Case
A 46-year-old woman with facioscapulohumeral muscular dystrophy for left knee arthroscopy.

Questions
- What potential abnormalities should concern the anaesthetist?
- What anaesthetic technique should be used?
- What agents may be safely used for general anaesthesia?
Muscular dystrophy comprises a group of hereditary diseases of skeletal muscles characterized by progressive weakness and degeneration of muscle.

Four obligatory criteria that distinguish them from other neuromuscular diseases:

1. It is a primary myopathy.
2. It has a genetic basis.
3. The course is progressive.
4. Degeneration and death muscle fibres occur at some stage in the disease.
# Types of Muscular Dystrophy

<table>
<thead>
<tr>
<th>Disease</th>
<th>Clinical Note</th>
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<tbody>
<tr>
<td>Duchenne MD</td>
<td>An X-linked recessive disorder, affects males almost exclusively. Incidence 1-3 cases per 10,000 live male births. Begins early childhood; death by late teens or early 20’s; most severe form. Affected individuals produce abnormal dystrophin, a protein found on the sarcolemma of muscle fibres.</td>
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<td>Becker’s MD</td>
<td>Like Duchenne but present later in life</td>
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<td>Emery-Dreifuss MD</td>
<td>The 3rd most common, characterized by early contractures/muscle weakness in a humeroperoneal distribution, and cardiomyopathy.</td>
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<td>Limb-girdle MD</td>
<td>Mostly benign clinical course.</td>
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<tr>
<td>Facioscapulohumeral MD</td>
<td>No heart involvement. Muscles in lower extremities are less affected, and respiratory muscles are usually spared.</td>
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## Types of Muscular Dystrophy

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<td>Myotonic Dystrophy</td>
<td>A multisystem disorder that is the most common cause of myotonia—slowing of relaxation after muscle contraction in response to electrical or percussive stimuli.</td>
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Clinical Findings in Duchenne MD

Proximal muscle weakness (gait disturbance)
Hypertrophied muscles (esp. calf muscles)
Progressive kyphoscoliosis
Respiratory disease
  # Inability to cough and clear secretions
  # Recurrent infections
  # Restrictive pulmonary defect
  # Respiratory failure
  # Pulmonary hypertension
Cardiomyopathy (dilated or hypertrophic ~10%, MR due to papillary muscle dysfunction ~25%, ECG: PR interval prolongation, QRS and ST abnormalities /prominent P waves / deep Q wave. Atrial arrhythmias are common)
Association with malignant hyperthermia?
# Clinical Findings in Myotonic Dystrophy

<table>
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<tr>
<th>Cardiac</th>
<th>Pulmonary</th>
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<tr>
<td>Cardiomyopathy</td>
<td>Pulmonary aspiration</td>
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<tr>
<td>Atrial arrhythmias</td>
<td>Alveolar hypoventilation</td>
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<td>Heart block</td>
<td>Reduced vital capacity</td>
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<td></td>
<td>Chronic hypoxaemia</td>
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<td></td>
<td>Increased sensitivity to drug-induced depression</td>
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<td></td>
<td>Central sleep apnoea and hypersomnolence</td>
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</tbody>
</table>

- Cataracts
- Frontal baldness
- Testicular atrophy
- Mild mental deficiency
Clinical Findings in Myotonic Dystrophy

Endocrine
- Pancreatic insufficiency
- Adrenal insufficiency
- Thyroid gland dysfunction
- Gonadal insufficiency

Gastrointestinal
- GI hypomotility
- Gastric dilatation
- Gastric regurgitation

Uterine atony

Associated with malignant hyperthermia.

Prolonged contraction in response to suxamethonium.

Myotonia is not relieved by anaesthesia or muscle relaxants.
Anaesthetic Considerations in Duchenne + Becker’s Muscular dystrophy

Preoperative assessment:
History/examination/investigations – information to effectively assess the likelihood of anticipated problems.

- Anaesthetic management is complicated not only by muscle weakness but also by cardiac and pulmonary manifestations.
- An association with malignant hyperthermia has been suggested but is controversial. No definitive genetic link to MH has been found, most paediatric anaesthetists consider these patients to be MH-susceptible based on case reports, and will perform a nontrigger technique. Use of inhalational agents, albeit for a relatively short period of time, appears to be safe when intravenous access is not available. Marked respiratory and circulatory depression may be seen with volatile agents in pts with advanced disease.
- Preoperative premedication with sedatives or opioids is best avoided due to pts may be at increased risk for aspiration from respiratory muscle weakness or gastric hypomotility.
- For major surgical procedures, esp. major orthopaedic/upper abdominal/thoracic procedures, pre-operative pulmonary function tests and echocardiography are appropriate.
Suxamethonium has been used safely in some pts but is best avoided because of unpredictable responses and the risk of inducing rhabdomyolysis/severe hyperkalaemia or triggering MH. Be aware that acute rhabdomyolysis in the perioperative period can be induced by other factors besides SUX, including GA and electrolytes disorders.

Although some pts exhibit a normal response to non-depolarizing muscle relaxants, others may be very sensitive.

Regional or local anaesthetics may therefore be preferable in these pts.

Respiratory complications are largely responsible for perioperative morbidity. Pts with vital capacities < 30% of predicted appear to be at greatest risk and often require postoperative mechanical ventilation.
Anaesthetic Considerations in Myotonic Dystrophy

Preoperative assessment:
Focus on evaluation of cardiopulmonary system.
ECG is mandatory- taking special note of any rhythm disturbances or conduction defects.
Cardiac failure should be excluded.
Pulmonary evaluation should elicit any history of recent changes in pulmonary signs or symptoms including recent infections or changes in cough pattern or sputum production.
Clinical evidence of thyroid/pancreatic/adrenal hypofunction should be investigated, perioperative steroid supplementation prn.
Drug history is very important- interactions with anaesthetic agents.
Phenytoin- induce enzymes/decrease MAC/enhances the CNS toxicity of LA
Procainamide-prolongs the duration of action of muscle relaxant and antagonizes the effects of neostigmine.
Quinine –increases the risks of ventricular arrhythmia in pts with anti-histamine, and potentiate the effects of digoxin.
Patients with Myotonic dystrophy are at high risk for perioperative respiratory and cardiac complications. Surgery with GA should be avoided, therefore, when not absolutely necessary.

Induction of anaesthesia without complications has been reported for a number of agents including thiopentone, inhalational agents, and propofol (with or without ketamine)

- Reported a prolonged recovery time of 120 min after an induction dose of propofol 50 mg in an adult pt.
- Furthermore generalized myotonia has been precipitated by the use of propofol.

Neuromuscular blockade, if needed, should be performed with short-acting agents (atracurium or cisatracurium).

N2O and inhalational agents can be used as maintenance anaesthesia. Reversal with anticholinesterases is to be avoided.

The choice of intraoperative analgesia, short acting opioids, such as alfentanyl and remifentanyl.

NSIAID’s devoid of problems of respiratory depression, but care must be taken to minimize the risks of renal toxicity.
Anaesthetic Considerations in Myotonic Dystrophy

Perioperative factors associated with the development of myotonia

- Suxamethonium has been relatively contraindicated because it may precipitate intense myotonic contractions.
  - # Trismus can prevent opening the mouth for intubation.
  - # Myotonic contraction of respiratory, chest wall, or laryngeal muscles can make ventilation difficult or impossible.

- Non-depolarizing muscle relaxants appear to behave normally, however, they do not consistently prevent or relieved myotonic contractions. Because of variable sensitivity to it, which may be as a result of the disease process or a side-effect of drug therapy (e.g. phenytoin).
  close monitoring of neuromuscular function is essential, be aware of triggering myotonic episodes.
Perioperative factors associated with the development of myotonia

- Anticholinesterases drugs may precipitate myotonia, presumably because myotonic muscle has increased sensitivity to stimulatory effects of Acetylcholine (Ach). Use short acting agents such as atracurium, which do not require antagonism.

- Avoid potassium containing solutions—may worsen clinical myotonia.

- Regional anaesthesia can be employed but does not always prevent myotonic contractions.

- Cold and shivering independently induce myotonia. Wherever possible, increases theatre ambient temperature/ warms iv fluids. Core temp. monitored.

- Myotonic contraction during Surgical manipulation and electrocautery is a major management problem.

- Troublesome myotonia rarely occurs, but can be reduced by injecting/tropical LA’s in the muscles or by giving quinine hydrochloride 300 to 600mg iv. Large concentrations of VA’s may abolish myotonic contractions at the expense of cardiovascular depression and post-op shivering.
Post operative management

- The main post-op. complications are pulmonary: prolonged ventilatory failure, pneumonia, and atelectasis.
- Perioperative cardiac conduction abnormalities are less likely to occur but still warrant close cardiovascular monitoring.
- ICU/HDU bed must be available.
  Whichever choice is made, a period of at least 24h post-op care should be planned.

As association between Myotonic dystrophy and MH has been suggested but has not been firmly established, it does not seem, therefore, that pts with MD are at an increased risk for MH. Interestingly, both disorders map to chromosome 19, albeit in different locations. (ryanodine receptor on chromosome 19)