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Review article

Neuromuscular diseases: Diagnosis and management

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ABSTRACT

Neuromuscular diseases (NMDs) affect the peripheral nervous system, which includes the motor neurons and sensory neurons; the muscle itself; or the neuromuscular junction. Thus, the term NMDs encompasses a vast array of different syndromes. Some of these syndromes are of direct relevance to paediatric orthopaedic surgeons, either because the presenting manifestation is a functional sign (e.g., toe-walking) or deformity (e.g., pes cavus or scoliosis) suggesting a need for orthopaedic attention or because orthopaedic abnormalities requiring treatment develop during the course of a known NMD. The main NMDs relevant to the orthopaedic surgeon are infantile spinal muscular atrophy (a motor neuron disease), peripheral neuropathies (chiefly, Charcot-Marie-Tooth disease), congenital muscular dystrophies, progressive muscular dystrophies, and Steinert myotonic dystrophy (or myotonic dystrophy type 1). Muscle weakness is a symptom shared by all these conditions. The paediatric orthopaedic surgeon must be familiar, not only with the musculoskeletal system, but also with many other domains (particularly respiratory and cardiac function and nutrition) that may interfere with the treatment and require preoperative management. Good knowledge of the natural history of each NMD is essential to ensure optimal timing of the therapeutic interventions, which must be performed under the best possible conditions in these usually frail patients. Timing is particularly crucial for the treatment of spinal deformities due to paraspinal muscle hypotonia during growth: depending on the disease and natural history, the treatment may involve non-operative methods or growing rods, followed by spinal fusion. A multidisciplinary approach is always required. Finally, the survival gains achieved in recent years increasingly require attention to preparing for adult life, to orthopaedic problems requiring treatment before the patient leaves the paediatric environment, and to the transition towards the adult healthcare system.

1. Introduction

Neuromuscular diseases (NMDs) clearly fall within the purview of paediatric orthopaedics. The nature of the presenting manifestation may lead the patient to the paediatric orthopaedic surgeon before any other physicians are consulted. Surgeons must therefore be trained to detect the clinical, apparently orthopaedic, signs suggesting NMD and to perform the initial work-up. NMDs are usually progressive and very often cause postural abnormalities and deformities. Surgical decisions must be guided by in-depth knowledge of the NMD and its natural history. As with all surgical procedures, the goal is not only to correct an anatomical abnormality but also to determine the desired benefits in terms of musculoskeletal or respiratory function. Surgery is only very rarely performed on an emergency basis. The surgeon must therefore ensure optimal preparation of the patient to minimise the operative risks. Finally, the survival gains achieved in recent years increasingly require attention to prospects for adult life and to orthopaedic problems requiring treatment before the patient leaves the paediatric environment. Readers can find details on the nosology and treatment of NMDs in the SOFOP treatise on NMDs (see References).

2. Clinical findings that should suggest neuromuscular disease (NMD)

2.1. Before birth

The paediatric orthopaedic surgeon may be asked for advice when a prenatal sonogram shows a clubfoot or clubhand deformity or hip dislocation. NMD is among the diagnoses suggested by the absence of foetal movements. The mother should then be referred to a prenatal diagnosis centre for a detailed aetiological work-up, whose results may prompt an offer to terminate the pregnancy.

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2.2. At birth

The neonate with peripheral paralytic hypotonia has no spontaneous movements. Elicited movements are scarce and occur only along the plane of the bed. In contrast, eye contact is good. Peripheral hypotonia combined with orthopaedic deformities suggest Steinert's disease or myotonic dystrophy type 1 (DM1), a congenital myopathy (with involvement of the facial muscles and often of the respiratory muscles), congenital muscular dystrophy (usually responsible for contractures), motor neuron disease (infantile spinal muscular atrophy), or arthrogryposis (if joint motion is limited). Patients with these manifestations should be referred promptly to the neuropaediatrician for a detailed diagnostic evaluation. This referral should not delay the symptomatic management of the orthopaedic abnormalities.

2.3. From birth to 2 years of age

At this age, the predominant symptom remains diffuse hypotonia affecting not only the torso but also the limbs (whereas central neuromuscular diseases are characterised by peripheral spasticity). Muscle strength is decreased overall, leading to difficulty with posture changes and a paucity of spontaneous gestures. The gaze is attentive and the child is interested in proffered objects but does not seek to grasp them. The muscle-wasting may be difficult to detect. Peripheral hypotonia combined with cranial nerve involvement (e.g., paucity of facial expressions, limited ocular tracking, or swallowing disorders) strongly suggests NMD. Osteo-articular deformities may be either congenital or recent and progressive and may affect the feet (pes planus, foot stiffness, medial pes cavus, pes equinus, etc.) or spine (kyphosis due to spinal collapse or spinal stiffness).

At this age, two other broad groups of NMDs produce similar presentations:

- central neurological disorders (e.g., cerebral palsy): the history, impaired contact, and peripheral spasticity assist in the diagnosis;
- inherited collagen disorders (Marfan, Ehlers–Danlos, osteogenesis imperfecta): in addition to neurological and orthopaedic signs, these diseases cause suggestive abnormalities of other systems (cardiac, ocular, cutaneous).

In some cases, the diagnosis is established only based on the course of the clinical manifestations over time.

Delayed walking is another symptom that may prompt a visit to the paediatric orthopaedic surgeon and lead to a diagnosis of NMD. The physical findings are strongly suggestive in the vast majority of cases. In doubtful cases, advice from a specialist or a second evaluation 3–4 months later is desirable.

2.4. After 2 years of age

After 2 years of age, the orthopaedic surgeon is very often the first in line to diagnose NMD [1]. The main manifestation is muscle weakness, with its consequences on bones and joints. The expression of muscle weakness varies widely, although abnormal fatigability predominates. The parents seek medical attention because of frequent falls, difficulty climbing stairs, or an inability to run or stand from the sitting position on the floor. Gowers' manoeuvre consists in asking the child to stand from the sitting position on the floor. It is positive if the child must push with the hands on the tops of the thighs to stand, due to weakness of the quadriceps. Weakness of the upper limb muscles results in difficulty performing activities with the arms raised and carrying heavy loads. Each sign considered alone may simply indicate excessive anxiety on the part of the parents. When the physical findings are reassuring, the patient should nevertheless be re-evaluated a few months later to allow an assessment of the time-course of the symptoms.

The muscle weakness may be overshadowed by the progressive orthopaedic abnormalities that it induces. An example is recent and worsening toe-walking, which indicates contracture of the triceps surae. Patients with Duchenne muscular dystrophy (DMD) also have enlarged calves, due to fatty infiltration of the triceps surae. Progressive pes cavus, pes planus due to muscle weakness or an inability to walk on the heels with recruitment of the toe extensors suggests a peripheral neuropathy.

Most NMDs are progressive, and patient follow-up is therefore essential.

3. Suspected neuromuscular disease (NMD): practical course of action – initial work-up

When a family seeks advice for what they believe is a purely orthopaedic, mechanical problem, raising the possibility of an NMD requires sensitivity. A cautious approach, in which the parents are told that some diseases must be ruled out on principle, is in order. Ideally, the patient should then be managed chiefly by a neuropaediatrician. Until this can be arranged, however, a useful measure consists in obtaining a few first-line investigations, such as muscle enzyme assays and an electromyogram (EMG). The EMG prescription should include a specific question (e.g., presence of a neuropathy or of a neuromuscular conduction block). When these investigations are prescribed, the surgeon must be capable of explaining their objective to the parents. The search for a cause should not delay the non-operative treatment of the orthopaedic abnormalities (e.g., via rehabilitation therapy or orthoses). In contrast, no surgical procedures should be performed until the diagnosis is established.

3.1. Muscle enzymes

Creatine kinases (CK) are enzymes found chiefly in the skeletal muscle and myocardium. In neonates, the plasma CK level is normally below 200 IU/L. An increase to at least 3 times the upper limit of normal (in some cases, to more than 1000 IU/L) suggests a congenital muscular dystrophy or infantile spinal muscular atrophy [2]. In boys, the main diagnosis to consider is a dystrophinopathy [DMD or Becker muscular dystrophy (BMD)], in which, the plasma CK levels are at least 50-fold the upper limit of normal. In general, CK elevation to more than 3 times the upper limit of normal requires referral of the patient to a neuropaediatrician, who will determine whether genetic testing and/or a muscle biopsy are in order.

3.2. Electromyogram (EMG) with nerve conduction velocity measurements

This investigation is performed in a paediatric department. The muscle response to electrical nerve stimulation or to a voluntary contraction is tested, and conduction velocities within the nerves are measured [1]. The results distinguish between an abnormal muscle response (myopathic pattern) and loss of motor nerve fibres (neuropathic or denervation pattern), i.e., between diseases involving only the muscle and motor neuron diseases. In combination with nerve conduction velocity measurements, the EMG helps to separate pure motor neuropathies, demyelinating sensorimotor neuropathies (marked prolongation of latency and very low conduction velocities in both motor and sensory nerves), and axonal sensorimotor neuropathies (decreased response with normal latency and conduction velocities).

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