

orphananesthesia

Anaesthesia recommendations for patients suffering from **Emery-Dreifuss Muscular Dystrophy**

Disease name: Emery-Dreifuss Muscular Dystrophy (EDMD)

ICD 10: G71.0

Synonyms: Benign Scapulooperoneal Muscular Dystrophy, Hauptmann-Thannhauser Muscular Dystrophy, EDMD 1 (X-linked affecting EMD gene), EDMD 2/3 (autosomal dominant/recessive affecting LMNA gene)

Other laminopathies may be phenotypically similar.

Medicine in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

Disease summary

Joint contractures (Achilles tendons, elbows and spine). Posterior neck and lower back involvement may result in a rigid spine.

Humeroperoneal muscle weakness by adolescence, later involving the proximal limb girdles.

Cardiac conduction abnormalities (PR prolongation & complete heart block), atrial and ventricular arrhythmias and dilated cardiomyopathy usually beginning in the 2nd – 3rd decade of life.

Typical surgery

- Tendon release
- Corrective spinal surgery
- Caesarean Section
- Permanent pacemaker / defibrillator insertion

Type of anaesthesia

All forms of anaesthesia are possible. Intubation and central neuro-axial blocks may be difficult due to joint contractures.

Although evidence is lacking it may be prudent to avoid suxamethonium and inhalational anaesthetics during the first decade of life to avoid anaesthesia-induced rhabdomyolysis.

Necessary additional diagnostic procedures (preoperative)

Electrocardiogram (ECG).

Echocardiography and 24 hour ambulatory ECG telemetry are recommended.

Cardiac electrophysiological testing should be considered in patients with conduction defects.

Particular preparation for airway management

There is the potential for reduced gut motility, aspiration risk should be assessed and managed. The airway plan should include options for dealing with a difficult airway secondary to restricted neck movement.

Particular preparation for transfusion or administration of blood products

May be at increased risk of intra-operative bleeding; mechanism not fully understood.

Consider anti-fibrinolytics and early treatment of acquired coagulopathy.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transport or mobilisation

Take care when positioning contractures.

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

Not reported.

Anaesthesiologic procedure

An opioid sparing technique and careful titration of muscle relaxants will limit the impact on post-operative respiratory function.

Judicious use of fluids and a means of external pacing should it be necessary (pads or temporary pacing wire) will reduce the risks of cardiac failure. Electrolyte monitoring and DC cardioversion should be available.

Potentiation of neuromuscular blockade by hypothermia should be avoided.

Particular or additional monitoring

Invasive arterial pressure monitoring is prudent. Central venous pressure monitoring may be of benefit.

Neuromuscular blockade should be monitored routinely.

Possible complications

Decompensation of existing cardiac abnormalities:

- Conduction defects, including complete heart block and atrial standstill
- Cardiac failure
- Ventricular and supraventricular arrhythmias.

Prolonged neuromuscular blockade.

Postoperative care

Patients are at risk of respiratory compromise. High dependency care should be considered particularly following intra-abdominal or thoracic surgery.

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

Anaesthesia induced rhabdomyolysis may present as a Malignant Hyperthermia type picture. Hyperkalaemia and life-threatening arrhythmias are possible.

Ambulatory anaesthesia

Joint contractures and muscle weakness may affect gait and stability.

Obstetrical anaesthesia

Require early investigation to inform decision making.

Elective Caesarean Section may be necessary, and the feasibility and impact of central neuro-axial blocks need to be assessed.

Literature and internet links

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