orphanaesthesia

Anaesthesia recommendations for patients suffering from

Coffin-Siris syndrome

Disease name: Coffin-Siris syndrome

ICD 10: Q87.1

Synonyms: CSS

Coffin-Siris syndrome (CSS) is a rare congenital multi-systemic genetic disorder characterized by aplasia or hypoplasia of the distal phalanx or nail of the fifth digit, developmental delay, intellectual disability, coarse facial features, hypotonia, microcephaly, sacral dimple, spina bifida occulta, scoliosis/kyphosis, cleft palate, macroglossia, micrognatia, chronic respiratory upper infections, cardiac abnormalities and gastroesophageal reflux. More than 100 cases of confirmed CSS have been clinically and/or genetically reported to date. Until now, at least seven responsible genes for CSS have been identified; *SMARCB1*, *SMARCA4*, *SMARCE1*, *ARID1A*, *ARID1B*, *SOX11*, and *PHF6*. The inheritance mode of CSS by these genes is autosomal dominant. However, exact prevalence and incidence are not known but the disorder is probably under-recognized.

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

Cardiac surgery, nutritional gastrostomy, fundoplication, hernia repair, ENT procedures (mainly adenoidectomy) and dental surgery.

Type of anaesthesia

Due to the rarity of the disease, only few case reports or letters are available in medical literature. Two of these reports were published in Japan and written in Japanese language.

General anaesthesia with tracheal intubation or laryngeal mask was reported. Caudal block and spinal anaesthesia were reported.

Necessary additional diagnostic procedures (preoperative)

Cardiac function test including electrocardiography and echocardiography should be performed to evaluating congenital heart diseases.

If a central block is considered, ultrasounds or MR to detect spina bifida occulta or tethered cord should investigate the spine.

Upper airway infections are very common and timing of elective surgery should consider the best moment of the airway clearance.

Particular preparation for airway management

Difficult ventilation and difficult intubation were described. New-generation laryngoscopies and fiberoptic intubation should be available. Surgical airway might be required.

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

Some cases of uneventfully general anaesthesia, a case of light general anaesthesia combined with caudal block and a spinal anaesthesia for Caesarean section were reported. Some problems regarding mask ventilation and tracheal intubation were reported. Probably, the difficult in airway management due to their characteristic face (e.g. high and/or narrow palate) is age-dependent, increasing with the patient's age.

Particular or additional monitoring

Not described.

Possible complications

Obstruction of tracheal tube by massive bronchial secretion was reported.

Postoperative care

A case of post-operative apnoea was reported. Consider admission in post-operative intensive care unit. Take special care of intellectual disabilities and sometimes autism spectrum disorder that could lead to anxiety and hyperactivity during pre- and post-operative period.

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

Not recommended.

One case reports described an uneventfully spinal anesthesia in a 22-year-old pregnant for urgent Caesarean section. Although difficult airway predictors have been present, a direct laryngoscopy, performed in deep sedation, revealed a sufficient Cormack-Lehane score (6).

Literature and internet links

1. http://www.orpha.net
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6. Fornet I, Morillas P, López MA, Palacio FJ, Aguilar JM, Mesa JL.Emergency cesarean in a patient with Coffin-Siris syndrome. Rev Esp Anestesiol Reanim. 2007 Nov;54(9):563-5
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This guideline has been prepared by:

Author
Paolo Silvani, Anaesthesiologist, Ospedale San Raffaele, University "Vita–Salute", Milano, Italy
silvani.paolo@hsr.it

Peer revision 1
Noriko Miyake, Department of Human Genetics, Yokohama City University Graduate School of Medicine, Yokohama, Japan
nmiyake@yokohama-cu.ac.jp

Peer revision 2
Tomoki Kosho, Department of Medical Genetics, Shinshu University School of Medicine Matsumoto, Nagano, Japan
ktomoki@shinshu-u.ac.jp

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