Benign Congenital Hypotonia

Synonym: congenital hypotonia with favourable outcome (CHFO)

The diagnosis of benign congenital hypotonia (BCH) is now a controversial one.[1] Since the first description in 1956 it has been possible to diagnose children more accurately who would otherwise have received this diagnosis.[2]

However, the term 'benign congenital hypotonia' is still used to describe children with mild hypotonia who appear to have a favourable outcome and in whom no other diagnosis can at this stage be made.

Diagnostic criteria

The criteria initially suggested for the diagnosis are:[3]

- Early hypotonia - usually since birth.
- Active movements of the limbs and normal tendon reflexes.
- Normal or mild motor retardation that improves with age.
- Normal levels of muscle enzymes.
- Normal results of electromyography (EMG) and nerve conduction studies.[4]
- Normal muscle biopsy studies.

Incidence

- The true incidence of BCH is not known but is less than first thought, as many have now received other diagnoses.
- There is a familial tendency with approximately 30% of affected children having some family history.
- Boys and girls appear to be equally affected.

Aetiology

The underlying pathology for all hypotonias can be divided into four categories:

- The central nervous system (CNS) - 66-88%. [5]
- The peripheral nerves (motor and sensory).
- The neuromuscular junction.
- The muscle.

Some congenital disorders have both central and peripheral origins to the hypotonia.

Presentation

The children present at birth as 'floppy babies' with generalised hypotonia. They may also have any combination of the following features:

- Accentuation of the spinal curve - eg, hyperlordosis.
- Abdominal protrusion.
- Flat feet when standing.
- Pes cavus - when not weight-bearing.
- Walking on tip toe,
- Inability to walk on heels (common).
- Developmental delay, ie failing to meet gross motor milestones for sitting, standing and walking.
- ± Muscle contractures (not before the age of 8 years).
- Joint hyperlaxity.
- Recurrent episodes of myalgia when using muscles.

A full history should be taken from the parents of any child with hypotonia, looking for evidence of:

- Oligohydramnios or polyhydramnios.
- Birth trauma.
- Family history of hypotonia (found in 46%).[5]
- Perinatal asphyxia.
- Infection and/or drugs taken by the mother during pregnancy.
- Mother's description of fetal movements.
Examination

This should include: [6]

- Head circumference (central hypotonia more likely to have microcephaly).
- Developmental assessment.
- Evaluation of muscle tone.
- Reflexes.
- Resting postures in prone and supine.
- Pull-to-sit.
- Antigravity movements.
- Visual following/alertness.

Infants with hypotonia of peripheral origin (eg, congenital muscular dystrophies, spinal muscular atrophy or congenital myasthenic syndrome) also often have joint contractures.

Differential diagnosis [2]

Differentiation of central from peripheral origins of hypotonia should be made to aid diagnosis. [5]

Other causes of hypotonia must be excluded. These include:

- Congenital myotonic dystrophy.
- Nemaline myopathy: congenital myopathy characterised by abnormal rod-like structures in muscle fibres, on histological examination. [7]
- Metabolic disorders - eg, Zellweger's syndrome.
- Prader-Willi syndrome.
- Werdnig-Hoffman disease.
- Down's syndrome.
- Infantile botulism.

Investigations

These may include:

- Infection screen - including CSF and blood culture.
- Blood tests - eg, for glucose, magnesium, creatine kinase.
- Karyotyping.
- CT/MRI scan.
- Muscle biopsy.
EMG:
- Normal EMG examination helps investigators to exclude several neurological diseases characterised by hypotonia.
- It can provide valuable information to confirm the clinical diagnosis of BCH. [4]

- Nerve conduction studies.

Management

General measures
Although no drug therapy is currently available for the treatment of BCH, children benefit from treatment with physiotherapy to help both active and passive movements in order to optimise muscle strength and prevent the development of any shortening of the muscles. [3]

Prognosis
Parents will benefit from reassurance that the disorder is usually self-limiting, is not associated with any intellectual deficit and disappears by puberty in the majority of cases. [3]

Further reading & references
7. Nemaline Myopathy 3, NEM3; Online Mendelian Inheritance in Man (OMIM)

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<th>Document ID:</th>
<th>Last Checked:</th>
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<tr>
<td>1853 (v24)</td>
<td>19/11/2015</td>
<td>17/11/2020</td>
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