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Stüve-Wiedemann syndrome

Systemic sclerosis

orphanaesthesia

a project of the German Society
of Anaesthesiology and Intensive Care Medicine

SUPPLEMENT NR. 17 | 2018

OrphanAnesthesia –
ein krankheitsübergreifendes Projekt des Wissenschaftlichen Arbeitskreises Kinderanästhesie der Deutschen Gesellschaft für Anästhesiologie und Intensivmedizin e.V.

Ziel des Projektes ist die Veröffentlichung von Handlungsempfehlungen zur anästhesiologischen Betreuung von Patienten mit seltenen Erkrankungen. Damit will Orphan Anesthesia einen wichtigen Beitrag zur Erhöhung der Patientensicherheit leisten.

Patienten mit seltenen Erkrankungen benötigen für verschiedene diagnostische oder therapeutische Prozeduren eine anästhesiologische Betreuung, die mit einem erhöhten Risiko für anästhesieassoziierte Komplikationen einhergehen. Weil diese Erkrankungen selten auftreten, können Anästhesisten damit keine Erfahrungen gesammelt haben, so dass für die Planung der Narkose die Einholung weiterer Information unerlässlich ist. Durch vorhandene spezifische Informationen kann die Inzidenz von mit der Narkose assoziierten Komplikationen gesenkt werden. Zur Verfügung stehendes Wissen schafft Sicherheit im Prozess der Patientenversorgung.

Die Handlungsempfehlungen von OrphanAnesthesia sind standardisiert und durchlaufen nach ihrer Erstellung einen Peer-Review-Prozess, an dem ein Anästhesist sowie ein weiterer Krankheitsexperte (z.B. Pädiater oder Neurologe) beteiligt sind. Das Projekt ist international ausgerichtet, so dass die Handlungsempfehlungen grundsätzlich in englischer Sprache veröffentlicht werden.

Ab Heft 5/2014 werden im monatlichen Rhythmus je zwei Handlungsempfehlungen als Supplement der A&I unter www.ai-online.info veröffentlicht. Als Bestandteil der A&I sind die Handlungsempfehlungen damit auch zitierfähig. Sonderdrucke können gegen Entgelt bestellt werden.

Bisher in A&I publizierte Handlungsempfehlungen finden Sie unter:

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OrphanAnesthesia –
a common project of the Scientific Working Group of Paediatric Anaesthesia of the German Society of Anaesthesiology and Intensive Care Medicine

The target of OrphanAnesthesia is the publication of anaesthesia recommendations for patients suffering from rare diseases in order to improve patients' safety. When it comes to the management of patients with rare diseases, there are only sparse evidence-based facts and even far less knowledge in the anaesthetic outcome. OrphanAnesthesia would like to merge this knowledge based on scientific publications and proven experience of specialists making it available for physicians worldwide free of charge.

All OrphanAnesthesia recommendations are standardized and need to pass a peer review process. They are being reviewed by at least one anaesthesiologist and another disease expert (e.g. paediatrician or neurologist) involved in the treatment of this group of patients.

The project OrphanAnesthesia is internationally oriented. Thus all recommendations will be published in English.

Starting with issue 5/2014, we'll publish the OrphanAnesthesia recommenations as a monthly supplement of A&I (Anästhesiologie & Intensivmedizin). Thus they can be accessed and downloaded via www.ai-online.info. As being part of the journal, the recommendations will be quotable. Reprints can be ordered for payment.

A survey of until now in A&I published guidelines can be found on:

www.ai-online.info/Orphsuppl
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orphananesthesia

Anaesthesia recommendations for patients suffering from

Stüve-Wiedemann syndrome

Disease name: Stüve-Wiedemann syndrome

ICD 10: Q78.8

Synonyms: (In older literature also known as Schwartz-Jampel type 2 syndrome)

Stüve-Wiedemann syndrome (SWS) is an autosomal recessively inherited disorder characterised by congenital skeletal dysplasia, and life-threatening autonomic nerve dysfunction. SWS is caused by a mutation in the leukemia inhibitory factor receptor (LIFR;151443) gene on chromosome 5p13.1. SWS has been reported in different ethnic groups including Europeans, North Africans, Gypsies and Arabs. However, it seems to be particularly common in the United Arab Emirates.

Clinical characteristics of SWS include bowing of the long bones (bent-bone dysplasia), camptodactyly, deformities of joints and extremities, facial dysmorphism, hypotonia, growth retardation, and difficulties with feeding and swallowing.

The clinical course is generally complicated by unexpected hyperthermic episodes, respiratory insufficiency and feeding difficulties. Therefore, the disease is associated with a poor life expectancy and most patients die early in life.

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

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SWS is caused by a mutation in the leukemia inhibitory factor receptor (LIFR;151443) gene on chromosome 5p13.1 [4]. It is particularly common in the United Arab Emirates population with a prevalence of 0.5/10,000 births. [5].

Clinical characteristics of SWS include bowing of the long bones (bent-bone dysplasia), hypertelorism, micrognathia, a single transverse palmar crease, trismus, camptodactyly, deformities of joints and extremities, facial dysmorphism, hypotonia, growth retardation, and difficulties with feeding and swallowing [6,7]. Neurological features resemble dysautonomia with temperature instability, reduced pain sensation, and absent corneal reflexes; however, intellectual capabilities are normal.

The clinical course is generally complicated by unexpected hyperthermic episodes, respiratory insufficiency and feeding difficulties [7,8]. Therefore it has a poor life expectancy and most patients die in early life. SWS may also be associated with cardiovascular abnormalities, especially pulmonary hypertension due to arterial wall abnormality [9].

Typical surgery

Orthopaedic (bone, joint and spine deformities) and eye surgery (cataract, corneal opacities).

There are a few other reports, as well as some theoretical risks, although the clinical significance of these are uncertain [20]. There is also a report of delayed tetraplegia after spinal surgery in two cases of SWS [22].

Type of anaesthesia

Only one report of a 3-year-old child that received uneventful sevoflurane anaesthesia five times.

Necessary additional diagnostic procedures (preoperative)

A thorough investigation of respiratory and cardiac function and reserves is mandatory. Several cardiac abnormalities have been noted in association with SWS [23] and, if present in a patient, an anaesthetic experienced in cardiac anaesthesia would be ideal.

Particular preparation for airway management

No specific deformities impairing airway management have been reported, but more difficult tracheal intubation may be anticipated. Plan a clinical pathway for induction of anaesthesia and prepare additional equipment for airway management and tracheal intubation.

Particular preparation for transfusion or administration of blood products

Follow usual guidelines.

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Particular preparation for anticoagulation

Follow usual guidelines.

Particular precautions for positioning, transport or mobilisation

Due to deformities of the extremities, positioning during anaesthesia should receive full attention.

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

No reported data available for SWS, but interactions are probable.

Anaesthesiologic procedure

No particular anaesthetic agents are advised or contraindicated, but extreme care with dosing of all drugs is essential.

Particular or additional monitoring

Continuous temperature monitoring during the whole perioperative period.

Respiratory and haemodynamic monitoring are very important due to the vulnerability of SWS patients in these organ functions.

Possible complications

Hyperthermic episodes	+
Respiratory insufficiency	+
Difficult airway/tracheal intubation	+
Haemodynamic disturbances	+
Pulmonary aspiration	+
Abnormal response to NMBDs	?
Malignant hyperthermia susceptibility	+?

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Postoperative care

Observation in a PACU or ICU setting is strongly advised depending on the preoperative problems and the course of surgery and anaesthesia.

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.:

No information.

Ambulatory anaesthesia

Not reported.

Obstetrical anaesthesia

Never reported. Pregnancy not probable.

Literature and internet links

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Attachment 1 Relationship with other diseases

There is a clinical overlap between SWS and Crisponi syndrome, especially concerning the unexpected hyperthermic episodes. Both are caused by mutations of genes involved in the ciliary neurotrophic factor (CNTF)-receptor pathway. The Crisponi syndrome is caused by a mutation in the cytokine receptor-like factor 1 (CRLF1) [13]. CRLF is involved in the CNTF-receptor pathway where the LIFR receptor is also part of [14]. These diseases cause unprovoked episodic high temperatures which reflect a systemic release of cytokines. Recently it has been hypothetised that those apparently unprovoked high temperatures may put the children at risk for sudden death [15].
When encountering a child with bowing of the long bones, one should also consider Campomelic dysplasia, Schwartz-Jampel syndrome and Ehlers-Danlos type IX syndrome as possible causes [5,7].
SWS may also be associated with mitochondrial disorders by ND1 gene mutations, leading to impaired oxidative phosphorylation [16,17]. It is unclear whether this is coincidence or a true relationship.

In textbooks and other literature, the risk of malignant hyperthermia susceptibility (MHS) in association with SWS has been speculated. There have been two case-reports which mention this possibility, but both are not very specific and the hyperthermic episodes were actually in patients with Schwartz-Jampel syndrome 1. This was reported when SWS was still considered identical to Schwartz-Jampel syndrome. One case-report dates from 1978 [18]. The patient of this report received atropine, ketamine, N_2O/O_2 and curare. She developed a temperature rise of 1.5°C within 10 minutes and a mild increase in creatinine kinase (216 $mU \cdot ml^{-1}$). The procedure was cancelled, the patient recovered and went home the following day. No muscle biopsy and IVCT test were taken to establish the diagnosis. Remarkably, no MHS triggering agents were used. The other case dates from 1974 [19]. This patient received N_2O/O_2 and halothane. During this procedure '*thermoregulatory control was disturbed resulting in a moderate hyperthermia*'. Neither could the diagnosis of MHS be confirmed in this case. Despite the confusion it should be realized that hyperthermic episodes are a hallmark of SWS and are the result of impairment of the ciliary neurotrophic factor receptor pathway without any relationship with the molecular or genetic background of MHS.

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