orphanaesthesia

Anesthesia recommendations for patients suffering from

Russell-Silver-Syndrome

Disease name: Russell-Silver-Syndrome

ICD 10: Q87.1

Synonyms: Silver-Russell-Syndrome; Russell-Silver dwarfism

Russell-Silver-Syndrome is a rare disease and is characterized by growth retardation with antenatal onset, characteristic facies and limb asymmetry. One major clinical characteristic is relative macrocephaly. The incidence is evaluated at 1-30/100 000 cases and about 400 cases have been reported in the literature. It was first clinically described by Russel and Silver et al. in 1953/54 independently (Russel 1954; Silver et al. 1953). The clinical picture of RSS is heterogenous. Weight is often more affected than size, with little subcutaneous fat tissue. Bone maturation is delayed, in accordance with small stature. The fontanelle may be late to close. The skull has a normal circumference, which may contrast with the rest of the body and confers a pseudohydrocephalic appearance. The wide prominent forehead contrasts with the small, triangular face with a small pointed chin, a wide mouth with thin lips and down-turned corners, large eyes and bluish sclera. Lateral and usually partial asymmetry of the limbs is observed in 60% to 80% of cases, but is not progressive. Shortness and/or clinodactyly of the fifth fingers is a common finding. Patients may be slow to learn motor skills, and in rare cases, may be mildly intellectually deficient. They may have feeding difficulties and hypoglycaemia (Toutain 2007; Price et al. 1999).

The genetic aetiology of RSS is only partly understood. About 10% of RSS patients have maternal uniparental disomy (mUPD) for chromosome 7 and up to 50% have methylation defects in the imprinted domain on chromosome 11p15 (Abu-Amero et al. 2008).

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

Hypospadias are more frequent than in the general population. Therefore, hypospadia surgery can be regarded as a typical surgery in male SRS.

Apart from that entire pediatric spectrum.

Type of anaesthesia

All common techniques for regional and general anesthesia can be used.

Necessary additional diagnostic procedures (preoperative)

Recent blood electrolyte measurement because of possible hypoglycaemia.

Particular preparation for airway management

Anesthetic problems with these children can be profound and mostly concern a difficult airway due to facial dysmorphism (retrognathia or hypognathia) with a small mouth opening occasionally combined with over-riding maxillary dentition (Dinner et al. 1994; Hara and Matsunaga 2006; Imaizumi et al. 1986; Scarlett and Tha 2006). Mask ventilation can be difficult due to the small face/ mouth. Larnygoscopy is diffícult due to small mouth opening and a anterior and small larynx. Additionally subglottic stenosis is possible. Thus preparation for difficult airway management is recommended. It's wise to have a clinical pathway and technical devises for difficult airway management and to follow that algorithm. It can be recommended to prepare a smaller fiberoptic bronchoscope as suggested by the age of the child.

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transport or mobilisation

Not reported.

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

Not reported.

Anaesthesiologic procedure

All common techniques for regional and general anesthesia can be used.

Be aware of difficult airway.

Particular or additional monitoring

None.

Possible complications

It is recommended to be prepared for the management of a difficult airway as suggested by local clinical pathway. The young very thin SRS patients are prone to hypoglycaemia because their fasting tolerance is reduced. Hypoglycemia can happen before and after surgery.

Postoperative care

No special treatment is necessary.

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.:

Not reported.

Ambulatory anaesthesia

Ambulatory anesthesia is not recommended since there has to be an infrastructure to manage difficult airway.

Obstetrical anaesthesia

Not reported.

Literature and internet-links

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