AUTHORS NOTE: The following respiratory advice on scoliosis management in patients with congenital myopathy is based on two papers, due to be published soon. Once published, updates will be posted here.

Myotubular Trust.

RESPIRATORY CARE ADVICE : SCOLIOSIS
For patients with myotubular and centronuclear myopathy and other congenital myopathies.

What are Congenital Myopathies?

Congenital myopathies are a group of rare neuromuscular disorders that more often present at birth or in early childhood, and which vary widely in how they present and manifest. Myotubular Myopathy and Centronuclear Myopathy is one of a number of conditions which come under the umbrella term "Congenital Myopathies". "Congenital" means present from birth, and "Myopathy" means muscle condition or weakness.

What is Scoliosis?

All spines have curves, and some curvature in the neck, upper trunk and lower trunk is normal. Humans need these spinal curves to help the upper body maintain proper balance and alignment over the pelvis. When there are abnormal side-to-side (lateral) curves in the spinal column, this is referred to as Scoliosis.

Scoliosis is often cosmetically unacceptable and makes sitting comfortably difficult due to pelvic obliquity and trunkal deformity. It can also worsen the ability to walk, and frequently becomes painful. Critically, scoliosis can compromise respiratory function.

Evaluating Scoliosis in Congenital Myopathy

Scoliosis in patients with Congenital Myopathy is not uncommon. All patients should be regularly checked for signs of spine curvature by their doctors during routine neuromuscular clinic appointments. Patients should be referred to an orthopaedic surgeon by the neuromuscular doctors at the first sign of any increase in curvature, and also to a doctor with an interest in pulmonary function, if not already being screened for breathing problems. Lung function tests will help identify a patient's ability to cope with respiratory infection, aspiration, and sleep-disordered breathing which may become more of an issue as the curve of the spine increases.

There are several different signs that a patient can be aware of themselves, to help determine whether they actually have scoliosis. If one or more of the following signs are present, it is advisable to schedule an exam with a doctor.

- Shoulders are different heights—one shoulder blade is more prominent than the other
- Head is not centered directly above the pelvis
- Appearance of a raised, prominent hip
- Rib cages are at different heights
- Uneven waist (more prominent crease on one side)
- Changes in look or texture of skin overlying the spine (dimples, hairy patches, color change)
- Leaning of entire body to one side and loss of balance when sitting

It is important that when evaluating a patient for surgery, the orthopaedic surgical team adopts a multidisciplinary approach. This means that the patient's individual needs and issues should be reviewed and assessed by a team of other experts too, which include neuromuscular, respiratory, cardiac, physiotherapy, speech and language, occupational therapy and dietary experts. These evaluations will help guide the patient on the optimal time to have surgery, what type of surgical

procedure is the best option for them and help manage the period following surgery. In planning for the actual surgery, it is vital that a senior anaesthetist is a member of the multi-disciplinary team from the outset, as the patient's breathing support both during and after the surgery will be very different to the needs of a patient without a neuromuscular condition. In particular the need for, and amount of muscle relaxant during surgery, is a critical decision.

The primary consideration when planning surgery for children with scoliosis associated with neuromuscular weakness should be comfort and quality of life. The primary goals of surgery in children with neuromuscular weakness is to control the progression of the curve, to balance the spine so that head is centrally placed over the pelvis, that standing posture is near perfect as possible and to protect the chest wall to help maximise respiratory function.

Chest wall deformities can be often be corrected during spinal surgery which may help to maximise respiratory function too.

Please also refer to the December 2011 Congenital Neuromuscular Disorders Scoliosis Webinar for further advice from David P Roye Jr MD, Professor of Orthopaedic Surgery, New York Presbytarian Hospital, USA. http://www.youtube.com/watch?v=ZBK8lo0Fz7M&feature=youtu.be

How Scoliosis Can Affect Breathing

Worsening scoliosis, through the increasing sideways curvature of the spine, alters the shape of the thoracic cage and diaphragm, limiting vital capacity by causing asymmetric inspiration and decreased chest wall compliance. The extent to which lung volume is reduced is variable; lung hypoplasia may occur in children with very early scoliosis, but in older children the lungs are likely to have developed normally, and compromise is due to mechanical limitations. Physiological dead space is increased, with a decreased tidal volume to dead space ratio, increasing the risk of carbon dioxide retention.

Regular pulmonary and cardiac assessment will help the doctor plan for and put in place any preventative measures to help maintain good respiratory function and where possible try to improve a patient's breathing efficiency in preparation for any surgery.

Doctors and patients should seriously consider and weigh up the pros and cons of wearing a spinal brace, as this can potentially have a negative effect on a patient's respiratory function. There is limited evidence to suggest that bracing actually has an impact on delaying or preventing severe curvature.

Planning Where Surgery Should Take Place

Surgery in children with neuromuscular weakness should take place in units with experienced paediatric surgeons, anaesthetists, and physiotherapists, and where there are facilities for paediatric intensive care and non-invasive ventilation.

Children with neuromuscular weakness who require scoliosis surgery, or indeed any surgical intervention, should be assessed by a multidisciplinary team in the days and months leading up to surgery.

Perioperative Airways Management

Airway management during surgery can sometimes present challenges in patients with congenital myopathies. Pre-operative assessment is recommended to identify those children with neuromuscular weakness who are at risk of an adverse outcome after surgery. Risk factors include children who use daytime and/or night-time bi-pap or c-pap, have a vital capacity less than 60% predicted and have an ineffective cough. The nutritional status of a child also needs to be taken into consideration.

While it's not unusual for patients to require some form of ventilation support post operatively, anaesthesia, even during a minor surgical procedure, can carry risk of long-term respiratory complications and therefore a potential for prolonged respiratory support. Furthermore, respiratory muscle strength is usually weaker in the post-operative period, more often due to the combined

effects of pain and sedation, and can result in hypoventilation and airway secretion retention in this group of patients.

Non-Invasive Positive Pressure Ventilation (NIPPV) is often needed in the perioperative / post-surgical period as a bridge to recovery from anaesthesia and should be anticipated for following spinal fusion procedures, or indeed other surgical procedures. For children who have a vital capacity less than 60% predicted and/or an ineffective cough and/or who already use NIPPV (Bi-level positive airway pressure (BiPAP) or continuous positive airway pressure (CPAP)) other effective airway clearance techniques such as breath stacking with a lung volume recruitment bag or mechanical insufflation/exsufflation (MI-E) should also be considered following surgical procedures.

Malignant hyperthermia (MH) is a concern for myopathy patients undergoing general anaesthesia and unfortunately there is no simple, straightforward test to diagnose the condition. Although believed to be more specifically a problem associated with RYR1-related myopathies, some cases have been recorded in other variants of congenital myopathy too.

Virtually all malignant hyperthermia (MH) cases in the past, have been associated with use of muscle relaxants such as succinylcholine, and inhalational agents such as halothane (but *not* nitrous oxide). In susceptible individuals, these drugs can induce a drastic and uncontrolled increase in skeletal muscle oxidative metabolism, which overwhelms the body's capacity to supply oxygen, remove carbon dioxide, and regulate body temperature, eventually leading to circulatory collapse and death if not treated quickly. A medical alert notice or bracelet detailing the risk of MH is recommended for patients with a known risk, in case of unexpected surgery or accident.

To summarise, new safe guidelines are as follows:

- Anaesthesiologists should consider that malignant hyperthermia is a distinct possibility *in all patients who have a neuromuscular weakness*, instead of trying to predict if a patient is likely to be affected. This is especially important when patients do not have a definitive diagnosis or do not know which gene is implicated in their condition.
- Anaesthesiologists should adopt the ‘clean technique’ using intravenous agents with known high safety profiles and only use anaesthesia machines and equipment which has been specifically cleaned immediately prior to surgery so as to remove all traces of anaesthetic agents used for other patients in previous surgical procedures.
- The patient’s cardiopulmonary parameters are to be closely and extensively monitored during surgery. This may involve the availability of dantrolene, capnography to measure CO2 concentration, and post-operative intensive care support.
- NIPPV should be considered for the periods pre- and post-operatively until the patient is well enough to resume normal breathing behaviour.
- Airways Clearance techniques should be considered to help clear excess secretions in the post-operative phase and taught to the carers and patients by experienced respiratory physiotherapists ideally in the pre-operative phase.

Airways Clearance and Exercise

Most independent airway clearance techniques start with the child taking a deep in-breath followed by a forced out-breath (huff) or cough. The deep breath gets air behind mucus, opens up the airway, loosens secretions in the small airways and mobilises the secretions to the larger airways. The secretions are then cleared from the central airways with an effective huff or cough.

However, independent airway clearance is hard for some children with more severe neuromuscular weakness to do, as they don’t have the ability to take a deep breath nor do they have an effective cough.

Chest physiotherapy techniques

Prior to chest physiotherapy inhaled treatment, including humidification, mucolytics, or hypertonic saline, may be beneficial to help promote secretion clearance. It is worth noting that with any secretion clearance, there is always a potential to mobilise a large volume of secretions and therefore

it is important to ensure that the appropriate emergency equipment (e.g. resuscitation bag and suction) is available in case of mobilising a large mucus plug into a central airway.

Some of the techniques for airway clearance include, and should be discussed with a doctor or physiotherapist with an interest in respiratory function:

- Standard chest percussion, using cupped hand and fingers on the chest
- Postural draining and positioning
- Intrapulmonary percussive ventilation (IPV) – which superimposes high frequency mini bursts of air on the patient's basic inspiratory and expiratory breathing pattern - creating an internal vibration (percussion) within the lungs and promotes secretion clearance
- High frequency chest wall oscillation(HFCWO) - provided by intermittent compression of the chest wall using an inflatable jacket or cuirass adjusted to fit snugly over the thorax
- Incentive spirometry (IS) – devices designed to mimic natural sighing or yawning by encouraging patients to take long, slow deep breaths. This can help increase inspiratory volumes, improve inspiratory muscle performance and re-establish or simulate the normal pattern of pulmonary hyperinflation. This device is often used in the postoperative period
- Augmented cough – this works in two parts a) to increase the volume of breath taken in and b) to increase the forces at which that volume of breath is breathed out (cough or huff). Both of these used together can increase expiratory flow rates during coughing and therefore clear secretions from the lungs. There are many effective ways to achieve an augmented cough, and these can be determined by a pulmonary expert according to the capabilities of the patient. These may include any combination of the following, air-stacking, frog breathing, manually assisted cough or insufflation-exsufflation devices.

Breathing Exercises:

Breathing exercises can play an important role in improving a patient's respiratory strength and endurance leading up to surgery. They can help by preserving or improving daytime and night-time ventilatory capacity and reducing respiratory problems by improving cough efficiency.

A respiratory physiotherapist or nurse can teach a number of exercises using some of the equipment used for airway clearance, or encourage other self-help exercises such as shouting, singing, etc.

Maintaining sitting and ambulation (walking):

Physiotherapy and occupational therapy can help maintain good sitting posture before and after surgery which will really help children who are able to sit maintain better respiratory function. Surgery will often help improve comfort and sitting balance in children.

Although children who can walk before surgery are at risk of losing the ability to walk after surgery due to pelvic fusion; most can continue to walk, at least with equipment after surgery. Standing frames, if safe to use, are excellent to retain good posture and bone strength. While a standing frame is also helpful to maintain good flexibility too (especially in hips and knees) caregivers should also be trained to perform other flexibility exercises as part of a routine daily programme.

Where possible physiotherapy and gentle exercise should be encouraged before surgery and as soon as the patient is pain free and willing to participate, following surgery. This will help maintain good cardiac and pulmonary function as well as aid physical /motor recovery. Prolonged periods of inactivity and without walking can lead to increased atrophy of the muscles, thereby prolonging rehabilitation and in some instances leading to total loss of ambulation.

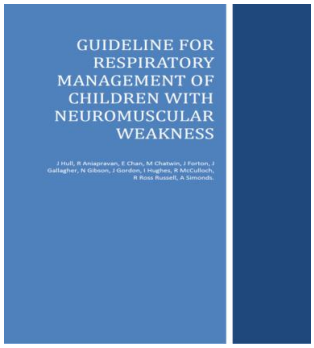
Further information

Please refer to the helpful Scoliosis Questions & Answers which were kindly answered by David P Roye, MD, Professor of Orthopaedic Surgery, New York Presbyterian Hospital, USA see: http://www.myotubulartrust.com/documents/Scoliosis-Webinar-Questions-And-Answers-December-2011.pdf

Read patient stories: http://www.myotubulartrust.com/talk-resources-scoliosis-index.htm

Join the Webinar on Respiratory Management on April 28th 2012 see our website http://www.myotubulartrust.org for more details.

Coming soon! Watch out for the British Thoracic Society Guidelines, 2012



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