orphanaesthesia

Anaesthesia recommendations for

Central Core Disease

Disease name: Central Core disease

ICD 10: G71.2

Synonyms: Shy-McGee syndrome

Disease summary: Central core disease (CCD) is an inherited (mostly dominant) neuro-muscular disorder characterised by central cores in type I fibres on muscle biopsy and clinical features of a congenital myopathy. Prevalence is unknown because of variable expression and incomplete penetrance. Associated in approximately 25% of cases with malignant hyperthermia because of gene proximity or overlap: the mutation then involves gene RYR1 (19q13.1-13.2). In case of rare recessive transmission gene MYH7 (14q11.1) is involved. Multiminicore, minicore myopathy and core-rod myopathy are closely related to Central core disease and probably carry the same risk for malignant hyperthermia.

CCD typically presents in infancy with hypotonia and motor developmental delay and is characterized by – typically non-progressive – predominantly proximal muscle weakness, pronounced in the hip girdle.

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

Typical surgery

Muscle biopsy; Orthopaedic surgery: correction of talipes equinovarus, scoliosis or dislocation of the hip or patella.

Type of anaesthesia

Succinylcholine and volatile anaesthetics have to be strictly avoided.

General anaesthesia performed as total intravenous anaesthesia or regional anaesthesia can both be done without complications. There are reports about the successful performance of spinal as well as epidural anaesthesia or a combination of both.

There is no contraindication for (analgo-)sedation beside common restrictions.

Necessary additional pre-operative testing (beside standard care)

Cardiac function is not typically impaired in patients with CCD, so preoperative cardiac function tests are not obligatory. There are some scarce reports of cardiac involvement. In this case and in case of severe scoliosis, preoperative echocardiography is necessary.

In some cases of neonatal onset, the respiratory system is impaired and lung function tests may be considered. This is also recommended in case of severe scoliosis.

If muscular weakness is present and regional anaesthesia is planned, neurological consultation is helpful for legal reasons.

Determination of preoperative creatine kinase level is not mandatory, but may be helpful in case of perioperative complications (eg. rhabdomyolysis or Malignant Hyperthermia).

Particular preparation for airway management

Manibular hypoplasia and impaired cervical mobility (kyphoscoliosis) may be found secondary to muscle weakness. Scrutiny for any stigmata of a difficult airway is advisable.

Particular preparation for transfusion or administration of blood products

There is one small study showing a higher intraoperative blood loss during surgery for scoliosis in patients with neuromuscular diseases in general compared to idiopathic scoliosis. In CCD, there is no evidence about bleeding abnormalities.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilisation

Not reported.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Strictly avoid succinylcholine and any volatile anaesthetic because of the risk for malignant hyperthermia.

Use of opiates (remifentanil, alfentanil, fentanyl, morphine), intravenous anaesthetics (propofol, midazolame), nitrous oxide, local anaesthetics (ropivacaine, bupivacaine) and non-depolarizing muscle relaxants (rocuronium, pancuronium) has been reported without complications.

When using non-depolarizing muscle relaxants, no prolonged neuromuscular blockade is reported. Antagonisation of neuromuscular blockade with neostigmine or reversal with sugammadex® (in case if rocuronium or vecuronium were used) have been reported as successful.

There is no need for strictly prophylactic postoperative ventilation.

Particular or additional monitoring

Although there are no reports about prolonged effects of non-depolarizing muscle relaxants, it cannot be ruled out from a pathophysiological view. Therefore, monitoring of the neuro-muscular blockade is recommended.

Possible complications

All CCD patients are highly susceptible for malignant hyperthermia.

Post-operative care

Avoid prolonged immobilization. Accompanying muscular atrophy may worsen disease.

The degree of postoperative monitoring and care is depending on the surgical procedure and on the preoperative condition of the patient.

Disease-related acute problems and effect on anaesthesia and recovery

If you suspect Malignant Hyperthermia, treat as soon as possible and aggressively.

Acute respiratory tract infections may impair respiratory function more than usual.

Ambulatory anaesthesia

In cases of stable disease without respiratory impairment, ambulatory anaesthesia is possible according to common guidelines.

Obstetrical anaesthesia

Obstetrical anaesthesia can be done in general (without succinylcholine and volatiles) as well as regional anaesthesia. Be aware that in some neuromuscular disorders, a disease progression can occur during pregnancy.

Use of syntocinon was reported without complications.

After the use of dantrolene (treatment in case of Malignant Hyperthermia) uterine atony is reported.

Because of inheritance of the disease, there is the possibility of impaired newborns with muscular hypotonia or respiratory distress.

References

1. Akiyama C, Nonaka I. A follow-up study of congenital non-progressive myopathies. Brain Dev 1996;18(5):404–408
2. Almenrader N, Patel D. Spinal fusion surgery in children with non-idiopathic scoliosis: is there a need for routine postoperative ventilation? Br J Anaesth 2006;97(6):851–857
3. Avila G. Intracellular Ca2+ dynamics in malignant hyperthermia and central core disease: established concepts, new cellular mechanisms involved. Cell Calcium 2005;37(2):121–127
4. Baur CP, Schara U, Schlecht R, Georgieff M, Lehmann-Horn F. [Anesthesia in neuromuscular disorders. Part 2: specific disorders]. Anasthesiol Intensivmed Notfallmed Schmerzther 2002;37(3):125–137
5. Brambrink AM, Kirsch JR. Perioperative care of patients with neuromuscular disease and dysfunction. Anesthesiol Clin 2007;25(3):483–509, viii-ix
6. Brini M. Ryanodine receptor defects in muscle genetic diseases. Biochem Biophys Res Commun 2004;322(4):1245–1255
7. Cohen ME, Duffner PK, Heffner R. Central core disease in one of identical twins. J Neurol Neurosurg Psychiatry 1978;41(7):659–663
8. Docquier MA, Veyckemans F, Prudhomme S, Rossillon R. [Anesthesia in a child presenting a anhydrotic ectodermic dysplasia associated with a multiminicore myopathy.]. Can J Anaesth 2000;47(5):449–453
9. Fananapazir L, Dalakas MC, Cyran F, Cohn G, Epstein ND. Missense mutations in the b-myosin heavy chain gene cause central core disease in hypertrophic cardiomyopathy. Proc Natl Acad Sci 1993;90:3993–3997
10. Finsterer J, Stollberger C. Cardiac involvement in primary myopathies. Cardiology 2000;94(1):1–11
11. Flick RP, Gleich SJ, Herr MM, Wedel DJ. The risk of malignant hyperthermia in children undergoing muscle biopsy for suspected neuromuscular disorder. Paediatr Anaesth 2007;17(1):22–27
12. Foster RN, Boothroyd KP. Caesarean section in a complicated case of central core disease. Anaesthesia 2008;63(5):544–547
13. Georgiou AP, Gatward J. Emergency anaesthesia in central core disease. Br J Anaesth 2008;100(4):567
14. Gozal D. Pulmonary manifestations of neuromuscular disease with special reference to Duchenne muscular dystrophy and spinal muscular atrophy. Pediatr Pulmonol 2000;29(2):141–150
15. Hackenberg T. Heart transplantation in a patient with central core disease. J Cardiothorac Vasc Anaesth 1992;6:386–387
16. Harper CM, Ambler G, Edge G. The prognostic value of pre-operative predicted forced vital capacity in corrective spinal surgery for Duchenne's muscular dystrophy. Anaesthesia 2004;59(12):1160–1162
17. Johi RR, Mills R, Halsall PJ, Hopkins PM. Anaesthetic management of coronary artery bypass grafting in a patient with central core disease and susceptibility to malignant hyperthermia on statin therapy. Br J Anaesth 2003;91(5):744–747
18. Jungbluth H. Central core disease. Orphanet J Rare Dis 2007;2:25
19. Loke J, MacLennan DH. Malignant hyperthermia and central core disease: disorders of Ca2+ release channels. Am J Med 1998;104(5):470-486.
20. Muenster T, Forst J, Goerlitz P, Schmitt HJ. Reversal of rocuronium-induced neuromuscular blockade by pyridostigmine in patients with Duchenne muscular dystrophy. Paediatr Anaesth 2008;18(3):251–255
21. Otsuka H, Komura Y, Mayumi T, Yamamura T, Kemmotsu O, Mukaida K. Malignant hyper-thermia during sevoflurane anesthesia in a child with central core disease. Anesthesiology 1991;75(4):699–701
22. Perrot A, Spuler S, Geier C, Dietz R, Osterziel KJ. Cardiac manifestations in muscular dystrophies. Z Kardiol 2005;94:312–320
23. Polat M, Tosun A, Ay Y, Ozer E, Serdaroglu G, Aydogdu S et al. Central core disease: atypical case with respiratory insufficiency in an intensive care unit. J Child Neurol 2006;21(2):173–174

24. Rudnik-Schoneborn S, Glauner B, Rohrig D, Zerres K. Obstetric aspects in women with facioscapulohumeral muscular dystrophy, limb-girdle muscular dystrophy, and congenital myopathies. Arch Neurol 1997;54(7):888–894
25. Sax TW, Rosenbaum RB. Neuromuscular disorders in pregnancy. Muscle Nerve 2006;34(5):559–571
26. Shapiro F, Sethna N. Blood loss in pediatric spine surgery. Eur Spine J 2004;13 Suppl 1:S6–S17
27. Waikar PV, Wadsworth R. A patient with severe central core disease. Br J Anaesth 2008;101(2):284
28. Weingarten AE, Korsh JI, Neuman GG, Stern SB. Postpartum uterine atony after intravenous dantrolene. Anesth Analg 1987;66(3):269–270.

Date last modified: **October 2019**

This recommendation was prepared by:

Author

Tino Münster, anaesthesiologist, Department of anaesthesiology and intensive care medicine, Hospital Barmherzige Brüder, Regensburg, Germany
Tino.Muenster@barmherzige-regensburg.de

Disclosure The author has no financial or other competing interest to disclose. This recommendation was unfunded.

This recommendation was reviewed by:

Reviewer 1

Francis Veyckemans, anaesthesiologist, University-hospital St. Luc, Bruxelles, Belgium
francis.veyckemans@uclouvain.be

Reviewer 2

Maggie Walter, neurologist, Friedrich-Baur-Institute, Ludwig-Maximilians-University of Munich, Germany
maggie.walter@lrz.uni-muenchen.de

Benedikt Schosser, neurologist, Friedrich-Baur-Institute, Ludwig-Maximilians-University of Munich, Germany
benedikt.schosser@med.uni-muenchen.de

Editorial review 2019

Johannes Prottengeier, anaesthesiologist, University-Clinic Erlangen-Nuernberg, Germany
Johannes.Prottengeier@googlemail.com

Disclosures The reviewers have no financial or other competing interest to disclose.