orphanaesthesia

Anaesthesia recommendations for patients suffering from

Smith-McCort Dysplasia (SMC)

Disease name: Smith-McCort Dysplasia (SMC)

ICD 10: -

Synonyms: Smith-McCort dwarfism, Orpha no: ORPHA178355

SMC, which was first described in 1958, is an autosomal recessive skeletal dysplasia. The disease bears similarity to Dyggve-Melchior-Clausen (DMC) syndrome except that patients with SMC have normal intelligence function, whereas patients with DMC have moderate to severe intellectual disabilities. The DMC gene is mapped to the 18q12-12.1 chromosomal region. SMC (OMIM: 607326) also mapped to this same region and as it turns out both conditions have mutations in the DYM gene. DMC results from mutations in the gene that results in loss of function, whereas in SMC, the mutation is a missense mutation resulting in residual DYM activity, thus a less severe phenotype. The gene codes for a novel protein known as Dymeclin, which is involved in Golgi organization and intracellular trafficking.

Recently, another gene, RAB33B, which decreases the level of another Golgi protein that is involved in retrograde transport of Golgi vesicles, has been implicated in the pathogenesis of SMC.

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

Disease summary

Patients with SMC have increased glycosaminaglycans (GAGs) in their urine but have normal electrophoretic patterns and enzyme assays. Lysosomal pathways are also normal in patients with SMC.

Patients with this syndrome have a severe generalized bony dysplasia with a short trunk, short stature, coarse facies, a short neck and a protuberant abdomen. Specific skeletal abnormalities include hypoplastic odontoid, pectus carinatum, kyphoscoliosis, platyspondyly, exaggerated lumbar lordosis, flared ribs, widened costochondral junction, small scapulas with concave inferior angles, small pelvis, hypoplastic acetabulums with small ilium, dislocated hips, widened symphysis pubis, deformed and displaced femoral neck, genu valgum or varum, rhizomelic limb shortening and limited joint extensibility. Pathognomic radiologic features include double-humped vertebral end plates and lace-like appearance of iliac crests.

Typical surgery

Atlantoaxial dislocation-induced spinal cord compression is a serious and treatable problem in these patients. Common surgeries for which these patients might present include posterior cervical spine fusion, orthopaedic femoral osteotomy, total hip arthroplasty, early meniscectomy and realignment osteotomy. However, we recommend postponement of any surgery (unless emergent) till adolescence in view of risks associated with anaesthesia and doubtful long term results in case of orthopaedic surgeries.

Type of anaesthesia

No particular recommendations are available regarding the type of anaesthesia, i.e., general versus regional. Congenital spine abnormalities must be ruled out if spinal or epidural block is planned. Moreover, if associated cervical spine disease has led to muscle weakness or spasticity, the same should be documented before contemplating regional anaesthesia.

Necessary additional diagnostic procedures (preoperative)

Chest radiographs and pulmonary function tests should be done if there are co-existing kyphoscoliosis and chest abnormalities. A blood gas analysis on room air may be useful for comparison at a later stage. Electrocardiography and echocardiography will help to rule out any congenital cardiac anomalies or cardiac dysfunction.

Particular preparation for airway management

A difficult airway may be expected in patients with Smith-McCort dysplasia as these individuals may have macroglossia, short neck and abnormality of neck flexion. But surprisingly, laryngoscopy may not be problematic. However, intubation may be possible only with smaller than expected endotracheal tube. Therefore, several sizes of endotracheal tubes should be readily available.

In a case report of anaesthesia in a patient with DMC, the authors did not find any airway difficulties. With one SMC patient with whom we have been involved, we anticipated difficulty with anesthesia and planned for fiberoptic intubation. However, on direct laryngoscopy, the vocal cords could be visualized without difficulty. We, however, could negotiate only a 5 mm uncuffed endotracheal tube into this 18-year-old patient.

In cases where a tracheostomy is required, post tracheostomy airway problems should be anticipated. In SMC patients, the biggest challenge is maintaining the position of the artificial airway because of distorted airway anatomy. Hence, wherever possible, direct extubation is preferable and tracheostomy should be performed only in unavoidable circumstances. Once a tracheostomy has been performed, all measures should be directed at appropriate positioning to prevent accidental misalignment of the tracheostomy tube.

Particular preparation for transfusion or administration of blood products

No recommendations are available.

Particular preparation for anticoagulation

No recommendations are available.

Particular precautions for positioning, transport or mobilisation

Not reported.

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

Not reported.

Anaesthesiologic procedure

Because the patient with Smith-McCort dysplasia has dwarfism, anthropometric measurements such as height and weight should be recorded and drug doses should be modified accordingly. Baseline complete haemogram and biochemistry profile should also be done.

Particular or additional monitoring

Depending on the surgery and expected blood loss, arterial blood pressure monitoring can be used and adequate intravenous access should be obtained.

Possible complications

Extubation should be performed carefully and only when the patient is fully awake. Doing so will reduce the need for reintubation especially if the surgery involved fusion of the cervical spine or if airway edema is expected post procedure, situations where reintubation could be even more difficult.

Postoperative care

The degree of postoperative monitoring will depend on the surgical procedure, the preoperative condition of the patient and the intraoperative course. When postoperative ventilation is needed, avoid prolonged ventilation and attempt to wean rapidly. Extubation should be attempted at the earliest possibility and tracheostomy should not be chosen as an easy alternative.

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 disease

Morquio syndrome (mucopolysaccharidosis type IVA): This disorder has similar clinical and radiologic findings but urinary mucopolysaccharide secretion is normal in SMC. Patients with this syndrome usually have corneal clouding, deafness and cardiac anomalies. Morquio syndrome is caused by a deficiency of N-acetyl-galactose-6 sulfatase enzyme. In both conditions, hypoplastic odontoid can be present.

Hurler syndrome (MPS type I): Occurs due to deficiency of α-L-iduronidase. Individuals with this condition also excrete dermatan sulfate and heparin sulfate in their urine.

Spondyloepimetaphyseal dysplasias (SEMDs): There is absence of the vertebral double hump with central constriction. In SEMDs, some vertebras show anterior pointing with narrow disc spaces, premature degenerative changes and no odontoid hypoplasia.

Achondroplasia: The vertebras are short and flat, pedicles are short and the spinal canal is narrow.

Metatrophic dysplasia (MD): This condition causes general shortening in all long and short tubular bones. The lace-like appearance of iliac crests in SMC is a significant sign for distinguishing between MD and SMC.

Ambulatory anaesthesia

Not reported.

Obstetrical anaesthesia

Not reported.

Literature and internet links

1. Mona SA, Samia AT, Ekram F et al. Dyggve–Melchior–Clausen syndrome: clinical, genetic, and radiological study of 15 Egyptian patients from nine unrelated families. J Child Orthop 2009; 3:451-8
2. Cohn DH, Ehtesham N, Krakow D, et al. Mental retardation and abnormal skeletal development (Dyggve-Melchior-Clausen dysplasia) due to mutations in a novel evolutionarily conserved gene. Am J Hum Genet 2003; 72:419-28
3. Nakamura K, Kurokawa T, Nagano A, Nakamura S, Taniguchi K, Hamazaki M. Dyggve-Melchior-Clausen syndrome without mental retardation (Smith-McCort dysplasia): morphological findings in the growth plate of the iliac crest. Am J Med Genet 1997; 72:11-7
4. Bayrak IK, Nural MS, Diren HB. Dyggve-Melchior- Clausen syndrome without mental retardation (Smith- McCort dysplasia). Diagn Intervent Radiol 2005; 11:163-5
5. Kandziora F, Neumann L, Schnake KJ et al. Atlantoaxial instability in Dyggve-Melchior-Clausen syndrome: Case report and review of the literature. J Neurosurgery: Spine 2002; 96:112-7
6. Paupe V, Gilbert T, Merrer ML et al. Recent advances in Dyggve-Melchior- Clausen syndrome. Molecul Genet Metabol 2004; 83:51-9
7. Eguchi M, Kadota Y, Yoshida Y, Masuda M, Masuyama T, Kammura Y. Anesthetic management of a patient with Dyggve-Melchior-Clausen syndrome. Masui 2001; 50:1116-7
8. Schorr S, Legum C, Ochshorn M, Hirsch M, Moses S, Lasch EE, El- Masri M. The Dyggve-Melchior-Clausen Syndrome. Am J Roentgenol 1977; 128:107-13
9. Dupuis N, Lebon S, Kumar M, Drunat S, Graul-Neumann LM, Gressens P, El Ghouzzi V. A novel RAB33B mutation in Smith-McCort dysplasia. Hum Mutat 2013; 34:283-6.

Last date of modification: February 2015

These guidelines have been prepared by:

Authors
Sonia Bansal, Anaesthesiologist, National Institute of Mental Health and Neurosciences, Bangalore, India
rashibpgi@yahoo.co.in
GS Umamaheswara Rao, Anaesthesiologist, National Institute of Mental Health and Neurosciences, Bangalore, India

Peer revision 1
Serge Dalmas, Anaesthesiologist, Hôpital Jeanne-de-Flandre, Lille, France
Eric Nectoux, Département de Chirurgie et Orthopédie de l'Enfant, Hôpital Jeanne de Flandre, Lille, France
Eric.NECTOUX@CHRU-LILLE.FR

Peer revision 2
David Weaver, Department of Medical and Molecular Genetics, Indiana University School of Medicine, Indianapolis, USA
dweaver@iupui.edu