orphanaesthesia

Anaesthesia recommendations for patients suffering from

Miller-Dieker syndrome

Disease name: Miller-Dieker syndrome

ICD 10: Q93.88

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. MDS is caused by visible deletion or microdeletion of 17p13.3 with haploinsufficiency of LIS1.

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. Most patients with MDS are suffered from epilepsy and severe developmental delay. Congenital heart diseases are frequently associated with MDS. Kidney anomalies, sacral dimple, omphalocele, genital anomalies, and clinodactyly are also associated with MDS.

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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

Typical surgery

Due to feeding and swallowing difficulties, aspiration pneumonia is common in patients with MDS. Typical surgeries in treatment of MDS complications include percutaneous gastrostomy and laryngotracheal separation.

Type of anaesthesia

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

General anesthesia with tracheal intubation is preferred due to increased risk of aspiration and gastroesophageal reflux.

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

Necessary additional diagnostic procedures (preoperative)

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. Electrocardiogram and echocardiogram are recommended to detect cardiac malformations.

Severe developmental delay and epilepsy are seen in most patients. 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.

Particular preparation for airway management

Patients with MDS have the characteristic facial appearance (a prominent forehead, bitemporal hollowing, short nose with upturned nares, prominent upper lip with downturned vermillion border, and micrognathia). 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. The airway needs to be managed after induction of general anaesthesia. Video laryngoscopy is the preferred choice of intubation devices.

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, transport or mobilisation

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

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

Most patients require anticonvulsant drugs to minimize seizure risk. 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.

Anaesthesiologic procedure

Special caution needs to be paid to avoid aspiration during the induction of general anaesthesia.

Intravenous anaesthetics are not recommended for maintenance of general anaesthesia, because children with MDS have extremely low Bispectral index (BIS) values even when they are awake. Volatile anaesthetics should be used for maintenance of general anaesthesia.

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

Particular or additional monitoring

BIS monitor does not provide adequate information for the depth of anaesthesia in patients with MDS.

Neuromuscular monitoring is recommended.

Invasive hemodynamic monitors may be considered in patients with congenital heart disease depends on their severities.

Possible complications

Aspiration pneumonia is the most common complication after surgery.

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.

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 diseases, e.g.:

Disease triggered emergency-like situations are not common in MDS.

Ambulatory anaesthesia

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

Obstetrical anaesthesia

Not reported. Patients with MDS rarely reach reproductive ages.

Literature and internet links

1. Chen CP, Chang TY, Guo WY, Wu PC, Wang LK, Chern SR, Wu PS, Su JW, Chen YT, Chen LF, Wang W, Chromosome 17p13.3 deletion syndrome: aCGH characterization, prenatal findings and diagnosis, and literature review. Gene 2013;532:152-9
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Internet links:

Genetic and Rare Diseases Information Center:
https://rarediseases.info.nih.gov/diseases/3669/miller-dieker-syndrome

Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/miller-dieker-syndrome

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This guideline has been prepared by:

Author
Kohei Godai, Department of Anaesthesiology and Critical Care Medicine, Graduate School of Medical and Dental Sciences, Kagoshima University, Kagoshima, Japan
kxg179@icloud.com

Peer revision 1
Volney Sheen, Associate Professor of Neurology, Harvard Medical School, Director of Epilepsy, Beth Israel Deaconess Needham Campus, Beth Israel Deaconess Medical Center, Boston, USA
vsheen@bidmc.harvard.edu

Peer revision 2
Paldeep Atwal, Medical Geneticist, Mayo Clinic, Jacksonville, Florida, USA
paldeep.atwal@googlemail.com

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