Management of respiratory disease in children with muscular weakness

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Abstract

Children with chronic muscular weakness from any cause are prone to develop varying degrees and patterns of respiratory muscle weakness. This leads to ineffective cough, atelectasis, pneumonia, restrictive pulmonary disease and eventually respiratory failure. Over the past 20 years there have been improvements in clinical care of children with muscular weakness, including improved monitoring of lung function and hypoventilation during sleep, coordinated multidisciplinary respiratory care, physiotherapy and introduction of non-invasive ventilation. This article reviews the current knowledge base and the evidence for management of children and provides practical advice for paediatricians.

Keywords cerebral palsy; children; neuromuscular disease; respiratory

Introduction

Children with muscle weakness include a large and heterogeneous group of patients, including those with neuromuscular disease (NMD), cerebral palsy or developmental delay, spinal injury and other rarer conditions. Muscle weakness often has a very significant effect on respiratory health and as a result these children require regular structured assessments.

NMD has a prevalence of 1 in 3000 in the UK population. Although, respiratory failure is the most common cause of death in this group, several factors, including the age of onset, disease severity and the rate of disease progression may vary for each individual. There has been considerable progress in medical care which has increased both duration and quality of life in these patients. Notable recent improvements include adjuncts to assist airway clearance, and the prompt evaluation of respiratory function by multidisciplinary teams.

Cerebral palsy (CP) is more common in the population, with a prevalence of approximately 2 per 1000 children. It is associated with prematurity and may be accompanied with concomitant developmental delay. Although CP and developmental delay affect the muscles and the airway protection in a different manner to NMD, the impact on respiratory health is similar. As with NMD, respiratory factors are the leading cause of death among CP patients, and careful consideration of respiratory care can improve outcomes as well. This article will predominately focus on patients with NMD, but will also reflect on the problems seen in CP. Muscle weakness caused by rare cases including spinal cord injury or Guillain–Barre syndrome will not be discussed specifically, although the principles that are described here remain the same.

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Pathophysiology, risk factors and problems

The range of presentations seen with NMD is wide, and it is beyond the scope of this article to review them in detail. However, it is very important to recognise the variability in the age of onset, disease severity and the pattern of disease progression that may occur. A brief summary is shown in Table 1. The most common neuromuscular childhood diseases are Duchenne muscular dystrophy (DMD), spinal muscular atrophy (SMA) and congenital muscular disorders (CMD).

DMD typically presents with an early onset and is often associated with some developmental delay. The disease is progressive, ultimately involving the heart muscle. Scoliosis develops in ~70–90% patients and current life expectancy is in the mid-20s. By contrast, SMA can vary significantly in severity: infants with Type I SMA usually die within two years, but for Type III SMA, life expectancy extends into adulthood. Scoliosis is universal in Types I and II, but there is no cardiac involvement. Children with CMD typically present early and it can be associated with significant weakness. However, progress of this weakness is usually slow or absent, and if present may occur in late childhood.

Despite these differences, the respiratory muscles are rarely spared in neuromuscular diseases even if the type of muscle involvement, severity and time course varies greatly between these three different diseases. Respiratory symptoms are, therefore, common and can occur at several different levels. We have grouped these respiratory complications into three categories: issues with the upper airway (airway protection, swallowing and obstructive sleep problems), ventilation problems (respiratory muscles, lung compliance, gas exchange) and infection.

Upper airway problems

One of the major dangers in children suffering from respiratory weakness is their reduced ability to protect their airways. Coughing, gagging and swallowing reflexes may all be impaired, increasing the risk of aspiration. In order to produce an effective cough, a deep breath (inspiratory muscles) has to be followed by a maximal contraction of the expiratory muscles, with initial glottis closure and then opening (oropharyngeal muscles), which generates an expiratory flow capable of eliminating secretions. All three muscle groups may be weak, leading to reduced airway clearance. Over time, the continuous presence of pooled secretions in the oropharynx may also lead to a reduced cough stimulus. Impairment of the oropharyngeal muscles, in addition to contributing to ineffective cough, will cause phonation and swallowing disorders with a risk of broncho-aspiration that may lead to acute respiratory failure.

Respiratory features of neuromuscular diseases

Condition	Respiratory failure	Secretion clearance difficulty	Recurrent pneumonia	Progression	Disease-specific features
SMA					
Type 1	All by 2 years	Marked	All	Rapid	All require full-time respiratory support
Type 2	~40% in childhood	Early	~25% in first 5 years	Slow	
Type 3	Rare in childhood	Rare in childhood	Rare in childhood	Slow	
SMA with respiratory distress type 1	All by 6 months	Marked	All	Rapid in first year, then slows	All require full-time respiratory support
DMD/severe childhood onset limb-girdle muscular dystrophy	After loss of ambulation	After loss of ambulation	Late		Cardiomyopathy usually occurs after respiratory problems but may precede them
Facioscapulohumeral muscular dystrophy	When onset <20 years	With infantile onset	With infantile onset	Slow	Severe infantile onset type is frequently associated with sensorineural deafness
Congenital muscular dystrophy					
All types	Any age depending on severity	Any age depending on severity	Any age depending on severity	Slow	
Ullrich	70% in adolescence	Mild	Infrequent		Proximal contractures with marked distal laxity
Rigid spine muscular dystrophy	Early while ambulation preserved	Mild	Infrequent		Hypoventilation may occur in ambulant children with relatively preserved vital capacity
Congenital myopathy					
Central core	Uncommon except in severe recessive type	Uncommon	Uncommon	Slow	Susceptible to malignant hyperthermia
Minicore	Early while ambulation preserved				
Nemaline	Early in severe neonatal form, mild later onset form may develop early while ambulation preserved	In severe form	In severe form	Slow	
Myotubular	85% in serve X-linked form	In severe form	In severe form	Slow	Ophthalmoplegia, rare coagulopathy and liver haemorrhage
Fiber type disproportion	Depends on genotype	Uncommon	Uncommon		
Myotonic dystrophy					
Myotonic dystrophy 1	Common in severe congenital onset, usually improves	Common in severe congenital onset	Common in severe congenital onset	Initial improvement, later slow deterioration	Prominent learning difficulty, somnolence, central hypoventilation
Myotonic dystrophy 2	Uncommon	Uncommon	Uncommon		
Congenital myasthenic syndromes	Often in neonatal period, may occur during inter-current illnesses	Especially during inter-current illnesses	Possible if weakness severe and persistent		Weakness may fluctuate, episodic apnoea in some. Congenital stridor in those with DOK7 mutations