orphanaesthesia

Anaesthesia recommendations for patients suffering from

Kabuki syndrome

Disease name: Kabuki syndrome

ICD 10: Q87.0

Synonyms: Kabuki make-up syndrome, Niikawa-Kuroki syndrome

Kabuki syndrome (KS) is a rare genetic disorder whose main clinical signs are multiple organ abnormalities and mental retardation. Niikawa et al. and Kuroki et al. simultaneously described this syndrome in a group of patients in 1981 [1,2]. Genetic transmission of KS is autosomal dominant in more than 50% of patients with an estimated incidence of 1 in 32,000 [3].

Niikawa et al. suggested the name of “Kabuki make-up syndrome” because the facial features of these patients were like the make-up actors in a Kabuki dance-drama in traditional Japanese theatre [1]. The facial features are characterized by an eversion of the lower lateral eyelid with long palpebral fissures, arched eyebrows, short columella, depressed nasal tip, and prominent ears.

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

Disease summary

Other manifestations include [4]:

- Postnatal growth retardation
- Microcephaly
- Mental retardation (mild-moderate)
- Seizures
- Ptosis
- Strabismus
- Hearing loss
- High arched/cleft palate
- Cleft lip
- Abnormal dentition
- Malocclusion
- Microdontia
- Feeding problems
- Diaphragmatic hernia
- Hernia (inguinal/umbilical)
- Renal anomalies
- Cryptorchidism
- Hypospadias
- Scoliosis
- Vertebral anomalies
- Hypermobile-loose joints
- Dislocation of hips/patella/shoulder
- Unusual dermatoglyphic patterns
- Short fifth finger
- Prominent finger pads
- Hypoglycemia
- Hypothyroidism
- Diabetes mellitus
- Premature thelarche
- Autism
- Autoimmune hemolytic anemia
- Idiopathic thrombocytopenic purpura (ITP)

Children with KS tend to develop obesity in later childhood [5]. These children may also have cardiovascular defects (atrial septal defect, coarctation of the aorta, patent ductus arteriosus, and transposition of the great vessels) with an incidence of 30–50% [6]. These patients also have susceptibility to infections such as recurrent otitis media, upper respiratory tract infections, and pneumonia [7]. Van Haelst et al. reported two cases, one with stenosis of the central airways and extrahepatic biliary atresia, the other with a congenital diaphragmatic hernia and severe bronchomalacia [8]. Another case with a tracheobronchial tree abnormality was a 6-yr-old child who presented with a right upper lobe bronchus originating from the trachea (tracheal bronchus) [9]. Another patient was reported with a small larynx [10]. Oto et al. reported a pulmonary haemorrhage in an adult patient with KS who presented with Henoch-Schönlein purpura; however, the authors were unsure about the relationship between the Henoch-Schönlein purpura and the KS. They attributed this complication to pulmonary hypertension secondary to an atrial septal defect [11]. Also, a latex allergy was reported in a single case [12].

The diagnosis for KS is primarily established by clinical findings. Mutations in the KMT2D gene (MLL2 gene) or the KDM6A gene have been reported. While a mutation in the KMT2D gene is inherited in an autosomal dominant pattern, a mutation in the KDM6A gene is inherited in an X-linked dominant pattern [13].

Typical surgery

Patients with KS can present for various surgical procedures like strabismus surgery, cleft lip/palate repair, bilateral myringotomy, cosmetic-restorative dental treatment, cosmetic surgery for prominent ears, congenital scoliosis correction, and treatment of hip dislocation. Other surgical treatments in KS patients may include corrective surgery for syndrome associated congenital heart defects, diaphragmatic hernia repair, gastrostomy tube placement, Nissen fundoplication, imperforate anus repair, and orchiopexy.

Type of anaesthesia

There is no definite recommendation for the type of anaesthesia to use in KS patients. Since KS is a genetic disorder that tends to involve multiple systems, and the symptoms can vary from patient to patient, clinical judgement concerning the anesthesia type can be made on a case-by-case basis. However, neuraxial anaesthesia can be difficult, especially in children with scoliosis and ITP. Patients with ITP often present with low platelet counts and the minimum platelet count for a safe neuraxial blockade has not been defined [14].

Necessary additional diagnostic procedures (preoperative)

Preoperative assessments of the patient's airway, cardiac, and pulmonary functions are essential. Since it is crucial to choose an appropriate anaesthetic agent for a patient who has poor cardiac functions, referral to a paediatric cardiologist should be considered for detection of suspected/diagnosed structural cardiac abnormalities. Although respiratory abnormalities are not common in KS, a preoperative evaluation of pulmonary function is useful for possible recurrent pneumonia and scoliosis.

These patients may have obstructive sleep apnea (OSA), therefore, the anaesthetist should be aware of the signs and symptoms of OSA while evaluating the patient preoperatively [1]. Furthermore, patients with low platelet counts should alert the anaesthetist to consult this issue with a paediatric haematologist to check for ITP.

Particular preparation for airway management

Difficult tracheal intubations can be expected in children with KS who have a high arched palate, abnormal dentition, cleft lip/palate, and malocclusions. Anticipation and preparation for a difficult airway management will reduce the risk of airway complications.

Particular preparation for transfusion or administration of blood products

Blood or blood products may be required if the patient undergoes scoliosis surgery. The major concern in a patient with ITP is perioperative bleeding, therefore, it is better to postpone surgery until the patient receives intravenous immunoglobulin preoperatively to achieve an adequate platelet count and good hemostasis [15].

Particular preparation for anticoagulation

None reported.

Particular precautions for positioning, transport or mobilisation

Caution must be taken while positioning the patient due to common joint laxity and dislocation.

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

Patients using anticonvulsant therapy may require a larger dose of nondepolarizing muscle relaxant during the operation [16].

Anaesthesiologic procedure

The majority of children with KS have hypotonia in which muscle biopsies have been normal. Neuromuscular blockade and malignant hyperthermia are important concerns with these patients. Remifentanil can be considered as an alternative to inhalation anesthesia [16].

Particular or additional monitoring

None reported.

Possible complications

The possibility of complications is related with the clinical manifestations of KS.

Postoperative care

None reported.

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

None reported.

Ambulatory anaesthesia

None reported.

Obstetrical anaesthesia

None reported.

Literature and internet links

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