maintain normal posture against gravity, exhibits diminished resistance to passive movements and has an excessive range of joint mobility. Table 1 lists some of the clinical signs with which a floppy infant may present; these features may or may not coexist in the same infant.

Common modes of presentation

The clinical consequences of hypotonia and/or weakness may be evident even in antenatal life. Specific questions in the history should address whether fetal movements were normal, as well as whether there was evidence of polyhydramnios .In the neonatal period, the manner of presentation depends on the severity of the condition. This ranges from the consequences of fetal immobilization, such as hip dislocation, arthrogryposis, talipes and flexion deformity of all limbs, to respiratory and feeding

Clinical signs in a floppy infant

- Observation of a 'frog-leg' posture. This generally implies reduced spontaneous movement, with the legs fully abducted and arms lying beside the body either extended or flexed
- Significant head lag on traction or pull-to-sit manoeuvre and excessively rounded back when sitting (>33 weeks)
- Rag-doll posture on ventral suspension
- Vertical suspension test — feeling of 'slipping through the hands' when the infant is held under the arms
- Various associated examination findings such as flat occiput or congenital dislocation of the hips, arthrogryposis, paradoxical breathing

Table 1

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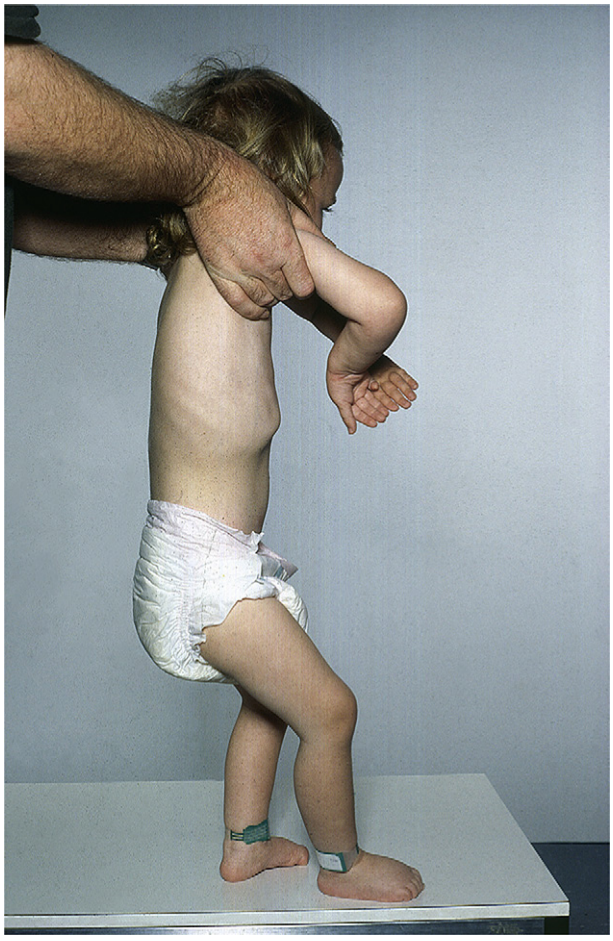

Figure 1 Head lag.

difficulties (slow feeding, recurrent choking or aspiration episodes). Later in infancy, hypotonia may be more obvious once delayed achievement of motor milestones becomes evident, with or without accompanying delay in other areas of development.

Clinical confirmation of hypotonia

Once the suspicion of hypotonia has been raised, the evaluation of the floppy infant should proceed by searching for those clinical signs that corroborate the diagnosis (Figures 1 and 2).

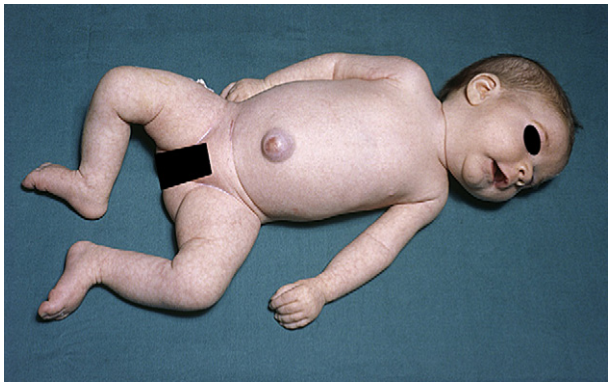

Figure 2 Classic posture in hypotonia.

Clinical features suggestive of hypotonia of central origin

- Social and cognitive impairment in addition to motor delay
- Dysmorphic features implying a syndrome or other organ malformations sometimes implying a syndrome
- Fisting of hands
- Normal or brisk tendon reflexes
- Features of pseudobulbar palsy, brisk jaw jerk, crossed adductor response or scissoring on vertical suspension
- Features that may suggest an underlying spinal dysraphism
- History suggestive of hypoxic-ischaemic encephalopathy, birth trauma or symptomatic hypoglycaemia
- Seizures

Table 2

Important diagnostic clues on examination

- Skin pallor, bruising, petechiae, or evidence of trauma — traumatic myelopathy
- Abnormalities of respiratory rate, pattern, or diaphragmatic movement — congenital myopathies
- Cardiomyopathy — consider *carnitine* deficiency, fatty acid oxidation disorders ,acid maltase deficiency, Pompe's disease
- Hepatosplenomegaly — storage disorders or congenital infections
- Renal cysts, liver dysfunctions, high forehead and wide fontanelles — Zellweger's spectrum disorder
- Congenital cataracts, glaucoma — Oculocerebrorenal (Lowe) syndrome
- Abnormal urine odour — metabolic disorders
- Hypopigmentation, undescended testes — Prader–Willi syndrome
- Abnormal fad pad and inverted nipples — congenital disorders of glycosylation (CDG).

Indicators of peripheral hypotonia

- Delay in motor milestones with relative normality of social and cognitive development
- Family history of neuromuscular disorders/maternal myotonia
- Reduced or absent spontaneous antigravity movements, reduced or absent deep tendon jerks and increased range of joint mobility
- Frog-leg posture or 'jug-handle' posture of arms in association with marked paucity of spontaneous movement
- Myopathic facies (open mouth with tented upper lip, poor lip seal when sucking, lack of facial expression, ptosis and restricted ocular movements)
- Muscle fasciculation (rarely seen but of diagnostic importance when recognized)
- Other corroborative evidence including muscle atrophy, muscle hypertrophy and absent or depressed deep tendon reflexes

Table 3