orphanesthesia

Anesthesia recommendations for

Miller-Dieker syndrome

Disease name: Miller-Dieker syndrome

ICD 10: Q93.88

ORPHAcode: 531

Synonyms: 17p13.3 deletion syndrome

Disease summary: Miller-Dieker syndrome (MDS) is a rare disorder that is characterized by type I lissencephaly (smooth brain), facial dysmorphism, and often other congenital abnormalities [3]. MDS is caused by visible deletion or microdeletion of 17p13.3 with haploinsufficiency of LIS1 [6].

Typical facial features include a prominent forehead, bitemporal hollowing, short nose with upturned nares, prominent upper lip with downturned vermillion border, low-set posteriorly rotated ears, and micrognathia [4,5]. Most patients with MDS have epilepsy and severe developmental delay [6]. Congenital heart diseases are frequently associated with MDS. Kidney anomalies, sacral dimple, omphalocele, genital anomalies, and clinodactyly are also associated with MDS [4,5].

Diagnosis may be incorrect; if uncertainty exists, the diagnosis should be re-evaluated.

Every patient is unique; individual circumstances must always guide clinical care.

Medicine is in progress; new clinical knowledge may not be yet reflected in this guideline Perhaps new knowledge.

Recommendations are not rules or laws; they provide a framework to support clinical decision-making. Although this recommendation has passed a structured review process, it does not meet the formal criteria of a guideline.

Translations may not always reflect the most recent updates of the English version.

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

Emergency information

A	**AIRWAY / ANESTHETIC TECHNIQUE**	Micrognathia and increased risk of aspiration and gastroesophageal reflux require careful airway management. Neuraxial anesthesia should be avoided, because MDS may be associated with spinal abnormalities. General anesthesia (GA) with tracheal intubation is preferred due to increased risk of aspiration and gastroesophageal reflux.
B	**BLOOD PRODUCTS (COAGULATION)**	The general rules for perioperative blood management may be applied.
C	**CIRCULATION**	Congenital heart diseases are frequently seen in patients with MDS. ECG and echocardiogram are recommended to detect cardiac malformations.
D	**DRUGS**	Long-term use of certain anticonvulsant agents may induce rapid metabolism of neuromuscular blockers and opioids. Because electroencephalographic monitoring, such as BIS, does not reliably reflect the depth of anesthesia in patients with MDS, intravenous anesthetics are not recommended for the maintenance of GA. No risk for MH.
E	**EQUIPMENT**	No specific recommendations are given.

Typical surgery and procedures

Due to feeding and swallowing difficulties, aspiration pneumonia is common in patients with MDS [1,2]. Typical surgeries to manage these complications of MDS include percutaneous gastrostomy and laryngotracheal separation [2].

Type of anesthesia

Neuraxial anesthesia should be avoided, because sacral dimple is frequently seen in patients with MDS [3]. Sacral dimple may be associated with spinal abnormalities.

General anesthesia with tracheal intubation is preferred due to increased risk of aspiration and gastroesophageal reflux [1,2]

There is no report describing regional anesthesia in patients with MDS.

Necessary additional preoperative testing (beside standard care)

Because patients may have lung damage due to repeated aspiration, chest X-ray and oxygen saturation must be evaluated preoperatively.

Congenital heart diseases are frequently seen in patients with MDS [2,4,5]. ECG and echocardiogram are recommended to detect cardiac malformations.

Severe developmental delay and epilepsy are seen in most patients [6]. Appropriate measures to treat breakthrough seizures should be in place. Other reported organ malformations such as kidney anomalies and omphalocele may require further evaluation to exclude any potential issues arising with fluid management, renal clearance, or gastrointestinal absorption [7].

Particular preparation for airway management

Patients with MDS have a characteristic facial appearance (e.g., prominent forehead, bitemporal hollowing, short nose with upturned nares, prominent upper lip with downturned vermillion border, and micrognathia) [2,4,5]. Micrognathia and increased risk of aspiration and gastroesophageal reflux require careful airway management.

Because most patients need surgical treatment within the first few years of life, awake intubation is rarely a choice for the airway management [1,2]. The airway needs to be managed after induction of general anesthesia. Video laryngoscopy is the preferred choice of intubation technique [8].

Particular preparation for transfusion or administration of blood products

Not reported. The general rules for perioperative blood management may be applied.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilization

Because of developmental delay, patients with MDS may not be able to move independently [2,4]. Extra caution should be taken in patients with contractures.

Interactions of chronic disease and anesthesia medications

Most patients require anticonvulsant drugs to minimize seizure risk [8]. Long term use of certain anticonvulsant agents may induce rapid metabolism of neuromuscular blockers and opioids by

up-regulating hepatic P450 enzymes. Particular care should be taken for the older generation anticonvulsants for this reason.

Anesthetic procedure

Special caution needs to be paid to avoid aspiration during the induction of general anesthesia [1,2].

Intravenous anesthetics are not recommended for maintenance of general anesthesia, because children with MDS have extremely low Bispectral index (BIS) values even when they are awake [1,9]. Volatile anesthetics should be used for maintenance of general anesthesia.

Muscle relaxants and opiates may be metabolized more rapidly due to use of anticonvulsant drugs.

Particular or additional monitoring

Electroencephalography, such as BIS monitor does not provide adequate information for the depth of anesthesia in patients with MDS [1,9].

Neuromuscular monitoring is recommended.

Invasive hemodynamic monitors may be considered in patients with congenital heart disease depending on the severity [4,5].

Possible complications

Aspiration pneumonia is the most common complication after surgery [1,2].

Postoperative seizures may occur. Continuation of anticonvulsant drugs is recommended prior to, during and after the operative procedure.

Postoperative care

Respiratory monitors should be used postoperatively, due to risks of respiratory complications.

Disease-related acute problems and effect on anesthesia and recovery

No specific recommendations are given.

Ambulatory anesthesia

Not reported. Ambulatory anesthesia is not recommended because patients with MDS require extensive perioperative care, as mentioned above.

Obstetrical anesthesia

Not reported. Patients with MDS rarely reach reproductive ages [4,5].

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Please note that this guideline has not been reviewed by two anesthesiologists, but two disease experts.

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