

## Anaesthesia recommendations for patients suffering from

### Opitz G/BBB syndrome

**Disease name:** Opitz G/BBB syndrome

**ICD 10:** Q87.8

**Synonyms:** Hypertelorism-hypospadias syndrome, hypospadias-dysphagia syndrome, Opitz BBB/G syndrome, Opitz BBBG syndrome, Opitz-Frias syndrome, Opitz G syndrome, Opitz syndrome, hypertelorism with esophageal abnormalities and hypospadias

Opitz G/BBB syndrome is a congenital malformation syndrome characterized by the defects in the midline of the body. Opitz syndrome is inherited either as X-linked, caused by mutations in the MID1 (midline 1) gene located on Xp 22.3, or autosomal dominant trait with male sex limitation with variable penetrance on chromosome 22q11.2.. One out of every 50,000 to 100,000 males is born with X-linked type II Opitz G/BBB syndrome. The incidence of autosomal dominant Opitz G/BBB syndrome is unknown. It is part of a larger condition known as 22q11.2 deletion syndrome, which affecting 1 in 4,000 people. It is possible to diagnose the syndrome by chromosomal microarray prenatally. Recently, mutations in the SPECC1L gene have been associated to the autosomal dominant condition.

Typical presenting features include hypertelorism, hypospadias, cleft lip/palate, laryngo-tracheoesophageal (LTE) abnormalities, and imperforate anus, developmental delay, and cardiac defects. Recently, however, diaphragmatic hernia and low dural sac have been reported as possible additional features in patients with Opitz G/BBB syndrome.

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Medicine in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic 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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Repair of laryngeal cleft, cleft palate and lip, hypospadias, colostomy formation for imperforate anus, posterior sagittal anorectoplasty (PSARP, pull-through).

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### **Type of anaesthesia**

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Both intravenous and inhalation anaesthesia can be used. Increased risk of aspiration on induction of anaesthesia needs to be considered due to possible associated laryngeal cleft. Also, lung damage from chronic aspiration may impact on ventilation. Anaesthesia needs to be tailored according the presence or absence of cardiac abnormality, i.e. avoiding cardio depressing drugs as nitric oxide.

Regional analgesia is recommended to avoid higher doses of opioid analgesics. However, spinal anomaly has been reported in patient with Opitz GBBB syndrome., Ultrasound has been shown useful in this case to diagnose low termination of dural sac in patients with Opitz GBBB syndrome and facilitate caudal block despite this abnormality.

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### **Necessary additional diagnostic procedures (preoperative)**

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Diagnosis is made on the basis of clinical features in males with ocular hypertelorism along with one or more of the major anomalies. Molecular genetic testing is difficult due to the complicated etiology. Identification of a MID1 mutation confirms the diagnosis.

Prenatal screening is difficult due to subtle changes on ultrasound pictures. Prenatal testing is possible for at-risk pregnancies if a MID1 mutation has been identified in a family member. Fetal sex determination can be performed by chromosome analysis, followed by DNA screening for disease-causing mutations.

Cardiology investigation is necessary to rule out any cardiac involvement.

Bronchoscopy is indicated in suspected laryngo-tracheal abnormalities, i.e. recurrent aspirations, or recurrent chest infections.

MRI of spine can help to avoid complications with neuraxial nerve blocks if planned.

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

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Intubation might be difficult due to cleft palate, but mainly due to trachea-laryngeal abnormalities as laryngeal cleft. Downsizing of tracheal tube might be needed due to tracheal stenosis. Rarely, an extensive laryngo-tracheo-oesophageal cleft will cause the tracheal tube to repeatedly fall back from the trachea into the oesophagus through the cleft: the tube may need to be advanced further to prevent this, even into a bronchus.

If unrepaired cleft palate is present, more attention is needed to secure the tracheal tube properly to avoid inadvertent extubation during the surgery.

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

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No special recommendations reported.

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### **Particular preparation for anticoagulation**

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There is no reason for additional anticoagulation compared to general population.

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### **Particular precautions for positioning, transport or mobilisation**

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Lateral position for caudal blockade with leg flexed might help to avoid dural tap. As dural sac is also midline structure, might terminate lower than usual. The lateral position has been shown to move dural sac cranially compared to prone or supine positions.

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

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Not reported.

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### **Anaesthesiologic procedure**

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Inhalation or intravenous anaesthesia is suitable for the patients with GBBB syndrome. Due to associated laryngo-tracheal abnormalities, it is important to take preventable measures to avoid aspiration.

Due to risk of tracheal stenosis, the lower tube size should be considered and cuff pressure should be monitored closely.

In case of cardiac involvement, nitrous oxide should not be used and the doses of anaesthetic agents need to be tailored to patient needs to avoid impact on cardiac function.

The features of the syndrome have no impact on the use of muscle relaxants and its' reversal.

The use of opioids, non-opioid analgesic are not different to general population. Regional analgesia is beneficial, caution is required if neuraxial analgesia is to be used due to possible abnormalities (spine and the attached structures are also midline structures).

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### **Particular or additional monitoring**

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More invasive monitoring might be required if associated cardiac abnormalities are present.

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### **Possible complications**

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Higher risk of aspiration due to associated laryngeal cleft.

Tracheal stenosis in case of prolonged use of inappropriately large tracheal tube.

Due to associated risk of sacral abnormalities, it might be more difficult to perform caudal block with particular risk of dural tap and associated total spinal anaesthesia.

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### **Postoperative care**

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Prolonged ventilation should be avoided due to possible tracheal stenosis.

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### **Information about emergency-like situations / Differential diagnostics**

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Emergency situations may occur due to unrecognised cardiac anomalies or due to airway difficulties arising from extensive laryngeal clefts.

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### **Ambulatory anaesthesia**

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The surgeries who patients with Opitz G/BBB syndrome needs usually require overnight stay. However, some diagnostic or simple surgical procedures might be done in one day hospitalisation setting.

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### **Obstetrical anaesthesia**

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No special recommendations.

## Literature and internet links

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**Last date of modification: November 2016**

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*Please note that this guideline has not been reviewed by one anaesthesiologist, but two disease experts instead.*

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