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PURA syndrome

Rubinstein-Taybi syndrome

orphanaesthesia

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

SUPPLEMENT NR. 11 | 2022

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:

www.ai-online.info/Orphsuppl
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OrphanAnesthesia –

a 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.

Find a survey of the recommendations published until now on:

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Anaesthesia recommendations for

Rubinstein-Taybi syndrome

Disease name: Rubinstein-Taybi syndrome

ICD 10: Q87.2

Synonyms: Michail-Matsoukas-Theodorou-Rubinstein-Taybi syndrome, Broad thumb-hallux syndrome, Rubinstein syndrome

Disease summary: Rubinstein-Taybi syndrome (RTS) has an estimated prevalence of 1 in 100,000 to 125,000. It is considered to have autosomal dominant patterns of inheritance but most cases result from de novo mutations. Mutation in the CREBBP gene on chromosome 16 which acts as a regulator of other genes in cell growth and division is the commonest mutation. A small percentage have mutations of a similar gene (EP300) and tend to have milder skeletal abnormalities. A deletion of the genetic material on the short arm of chromosome 16 which includes the CREBBP (16p13.3) is associated with severe forms. However, in 50 % of cases no genetic basis is found.

Major causes of deaths, particularly during the first year, are aspiration pneumonia and heart disease. There are no definite diagnostic features in RTS. However, the following are usually present:

- Short stature
- Moderate to severe intellectual disability
- Distinctive facial features (which become more prominent with age, include highly arched eyebrows, long eyelashes, down-slanting palpebral fissures, convex nasal ridge, highly arched narrow palate, abnormally large or "beak-shaped" nose, retrognathia and micrognathia). An unusual smile with almost complete closure of the eyes is present in most cases. These lead to an increase risk of dental problems and obstructive sleep apnoea (OSA). There is also an association with choanal atresia.
- Broad thumbs and 1st toes (often angulated in the varus/valgus position).

RTS is also frequently associated with the following:

- Eye abnormalities: Over 80 % of children with RTS have some form of eye abnormality, e.g. lacrimal duct obstruction (43 % are bilateral), ptosis and strabismus (55 %). Congenital glaucoma or glaucoma which develops in early life has been described.
- Congenital heart abnormalities 35–40 % have a single defect (e.g. ASD, VSD or PDA).

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- Renal and urinary tract: Renal anomalies are present in 50 % and for this reason all children should have renal ultrasound on diagnosis. Almost all boys will have incomplete or delayed descent of testes with hypospadias being present in 11 %. As a result of these anomalies, there is an increased risk of chronic kidney disease and urinary infections.
- Musculoskeletal: There is an increased risk of scoliosis, hyperkyphosis and spina bifida with general hypermobility.
- An increased risk of tumours (mainly leukaemia in childhood and meningioma in adulthood) has been observed. There is also an association with neuroendocrine tumours.
- An increased risk of keloid scarring.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

Find more information on the disease, its centres of reference and patient organisations on Orphanet: www.orpha.net

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Typical surgery

ENT: Adeno-tonsillectomy

Orthopaedic surgery: correction of scoliosis

Plastic surgery: correction of thumb and hallux anomalies craniofacial/orthodontic surgery

Ophthalmic: DCR, syringe and probe, squint surgery

Cardiac: dependent on cardiac defect

Urological: hypospadias repair/orchidopexy.

Type of anaesthesia

There is no definite recommendation for either general or regional anaesthesia. There is no evidence to support the use of TIVA vs. inhalational agents.

Regional anaesthesia can be performed. There are reports of spinal, epidural and caudal anaesthesia without complications. However, given the high incidence of spinal deformities, a proper risk analysis should be undertaken prior to embarking on neuroaxial blockade.

In patients with OSA, caution with or avoidance of sedation should be considered.

Necessary additional pre-operative testing (beside standard care)

Cardiac function tests including electrocardiography and echocardiography should be performed to evaluate the possibility of cardiac disease. Patients will usually have been reviewed by a paediatric cardiologist at diagnosis due to the frequency of associated cardiac anomalies.

Baseline renal function blood tests and ultrasound of the kidneys should have been performed as part of their initial diagnostic work-up.

Formal sleep studies (if signs and symptoms of OSA are elicited)

MRI/US of spine may have been performed and would be useful if considering caudal anaesthesia due to high incidence of tethered cord.

Particular preparation for airway management

Congenital tracheal stenosis and tracheomalacia has been well described and therefore it may be prudent to use a relatively smaller size of endotracheal tube.

Difficult intubation and ventilation should always be anticipated as craniofacial abnormalities may make laryngoscopy and intubation difficult, particularly in the presence of micrognathia or microsomia. Alternative methods of intubation such as nasal intubation and fibre-optic

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intubation have been described as well as the successful use of laryngeal mask airways including second generation and intubating laryngeal masks.

The association with OSA means that consideration should be given to awake vs. deep extubation.

There is a higher risk of aspiration due to the increased incidence of gastro-oesophageal reflux.

Particular preparation for transfusion or administration of blood products

There is no published evidence to support an increased transfusion requirement in patients with RTS.

Particular preparation for anticoagulation

There is no published evidence to support the need for particular anticoagulation. However, these patients are likely to have greater periods of post-operative immobility and this may lead to a higher risk of thrombosis.

Particular precautions for positioning, transportation and mobilisation

Cervical hyperkyphosis can be seen in 62 % of patients with RTS, while scoliosis can be seen in 38 %. This can make it challenging to position patients during intubation and for the surgical procedure. Attention should be given to pressure area care intraoperatively.

Patients may be prone to fractures and therefore care should be taken on transferring and positioning.

Interactions of chronic disease and anaesthesia medications

None reported.

Anaesthetic procedure

Avoid long-acting sedatives and be cautious with use of opioids (due to high incidence of OSA).

There are anecdotal reports on the occurrence of arrhythmias following the use of suxamethonium and it is generally accepted that this drug is best avoided. Although other muscle relaxants have been used without adverse event, however, in the presence of hypotonia it may be prudent to avoid them, if possible.

Both TIVA and inhalation anaesthesia have been used and there is no evidence to suggest that one is superior to the other in this group of patients.

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Antibiotic prophylaxis, appropriate for the planned surgery, should be given to those with an underlying cardiac lesion.

In patients with cardiac disease, the use of anticholinesterase and anticholinergic drugs may increase the risk of arrhythmia. For this reason, drugs such as atropine and neostigmine should be avoided, if possible, in this cohort of patients. When considering the use of muscle relaxants in patients with cardiac disease, the use of rocuronium and sugammadex in combination may be preferable.

Local anaesthetics have been used without complication.

Particular or additional monitoring

Facial nerve monitoring of neuromuscular blocking agents (NMBs) may be more appropriate as thumb anomalies and difficulty assessing adductor muscle response to stimulations exist.

In cases of high-risk surgery, with major fluid shifts, arterial cannulation and central venous cannulation for invasive pressure measurements may be useful.

Possible complications

Sedative drugs and opioids may cause post-operative respiratory depression; therefore, these drugs should be carefully titrated to avoid possible complications. The use of regional anaesthesia may be beneficial in this regard.

Airway obstruction.

Aspiration pneumonia.

Post-operative care

The degree of post-operative monitoring depends on surgical procedure and pre-operative condition of the patient. ICU is not mandatory.

Disease-related acute problems and effect on anaesthesia and recovery

Preparation for the possible complications as outlined above.

There are no reports of idiosyncratic drug reactions in relation to anaesthesia.

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Ambulatory anaesthesia

Published data regarding ambulatory anaesthesia for RTS is limited.

Obstetrical anaesthesia

Published data is limited. Females with RTS have normal fertility. Their management should be tailored to their individual needs. Previous spinal surgery or spinal anomalies may preclude or limit neuroaxial blockade. Remifentanil PCA may be an option for intrapartum pain relief.

Active planning for a potential difficult airway should be in place.

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Please note that this recommendation has not been reviewed by an anaesthesiologist but by two disease experts instead.

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