

## Anaesthesia recommendations for **Dubowitz syndrome**

**Disease name:** Dubowitz syndrome

**ICD 10:** Q87.1

**Synonyms:** intrauterine primordial microsomia

**Disease summary:** Dubowitz Syndrome (DS) is a rare genetic condition, originally described in 1965, with considerable phenotypic variability suggesting genetic heterogeneity. Over 200 cases are reported in the literature. DS is characterized by intrauterine and/or postnatal growth failure, microcephaly, mild to moderate cognitive delay, hyperactivity, immune defect, and increased risk of blood dyscrasia (pancytopenia) and malignancy (leukaemia, neuroblastoma). Congenital abnormalities such as heart defects, genital hypoplasia, hypospadias, rectal malformations and skeletal anomalies have been reported. Peculiar, but variable morphological facial features are sloping forehead, epicanthic folds, blepharophimosis, ptosis, broad and flat nasal bridge, low-set and poorly-formed ears, micro- and retrognathia; submucous cleft palate and velopharyngeal insufficiency may be present. Ocular and dental abnormalities such as cataract, strabismus, hypo-/anodontia have been described. Eczema is also frequent.

Single gene mutations (de novo or biallelic variants) and genomic imbalances have been reported in patients with Dubowitz syndrome clinical phenotypes. Several cases remain without an identifiable genetic cause.

DS is a complex and multisystemic condition and requires a multidisciplinary approach.

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Medicine is in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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Find more information on the disease, its centres of reference and patient organisations on Orphanet: [www.orpha.net](http://www.orpha.net)

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### **Typical surgery**

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There is no curative surgery involved. Associated surgeries include among others tonsillectomy, adenoidectomy, bilateral tubal ligation, cardiac surgery (complex correction of malformation of the heart and great vessels), velopharyngeal insufficiency operation and orthopaedic surgery.

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### **Type of anaesthesia**

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There is no definite recommendation for either general or regional anaesthesia.

Succinylcholine and volatile anaesthetics should be avoided in patients immobilized in a wheelchair because there is a risk of rhabdomyolysis or hyperkalaemic cardiac arrest.

Therefore, total intravenous anaesthesia should be favoured. There are no reports of spinal, epidural and caudal anaesthesia yet.

(Analgo-) sedation should be done after carefully assessing the individual risks (pharyngeal muscle weakness, cardiopulmonary involvement) with a high awareness to respiratory failure and risk of aspiration.

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### **Necessary additional pre-operative testing (beside standard care)**

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Individuals with DS may present with muscular weakness and relevant involvement of other organ systems (e.g. cardiopulmonary involvement) which make further diagnostic procedures necessary.

Cardiac function test including ECG and echocardiography should be performed to evaluate the presence of cardiac anomalies, such as malformation of the heart and great vessels.

Lung function test (lung volumes and blood gas analysis) can be done if pulmonary involvement is assumed. Note that there is no correlation between lung function and postoperative respiratory complications.

Literature suggests a chest and cervical X-ray to determine syndrome-associated skeletal anomalies.

If muscular weakness is present and regional anaesthesia is planned, neurological consultation is helpful for juridical reasons.

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### **Particular preparation for airway management**

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Laryngeal masks of different sizes, fiberoptic bronchoscopy/ video laryngoscope and a coniotomy set should be prepared for possible use. For the endotracheal intubation, the use of a stylet should be considered.

Patients with weakness of the oropharyngeal muscles including swallowing difficulties have a higher risk of aspiration. Providing an aspiration prophylaxis with e.g. H2 receptor blockers seems to be reasonable.

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### **Particular preparation for transfusion or administration of blood products**

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Not reported.

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### **Particular preparation for anticoagulation**

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There is no evidence to support the need of particular anticoagulation. But the impaired mobility of patients with severe muscle weakness may suggest a higher risk of postoperative thrombosis.

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### **Particular precautions for positioning, transportation and mobilisation**

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Not reported. Note the possible immobilisation in a wheelchair.

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### **Interactions of chronic disease and anaesthesia medications**

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Not reported.

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### **Anaesthetic procedure**

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To our knowledge, only one report of an uneventful general anaesthesia has been made.

Avoiding succinylcholine in case of immobilisation due to the risk of hyperkalaemic cardiac arrest and rhabdomyolysis seems to be reasonable.

Opiates, propofol and non-depolarizing neuromuscular blocking agents have been used without any complication.

There is no need for prophylactic postoperative ventilation if neuromuscular blockade is monitored consistently.

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### **Particular or additional monitoring**

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Monitoring of the neuromuscular blockade is recommended if neuromuscular blocking agent is used.

Monitoring of the body temperature should be considered using warming management and to avoid post-operative shivering and increased oxygen demand.

In case of major fluids shift, advanced disease or high risk surgery, arterial cannulation for invasive blood pressure measurement and central venous line placement seem indispensable.

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### **Possible complications**

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Primarily, anaesthesiological complications may be caused by the challenging airway management due to anatomic characteristics (receding chin, microcephaly, micrognathia).

Anaesthetic management may be influenced by relapsing infections and the predisposition to allergies.

Sedative drugs (benzodiazepines) can cause respiratory insufficiency.

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### **Post-operative care**

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Close-meshed observation, including oxygen saturation, respiratory frequency and blood pressure measurement, especially during recovery, is highly recommended.

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### **Disease-related acute problems and effect on anaesthesia and recovery**

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Disease triggered emergency-like situations have not been reported so far.

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### **Ambulatory anaesthesia**

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Due to the lack of experience, postoperative monitoring is obligatory and excludes ambulatory anaesthesia in most cases.

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### **Obstetrical anaesthesia**

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There are no reports about obstetrical anaesthesia with patients suffering from Dubowitz syndrome.

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