# Anesthetic Management of Patients with Muscle Dystrophy

*Essay*

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***In Anesthiology***

*By*

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# List of Abbreviations

| | |
|---|---|
| **ABGs** | Arterial blood gases |
| **ACh** | Acetylcholine |
| **AD-EDMD** | Autosomal dominant Emery-Dreifuss muscular dystrophy |
| **AR-EDMD** | Autosomal recessive Emery-Dreifuss muscular dystrophy |
| **ATP** | Adenosine triphosphate |
| **BIPAP** | Biphasic positive airway pressure |
| **BMD** | Becker muscle dystrophy |
| **$Ca^{++}$** | Calcium |
| **CABG** | Coronary artery bypass graft |
| **CK** | Creatinine kinase |
| **CMD** | Congenital muscle dystrophy |
| **CPAP** | Continuous positive airway pressure |
| **CPK** | Creatine Phosphokinase |
| **DAGs** | Dystrophin-associated glycoproteins |
| **DD** | Distal dystrophy |
| **DMD** | Duchenne muscle dystrophy |
| **EC** | Excitation-contraction |
| **ECG** | Electrocardiogram |
| **ECHO** | Echocardiography |
| **EDMD** | Emery-Dreifuss muscle dystrophy |
| **EMG** | Electromyography |
| **FBS** | Fasting blood sugar |
| **FCMD** | Fukuyama congenital muscular dystrophy |
| **FSHD** | Facioscapulohumeral muscle dystrophy |
| **FVC** | Forced vital capacity |
| **GA** | General anesthesia |
| **HLAs** | Human leukocyte antigens |
| **IVCTs** | in vitro contracture tests |
| **ICU** | Intensive care unit |
| **$K^{+}$** | Potassium |
| **LGMD** | Limb Girdle muscle dystrophy |

# ☙List of Abbreviations (Cont.)❧

| | |
|---|---|
| **MD** | muscular dystrophy |
| **MDs** | Muscular dystrophies |
| **MEB** | Muscle-eye-brain |
| **MEP** | Maximal expiratory pressure |
| **MH** | Malignant hyper thermia |
| **MMD** | Myotonic muscle dystrophy |
| **$NaHCO_3$** | Sodium bicarbonate |
| **NMJ** | Neuromuscular junction |
| **NPPV** | Noninvasive positive-pressure ventilation |
| **OPMD** | Oculopharyngeal muscle dystrophy |
| **$PaCO_2$** | Arterial tension of carbon dioxide |
| **PACU** | Post-anesthesia care unit |
| **$PaO_2$** | Arterial tension of oxygen |
| **$PAO_2$** | Alveolar oxygen tension |
| **PCF** | Peak cough flow |
| **REM** | Rapid eye movement |
| **RV** | Residual volume |
| **RYR1** | Ryanodine receptor |
| **SDB** | Sleep-disorder breathing |
| **SR** | Sacroplasmic reticulum |
| **TIVA** | Total intravenous anesthesia |
| **TLC** | Total lung capacity |
| **VC** | Vital capacity |
| **VD** | Dead space |
| **VT** | Tidal volume |
| **WWS** | Walker-Warburg syndrome |
| **XL-EDMD** | X-linked Emery-Dreifuss muscular dystrophy |

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# INTRODUCTION

## Definition:

Muscular dystrophies (MDs) are a heterogeneous group of hereditary disorders characterized by muscle fiber necrosis and regeneration, leading to progressive weakness and degeneration of muscle ***(Morgan et al., 2006)*.**

## Incidence:

The incidence rates of MDs vary depending on the specific type. Duchenne MD is the most common MD and is sex-linked, with an inheritance pattern of 1 case per 3500 live male births ***(Dubowitz, 1995 and Emery, 1993)***. One third of cases occurs as a result of spontaneous new mutations ***(Emery, 1991)***. Becker MD is the second most common form, with an incidence of 1 case per 30,000 live male births ***(Shapiro and Specht, 1993)***. Other types of MD are rare. For example, limb-girdle dystrophy occurs in only 1.3% of patients with MDs. The oculopharyngeal type is more common in French Canadians than in other groups ***(Pratt and Meyers, 1986)***. Distal MD tends to occur more in Sweden.

## History:

The first historical account of MD was reported by ***Conte and Gioja in (1836)***. They described 2 brothers with progressive weakness starting at age 10 years. These boys later developed generalized weakness and hypertrophy of multiple

muscle groups, which are now known to be characteristic of the milder Becker MD. At the time, however, many thought that Conte and Gioja described tuberculosis; thus, they did not achieve recognition for their discovery ***(Conte and Gioja, 1836)***.

***Meryon (1852)*** reported in vivid details a family with 4 boys, all of whom were affected by significant muscle changes but had no central nervous system abnormality when examined at necropsy. He subsequently wrote a comprehensive monograph on MD and even went on to suggest a sarcolemmal defect to be at the root of the disorder. He further suspected that the disorder is genetically transmitted through females and affects only males.

Guillaume Duchenne was a French neurologist who was already famous for his application of faradism (the use of electric currents to stimulate muscles and nerves) in the treatment of neurologic disorders when he wrote about his first case of MD ***(Duchenne, 1868)*.**

In 1868, he gave a comprehensive account of 13 patients with the disease, which he called "paralysie musculaire pseudo-hypertrophique." Because Duchenne was already held in high esteem for his work in faradism and for his contributions to the understanding of muscle diseases, one of the most severe and classic forms of MD, Duchenne MD, now bears his name.

## Types:

1. Duchenne muscle dystrophy(DMD)
2. Becker muscle dystrophy(BMD)
3. Emery-Dreifuss muscle dystrophy(EDMD)
4. Facioscapulohumeral muscle dystrophy(FSHD)
5. Myotonic muscle dystrophy(MMD)
6. Limb Girdle muscle dystrophy(LGMD)
7. Oculopharyngeal muscle dystrophy(OPMD)
8. Congenital muscle dystrophy(CMD)
9. Distal Myopathies

***(Dubowitz, 1995)***

# CLASSIFICATION

There have been several attempts at classifying the different types of muscular dystrophy (MD).Walton and Nattrass proposed an original classification scheme which depends on two considerations: the mode of inheritance and the distribution of predominant muscle weakness ***(Alan and Emery, 1998).***

***Heritable MDs include the following (Yates et al., 1993):***

## A. X-Linked Inheritance:

***Occurs with the following dystrophies:***

- Duchenne muscular dystrophy(DMD)
- Becker muscular dystrophy(BMD)
- Emery-Dreifuss muscular dystrophy (XL-EDMD)

## B.Autosomal Recessive Inheritance:

***Occurs with the following dystrophies:***

- Limb-girdle muscular dystrophies (LGMD) 2A, 2B, 2C, etc
- Congenital muscular dystrophies(CMD)
- Distal dystrophies

## C.Autosomal Dominant Inheritance:

***Occurs with the following dystrophies:***

- Facioscapulohumeral muscular dystrophy(FSHD)
- Myotonic muscular dystrophy(MMD)

- Oculopharyngeal Muscular Dystrophy (OPMD).
- Limb-girdle muscular dystrophies (LGMD)1A,1B,1C, etc.
- Distal dystrophies.
- Emery-Dreifuss muscular dystrophy (AD-EDMD)

## 1. Duchenne's Muscular Dystrophy (DMD):

### *Incidence:*

Around 1 in 3,500 boys in the UK ***(Prior et al., 2005).***

### *Aetiology:*

DMD is caused by afault in the dystrophin gene which is absent or severely abnormal ***(Emery, 1998 )***.

### *Genetics of DMD:*

DMD is an X-linked recessive trait, "carried" by females and manifest in males ***(Bushby et al., 2000)***.

### *Presentation:*

The distinctive feature of Duchenne muscular dystrophy is a progressive proximal muscular dystrophy with characteristic pseudohypertrophy of the calves. All patients have symptoms by age 3 years, but diagnosis is often later. ***Symptoms and signs are***:

- Motor milestones delayed (50% not walking by 18 months)
- Inability to run - with waddling gait when attempting to do so.
- Inability to jump or hop, no spring in the step.
- Climbing up legs' using the hands when rising from the floor (Gower's sign).

- Hypertrophy of calf muscles (other muscles may also be hypertrophied, including deltoid, quadriceps, tongue and masseters.
- The child may 'slip through the hands' on being lifted, due to hypotonia of muscles around the shoulder.

***(Bushby, 2007)***

## ***Non-locomotor presenting symptoms are:***

- Speech may be delayed.
- Global developmental delay may be the presenting feature ***(Essex et al., 2001)***.
- Anaesthetic complications, rarely, are the presentation.
- If DMD is suspected, test the serum creatine kinase. This is always massively elevated in DMD (creatine kinase is also raised in some other muscular dystrophies). If so, refer to a specialist at this stage, for diagnosis and genetic counselling.

## ***Investigations:***

- Serum creatine kinase is always massively raised, even to >200 times normal.
- Electromyography (EMG) shows non-specific dystrophic changes.
- Muscle biopsy establishes the diagnosis, showing reduced or absent dystrophin.
- Genetic analysis can confirm the diagnosis in about 70% (when a deletion of the dystrophin gene is present).

- Carrier status can usually be identified by genetic analysis. Also, serum creatine kinase is usually high in carriers.

***(Bushby, 2007)***

## ***Differential diagnosis:***

- Other muscular dystrophies - particularly Becker MD which is similar but progresses more slowly. The clinical features, muscle biopsy and genetic analysis help distinguish DMD from the other muscular dystrophies.
- Other types of myopathy.
- Neurological causes of muscle weakness, e.g. spinal cord lesions, spinal muscular atrophy, motor neurone disease, multiple sclerosis. These conditions are likely to have additional features such as sensory loss, upper motor neurone signs or muscle fasciculation.

## ***Complications:***

- Joint contractures are usual.
- Respiratory muscle failure is progressive, leading to hypoventilation, loss of coughing and respiratory infections.
- Cardiomyopathy is common, although actual symptoms are less likely and usually occur late on in DMD. Cardiac arrythmias can occur.
- Respiratory failure, pneumonia and/or cardiomyopathy are the usual cause of death.
- Smooth muscle can also be affected, causing gastrointestinal symptoms such as gastric dilation or pseudo-obstruction.