Clinical Reports

Anaesthetic management of a child with Marshall-Smith syndrome

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Purpose: The Marshall-Smith Syndrome (MSS) is a rare disease characterized by orofacial dysmorphism, failure to thrive, accelerated osseous maturation and mental retardation. It has anaesthetic implications due to upper airway problems and possible atlanto-axial instability. We present the perioperative problems (difficult intubation, airway obstruction) encountered in a child with MSS who underwent several anaesthetics during his first two years of life.

Clinical features: At birth, the child presented with asphyxia due to obstructive apnoea. His trachea was, therefore, intubated immediately. The morphological diagnosis of MSS was confirmed by the pathognomonic radiological appearance of the bones (bone age was eight months at the age of four days). Upper airway difficulty was caused by functional problems at the level of the hypopharynx (inspiratory collapse at the level of the velum palatinum), and was solved by the use of a nasopharyngeal airway (NPA) during the induction of anaesthesia and early postoperative period.

Conclusion: The use of an NPA during both induction and recovery of anaesthesia may be particularly useful to prevent upper airway problems in children with MSS.

Objectif : Le syndrome de Marshall-Smith (SMS) est une maladie rare d'étiologie inconnue, caractérisée par une dysmorphie faciale, un retard de croissance, une maturation osseuse accélérée et un retard mental. Il a des implications anesthésiques liées à une possible instabilité cervico-atlantoïdienne et à une obstruction chronique des voies aériennes supérieures. Nous présentons les problèmes périopératoires (intubation difficile, obstruction des voies aériennes) rencontrés chez un enfant atteint de ce syndrome, et qui a bénéficié de plusieurs anesthésies durant ses deux premières années de vie.

Éléments cliniques : Le patient a présenté des problèmes respiratoires qui ont nécessité une intubation dès la naissance. Le diagnostic morphologique de SMS a été confirmé par l'aspect très caractéristique, voire pathognomonique, de la radiologie des mains et par l'âge osseux qui était de huit mois à l'âge de quatre jours. Les problèmes respiratoires étaient dus à un dysfonctionnement vélo-palatin; l'induction et les premières heures postopératoires ont été facilitées par la mise en place prophylactique d'un tube nasopharyngé (TNP), lors de chaque anesthésie.

Conclusion : Dans notre expérience, l'utilisation du TNP à l'induction et en postopératoire s'est révélée particulièrement utile pour résoudre les problèmes d'obstruction des voies aériennes supérieures rencontrés dans le SMS.

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WE report the case of a child with MSS, a rare disease of unknown aetiology, which is characterized by typical orofacial dysmorphism, failure to thrive, respiratory problems and accelerated osseous maturation. This child underwent five operations under general anaesthesia and presented respiratory problems due to functional obstruction of the upper airway.¹

Case report

The patient, the second child of parents in good health, was born at term by Caesarean section (BW: 2.800 g) with an Apgar score of 1 at 5 min because of asphyxia due to obstructive apnoea. His trachea was intubated immediately. Laryngoscopy revealed a hypoplastic epiglottis and a barely visible glottic aperture. Mechanical ventilation was provided for approximately 12 hr. After several attempts of extubation, each followed by severe upper airway obstruction, a nasopharyngeal airway (NPA) was inserted and left in place.

Clinical examination revealed: (Figure 1) hypertelorism with downslanting palpebral fissures, prominent eyes and forehead, largely opened frontal suture, flat nasal bridge with anteverted nostrils, micrognathia and a high-arched palate, large overflexed ears, large hands with long fingers, longilineal appearance with an underdeveloped musculature, and cryptorchidism to the right.

The morphological diagnosis of MSS was confirmed by the pathognomonic radiological appearance of the bones. First, bone age was eight months at the age of four days. Second, hand radiographs showed widened first and second phalanges of the last four fingers while the third phalanx had a slender and fine appearance; the metacarpals appeared dysmorphic with a stocky and massive appearance. Moreover, the radiographic examination of the vertebrae showed multi-levelled platyspondylia and a relatively large odontoid process of C2 but no atlanto-axial subluxation during anteflexion of the neck. Ultrasonic examination of the brain confirmed the presence of the corpus callosum but showed dilatation of both ventricles with an aspecific dysmorphic appearance. Chest X-ray showed an hypoplastic thorax. At the age of 21 days, diagnostic laryngoscopy was performed under general anaesthesia (GA) with halothane administered via the NPA before bronchoscopy. It showed a rudimentary and short epiglottis but no abnormality of the larynx nor of the trachea. An attempt of extubation failed because of severe obstructive apnoea: a NPA was re-inserted and left in place day and night.

At one month, the child developed pyloric stenosis and was operated upon. After induction of anaesthesia

FIGURE 1 Lateral view of the patient when aged eight months.

FIGURE 2 Lateral scan of the child when three years old: it shows microretrognathia and normal cervical spine.

(Courtesy from Drs Ph. Clapuyt and Ch. Saint Martin, Department of Paediatric Radiology)

with halothane via the NPA and using gentle cricoid pressure, orotracheal intubation was completed at the first attempt but without visualization of the glottis, arytenoids or posterior commissure. Before extubation, fiberoptic examination of the upper airway and of the nose was performed and showed stenosis of the choanae and an inspiratory collapse at the level of the velum palatinum. The insertion of a NPA at the end of the examination allowed maintenance of upper airway permeability. It was, therefore, left in place all the time for several days. Later, it was withdrawn during the day-time and inserted before the night. At five months, ophthalmological examination was performed under GA with halothane. After induction using a NPA, tracheal

intubation was again achieved at the first attempt, although no glottic structure was visualized at laryngoscopy. The trachea was extubated without any problem, a NPA being left in place prophylactically. Because of swallowing difficulty, exclusive gavage-feeding was necessary until the age of six months. At 11 mo, another ophthalmological examination was performed under general anaesthesia. Inhalational induction, without a NPA, was complicated by severe upper airway obstruction which necessitated emergency tracheal intubation which was achieved, once again, at the first attempt but without visualization of the glottic structures. During recovery, major inspiratory stridor necessitated the insertion of an NPA which was left permanently in place for several days. At 14 mo, a central venous catheter was placed percutaneously to allow antibiotic therapy of a pneumonia. This procedure was undertaken uneventfully under general anaesthesia with sevoflurane administered via an NPA and local anaesthetic infiltration. At 18 mo, adenotonsillectomy, an ophthalmological examination and orchidopexy were performed. Inhalational induction was performed with sevoflurane administered via an NPA. Intubation was completed at the first attempt, but again without visualizing the glottic structures. During the few postoperative days, an NPA was left in place day and night, then only at night because the problem of functional obstruction of the upper airway during sleep persisted despite the adenotonsillectomy (Table). At 18 mo, the bone age was of five to six years. At the time of writing, the child still presents some behavioural disorders but his physical development is satisfactory. Figure 2 shows the tridimensional scan of the child's head when he was three years old.

Discussion

The syndrome

In 1971, Marshall *et al.* described a new syndrome characterized by accelerated osseous maturation, orofacial dysmorphism and failure to thrive associated with mental retardation.¹ The facial dysmorphism includes prominent eyes and forehead, megalocornea, micrognathia, and anteverted nostrils.² The syndrome is of unknown aetiology and the risk of relapse in siblings is low, similar to that of a sporadic entity. Mortality is very high during the first two years of life (11 cases out of the 14 reported in the 90s). The main causes of death are recurrent pulmonary infections caused by laryngeal incompetence and/or pulmonary hypertension caused by chronic upper airway obstruction.³ It is of outmost importance that neonates and infants with MSS be managed early in the same way as neonates with the Pierre-Robin sequence in order to avoid the development of cor pulmonale and pulmonary hypertension caused by chronic upper airway obstruction.⁴

TABLE

age	anaesthetic agent	intubation	glottic aperture	NPA
birth	no	1	barely visible	R
21 days	H	no	-	I/R
1 mo	H	1	not seen	I/R
5 mo	H	1	not seen	I/R
11 mo	H	1 (emergency)	not seen	R (late)
14 mo	S	no	-	I/R
15 mo	S	1	not seen	I/R

Legend: H = halothane S = sevoflurane I = induction R = recovery
No = not intubated 1 = first attempt NPA = nasopharyngeal airway

Difficult intubation due to laryngeal anomalies is also described in MSS.⁵ Orthopaedic problems are frequent: mainly scoliosis and atlanto-axial instability which may induce medullary compression.⁶ Other abnormalities have been reported: hypotonia, choanal atresia, umbilical hernia, cardiovascular problems, hydronephrosis and immunological abnormalities.⁷ Children with MSS differ from those with Weaver-Syndrome⁸ who have a similar oro-facial dysmorphism but a superior life expectancy: X-Ray examination of the hands allows differentiation of both syndromes. In our case, the child presented a dysfunction of the velum palatinum that obstructed the upper airway. The presence of an NPA allowed maintenance of upper airway patency by stenting the membranous palate and thus preventing hypopharyngeal collapse. The use of an NPA in the postoperative care of cleft palate repair to prevent the closure of nasal airway and the collapse of the hypopharynx has already been described.⁹ Benjamin and Walcar¹⁰ described its use in the Pierre-Robin sequence.

Anaesthetic management

From the anaesthetist's point of view, taking care of children affected by MSS needs special attention. First, the facial dysmorphism may cause difficulty during ventilation with a face mask as well as for intubation. It is necessary to have a small fiberoptic bronchoscope (2.5 mm) and a laryngeal mask airway ready for use. Ideally, spontaneous ventilation should be maintained during induction of anaesthesia until the airway is controlled. Second, possible atlanto-axial instability can entail medullary compression during extension and/or flexion of the cervical spine: manipulation of the head and manoeuvres for intubation should be undertaken with great care and, if possible, after X-ray evaluation of that area. Third, functional problems at the level of the hypopharynx can be solved and prevented by the use of an NPA especially

during the induction of anaesthesia and in the early postoperative period, when the increased sensitivity of the upper airway muscles to anaesthetic agents may favour obstruction.¹¹ Most textbooks recommend that the appropriate length of an NPA is the distance from the nares to the tragus of the ear.¹² However, in case of oro-facial dysmorphism, the correct length of the NPA should be estimated clinically: when the tube is in the nasopharynx, maximal breath sounds indicate that the tube is correctly placed behind the tongue and above the epiglottis. Moreover, good air entry, the absence of stridor and retractions and, if available, the presence of a capnogram confirm proper positioning of the NPA. Fourth, it is necessary to take into account the associated hypoplastic thorax, which causes a restrictive syndrome and therefore a reduced reserve in oxygen.

In conclusion, MSS is characterized clinically by oro-facial dysmorphism, failure to thrive, accelerated osseous maturation and mental retardation. As far as the anaesthetist is concerned, anatomic abnormalities of the larynx, functional upper problems airway, possible atlantoaxial instability and a restrictive syndrome are additional difficulties. In our experience, the use of a NPA during both induction and recovery of anaesthesia may be particularly useful to prevent upper airway problems in children with similar functional problems of the upper airway.

References

- 1 Marshall RE, Graham CB, Scott CR, Smith DW. Syndrome of accelerated skeletal maturation and relative failure to thrive: a newly recognized clinical growth disorder. *J Pediatr* 1971; 78: 95–101.
- 2 Charon A, Gillerot Y, Van Maldergem L, Van Schaftingen MH, de Bont B, Koulischer L. The Marshall-Smith syndrome. *Eur J Pediatr* 1990; 150: 54–5.
- 3 Johnson JP, Carey JC, Glassy FJ, Paglieroni T, Lipson MH. Marshall-Smith syndrome: two case reports and a review of pulmonary manifestations. *Pediatrics* 1983; 71: 219–21.
- 4 LaPenna R, Folger GM Jr. Extreme upper airway obstruction with the Marshall syndrome. *Clin Pediatr* 1982; 21: 507–10.
- 5 Cullen A, Clarke TA, O'Dwyer TP. The Marshall-Smith syndrome: a review of the laryngeal complications. *Eur J Pediatr* 1997; 156: 463–4.
- 6 Pappas CTE, Rekate HL. Cervicomedullary junction decompression in a case of Marshall-Smith syndrome. *J Neurosurg* 1991; 75: 317–9.
- 7 Sperli D, Concolino D, Barbata C, Strisciunglio P, Andria G. Long survival of a patient with Marshall-Smith syndrome without respiratory complications. *J Med Genet* 1993; 30: 877–9.
- 8 Cole TRP, Dennis NR, Hughes HE. Weaver syndrome: seven new cases and a review of the literature. In: Donnai D, Winter RM (Eds.). *Congenital Malformation Syndromes*. London: Chapman & Hall Medical, 1995; 38: 267–80.
- 9 Donnelly J, Thirlwell J. Nasopharyngeal airways in infants after palatal surgery (Letter). *Br J Anaesth* 1992; 68: 227.
- 10 Benjamin B, Walker P. Management of airway obstruction in the Pierre Robin sequence. *Int J Pediatr Otorhinolaryngol* 1991; 22: 29–37.
- 11 Ochiai R, Guthrie RD, Motoyama EK. Effects of varying concentrations of halothane on the activity of the genioglossus, intercostals, and diaphragm in cats: an electromyographic study. *Anesthesiology* 1989; 70: 812–6.
- 12 Steven JM, Cohen DE, Sclabassi RJ. Anesthesia equipment and monitoring. In: Motoyama EK, Davis PJ (Eds.). *Smith's Anesthesia for Infants and Children*, 6th ed. St. Louis: Mosby, 1996: 229–79.