orphanaesthesia

Anaesthesia recommendations for patients suffering from

Limb-girdle muscular dystrophy

Disease name: Limb-girdle muscular dystrophy

ICD 10: G.71.0

Synonyms: -

Limb-Girdle muscular dystrophy (LGMD) is a group of genetic muscle diseases characterized pathophysiologically by a mismatch between muscle breakdown and muscle repair, and it has a frequency of 1 in 15000. The main clinical features include proximal muscle weakness involving hip and shoulder girdles, elevated creatine kinase (although serum CK level is not an absolute secreening test for all LGMD) and a wide range of age of onset, from early childhood to late adulthood. Clinical classification is based on the distribution of weakness early in the course and on the age of presentation. It may be difficult to reach a precise diagnosis because of the wide clinical and genetic variability, and the different relative local frequencies of the different forms. The base of the diagnosis includes a clinical thorough evaluation, electrodiagnostic testing, muscle biopsy, imunohistochemistry and genetic analysis. These disorders may be either autosomal recessive (LGMD2) or dominant (LGMD1), and there are a number of different proteins involved, which allow a further classification of LGMD: LGMD 1A (myotilin), LGMD 1B (lamin A/C), LGMD 1C (caveolin 3), LGMD 2A (calpain-3), LGMD 2B (dysferlin), LGMD 2C-F (sarcoglycans), LGMD 2G (telethomin), LGMD 2H (TRIM 32), LGMD 2I (FKRP), LGMD 2J (titin), LGMD2L (anoctamin 5).

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

Find more information on the disease, its centres of reference and patient organisations on Orphanet: www.orpha.net

Disease summary

There are different disorders that must be taken in account in these patients, some of them sharing similar gene involvement; among these disorders we should include nuclear envelopathies (Emery-Dreifuss muscular dystrophy), Bethlem myopathy/Ulrich disease, and milder presentations of dystrophinopathies (Becker muscular dystrophy and women who are manifesting dystrophinopathy gene carriers). Diagnostic evaluation depends on inmunostaining or blotting, although these tests may be difficult to be properly interpreted. Genetic testing is somewhat limited on a practice basis to the research setting, but newer technologies (such as next generation sequencing panels) are actually significantly less expensive than muscle biopsy and provide a definitive diagnosis. The importance of genetic testing must not be overlooked because it may allow genetic counselling and prediction of other organ system involvement, like cardiomyopathy and respiratory failure.

The main issue when performing an anaesthetic procedure in these patients, deals with the risk of cardiac involvement (presence of cardiac myopathy and cardiac arrhythmia in LGMD 1A, 1B, 2C-F, 2I) and respiratory failure (LGMD 1A, 1B, 2C-F, 2I, 2J). In the case of LGMD 1B, pacemaker and implantable cardioverter defibrillator may be needed because of conduction disorders, and regarding LGMD 2I, 50% of the patients in this group may suffer from dilated cardiomyopathy.

We must keep in mind whenever facing a surgical procedure that this patients may have an increased sensitivity to the effect of anaesthetics and neuromuscular blocking agents. Although there is scarce evidence supporting an increased risk of malignant hyperthermia, there is a higher risk of suffering disease related cardiac complications and acute rhabdomyolysis when exposed to inhaled anaesthetics, and succinylcholine should be avoided as in many muscle disease because of being associated to life-threatening hyperkalemia.

Typical surgery

Paediatric procedures (tonsillectomy, adenoid surgery), muscle biopsies, correction of progressive orthopaedic deformities including spinal surgery, cardiac transplantation, cardiac surgery with extracorporeal circulation, caesarean section.

Type of anaesthesia

There is insufficient data to recommend a specific type of anaesthesia; there are previous reports of caesarean sections performed under combined spinal-epidural anaesthesia, as well as different procedures using general anaesthesia and spinal anaesthesia. If general anaesthesia is chosen, inhaled anaesthetics and succinylcholine must be avoided in order to minimize the risk to trigger hyperkalemia and rhabdomyolysis; therefore total intravenous anaesthesia is probably the best option. General anaesthesia may have a beneficial role in the management of patients unable to tolerate lying supine position despite respiratory support, or those with bulbar muscle involvement, putting them at risk of aspiration and contraindicating non-invasive positive pressure ventilation (NPPV). Neuromuscular blocking agents may be used although careful dosing and meticulous monitoring is of paramount importance, since neuromuscular disease may predispose to an unpredictable response to neuromuscular block. There are several cases where rocuronium and sugammadex have been used safely, so this combination may be a safe option.

Concerning the use of opioids, short-acting agents are recommended to avoid the risk of drug accumulation. Remifentanil has been successfully used due to its short duration of action, so this agent could prevent prolonged postoperative respiratory depression and sedation.

There is no absolute contraindication to sedation if respiratory involvement is mild, although individual risks and benefits of the procedure must be assessed.

Regional anaesthesia should probably be preferred to general anaesthesia whenever it is a feasible option, including peripheral nerve blocks, and neuroaxial anaesthesia. A carefully conducted neuroaxial anaesthesia technique with no extent of the motor blockade could be performed in patients with preoperative history of functional dependence and poor lung function, in order to avoid significant postoperative pulmonary complications. If indicated, a combined spinal-epidural (CSE) anaesthesia may be the preferred option among neuroaxial techniques, since it allows a quick onset of sensorimotor block, with a reduced intratecal drug dose, the height of which can be titrated by further sequential epidural boluses. Furthermore, this combination may be useful to minimize the risk of hypotension, excessive motor blockade and further deterioration in lung function. The epidural boluses with a low concentration high volume local anaesthetic solution offer both a volume and a local anaesthetic effect, resulting in a more adequate block for surgery. The use of epidural opioids should be avoided because of the risk of postoperative respiratory depression.

Necessary additional diagnostic procedures (preoperative)

The course of anaesthetic management should be planned during the preoperative evaluation of the patient. Family history of muscular disease, and any signs or symptoms that evoke myopathy must be searched for.

Serum creatine kinase is variably elevated, it is useful to subdivide in different types of LGMD, and can help to evaluate the appearance of metabolic perioperative complications like rhabdomyolysis, comparing concentrations to baseline level. Electrocardiogram and Holter may be necessary to rule out any cardiac conduction disorders, and echocardiography should probably be performed every 2 to 5 years. If respiratory symptoms are present, functional respiratory tests may help us to plan perioperative care, and arterial blood gases on room air could assist perioperative dispositions. Ultrasound Doppler of lower limbs must be performed for patients on a wheel-chair and if intermittent pneumatic compression (IPC) is recommended. Consultation to specialists may be advised (Cardiology, Neurology, Respiratory Care). The patient should be programmed in the first position of the operative program.

Particular preparation for airway management

Tongue hypertrophy could make airway management more difficult, although there are not enough previous reports to support this concern; a delayed gastric emptying because of hypomotility of gastrointestinal tract may predispose to an increased risk of aspiration. The presence of somonolence and previous frequent chest infections may suggest muscle respiratory weakness, and increased risk of hypoventilation. Laryngeal mask has been used in at least one case for hemorrhoidectomy surgery, although laryngeal mask is not recommended in case of delayed gastric emptying.

A difficult intubation cart, a videolaryngoscope and a fiberoptic bronchoscope should be in close proximity.

Particular preparation for transfusion or administration of blood products

There are no particular recommendations.

Particular preparation for anticoagulation

Patients may be on oral anticoagulation because of suffering atrial fibrillation or flutter. The presence of an impaired mobility may predispose to deep venous thrombosis, so compression stockings and/or low molecular weight heparin may be indicated in the perioperative period. Intermitent pneumatic compression is recommended for high-risk surgeries, if there is no contraindications (significant arterial insufficiency, edema from congestive heart failure, active phlebitis, deep vein thrombosis, localized wound infection or cellulitis).

Particular precautions for positioning, transport or mobilisation

These patients may suffer spinal rigidity, scoliosis, and limb contractures, although uncommon; for this reason, positioning in the operating room must be careful.

Probable interaction between anaesthetic agents and patient's long term medication

Careful administration of opioids, neuromuscular blocking agents and anaesthetics is mandatory due to an increased sensitivity to these agents. Inhaled anaesthetics and succinylcholine should be avoided in order to prevent triggering muscular toxicity. Patients may be previously treated with antiinflamatory drugs, corticosteroids (reported improvements in LGMD 2C-F), and ACEi or betablockers for the treatment of left ventricular dysfunction, so perioperative adjustment of doses or increasing doses (in the case of corticoisteroids) could be necessary in the perioperative setting.

Anaesthesiologic procedure

Regional anaesthesia is recommended, and general anaesthesia has been used in several surgical procedures, and intravenous anaesthesia is the safest choice. Neuromuscular block agents may be used, although careful dosing and meticulous monitoring is of paramount importance, since neuromuscular disease may predispose to an unpredictable response to neuromuscular block. There are several cases where rocuronium and sugamadex have been used safely, so this combination may be a safe option.

Particular or additional monitoring

The anaesthesia machine should be prepared by using a new or disposable breathing circuit, a fresh CO2 absorbent, removing or disconnecting the vaporizers and flushing with O2 at a rate of 10 l/min for at least 20 minutes before use.

The regular intraoperative monitoring includes a a 5-lead electrocardiography, pulse oximetry, end-tidal capnography, non-invasive blood pressure, temperature monitoring, bispectral index, and neuromuscular block monitoring. Mechanical ventilation should be adjusted to maintain PaCO2 close to patient´s preoperative values. Temperature should be kept between 36.5 and 37.8 ºC using forced-air warming blankets and infusion warmer if necessary.

Possible complications

Cardiac conduction disorders (atrioventricular block, atrial fibrillation/ flutter), pacemaker dysfunction, cardiac failure, adrenal insufficiency due to improper corticosteroid perioperative management, respiratory complications (infections, hypoventilation, prolonged mechanical ventilation), muscular toxicity triggered by inhaled agents or succynicholine, hyperkaliemic cardiac arrest.

Postoperative care

Early mobilization, avoiding prolonged mechanical ventilation, and temperature control, must be among the mains goals in the postoperative period. Non-invasive positive pressure ventilation delivered by a facemask may be needed in the postoperative period to attenuate any exacerbation of respiratory symptoms associated with the general or neuroaxial anaesthesia in order to avoid intubation. The NVPP should be considered as a part of the perioperative management of these patients.

Information about emergency-like situations / Differential diagnostics

caused by the illness to give a tool to distinguish between a side effect of the anaesthetic procedure and a manifestation of the disease

The main complications that must be ruled out are hyperkalemia, rhabdomyolysis, cardiac conduction disorders, heart failure and respiratory failure. Hypoventilation may be more difficult to identify, so a special attention must be payed to indirect signs such as somonolence and bradipnea; Differential diagnosis should include residual effects of opioids and anaesthetics.

Ambulatory anaesthesia

Ambulatory anaesthesia may be feasible if cardiac and respiratory condition is stable and mild, although previous data is insufficient to make a particular recommendation, and individualized risk assessment and multidisciplinary approach is needed depending on the type of surgical procedure and anaesthetic technique.

Obstetrical anaesthesia

Regional anaesthesia may be a good option since it may allow us to avoid triggering agents, minimize the risk of respiratory depression and provide postoperative analgesia with the minimum doses of opioids. There are previous reports using epidural analgesia for elective caesarean section, as well as combined spinal-epidural anesthesia. Conventional management of vaginal delivery is safe, but epidural analgesia may be useful in order to avoid the adverse effects of opioids.

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These guidelines have been prepared by:

Author
Alexo López Álvarez, Anaesthesiologist, Hospital Meixoeiro de Vigo, Spain
lopez.alexo@gmail.com

Co-Author
Adriana Roman Fernandez, Anaesthesiologist, Hospital Meixoeiro de Vigo, Spain

Peer revision 1
Freda Richa, Anaesthesiologist, Hotel-Dieu de France Hospital, Beirut, Lebanon
fredrich24@yahoo.com

Peer revision 2
Matthew P. Wicklund, Neurologist, Penn State College of Medicine,
Penn State University, USA
mwicklund@hmc.psu.edu