

Russell-Silver Syndrome

Anaesthetic Implications and Management

MD Scarlett¹, MW Tha²

INTRODUCTION

The Russell-Silver Syndrome (RSS) is a form of dwarfism characterized by intrauterine growth retardation with subsequent severe postnatal growth impairment (*ie* low-birth-weight dwarfism). The characteristic features have been classified as universal, common and uncommon (1). Universal features include cranio-facial disproportion, facial dysmorphism, lateral asymmetry and incurved fifth digit (clinodactyly). The typical facial features are frontal bossing, triangular face with hypoplastic mandible and a downward incurving of the mouth, the so-called "shark" mouth (2). Endocrine abnormalities are common traits and include hypoglycaemia, hypopituitarism, adrenal insufficiency and various patterns of hypogonadism (3–6).

Silver and Russell were the first to diagnose this syndrome and independently reported the characteristic features in 1953 and 1954 respectively (7, 8). Hence the synonyms are Russell Syndrome, Silver Syndrome, Silver-Russell Syndrome, Russell-Silver Dwarfism and Silver-Russell Dwarfism.

The aetiology of RSS is unknown. Most cases are thought to be due to genetic mutation because it has appeared sporadically in an otherwise unaffected family. Other postulates are: autosomal dominant inheritance pattern with variable expressivity (9), familial occurrence (10), autosomal recessive inheritance (11), changes in chromosome-17 (12–15), chromosome-7 (16–20) and an X-linked dominant genetic trait (21).

There is no specific treatment for RSS. General treatment includes growth hormone therapy, periactin (to increase appetite), high calorie diet, gastrostomy feeding, limb lengthening/shoe lift, speech and physical therapy. Children with RSS tend to have low self-esteem and emotional problems related to their appearance and difficulty in speaking and walking. Many show improvement in growth and appearance as they get older. There is a RSS Support Group in the United States of America, which monitors all reported cases and publishes newsletters. In 1994, Miles *et al* reported the anaesthetic management of a patient with RSS who presented for surgery (3).

CASE REPORT

A 12-year-old boy of height 130 cm and weight 26.9 kg was scheduled for elective right orchidopexy for an undescended right testis and herniotomy for right inguinal hernia. Russell-Silver syndrome was diagnosed in early infancy based on the patient's low birthweight (2.2 Kg), postnatal growth deficiency and associated physical traits. His height and weight were below the third percentile. He also had congenital heart disease (atrial septal defect – ASD), hyperactive airway disease, frequent chest infections and was maintained on salbutamol inhaler. He had frequent hypoglycaemic episodes up to the age of four years, and had one episode of seizure-like activity at age two years, for which continued treatment was not necessary.

Physical examination revealed a very asthenic boy with little muscle development and subcutaneous fat, but the left side was more developed. He had a narrow elongated head, small triangular face, low set posterior-rotated ears, hypoplastic mandible, small mouth with down-turned corners and microdontia (small crowded teeth). A Mallampati score of II was given with regards to the ease of tracheal intubation. He had widely spaced nipples, lumbar lordosis and scoliosis, a larger left thumb with clinodactyly of the fifth digit of both hands. The right lower limb was shorter by 1.5 cm. The genitalia and scrotum were under-developed with the undescended right testis and the right inguinal hernia. Cardiovascular system examination findings were in keeping with the ASD, which was confirmed by echocardiography and cardiac catheterization. The chest X-ray showed enlarged main pulmonary trunk, but normal pulmonary vessels, lungs and cardiac shadow. The basic haematological and chemical blood tests results were all within normal limits and hypoglycaemia was ruled out.

The patient was fasted for six hours prior to surgery and intravenous 5% dextrose in 0.45% saline was administered. No premedication was given but prophylactic antibiotics (intravenous amoxicillin 1.5 g and gentamycin 40 mg) against infective endocarditis were administered one hour prior to surgery.

The anaesthetic plan was to avoid tracheal intubation since the surgical procedure was expected to be of short duration and profound muscle relaxation was not needed. Preparations were made for a possible difficulty, if tracheal intubation became necessary. After the application of all monitors and pre-oxygenation, anaesthesia was induced with intravenous propofol (60 mg) and maintained with 2.0L O₂/2.0L N₂O/1– 2.5% halothane mixture. Spontaneous respiration was maintained *via* "bag-and-mask" technique and patency of the airway maintained with a size 2 oro-pharyn-

From: Department of Surgery, Radiology, Anaesthesia and Intensive Care,¹ The University of the West Indies, Kingston 7 and Department of Anaesthesia,² Savanna-la-Mar Hospital, Westmoreland, Jamaica, West Indies.

Correspondence: Dr MD Scarlett, Department of Surgery, Radiology, Anaesthesia and Intensive Care, The University of the West Indies, Kingston 7, Jamaica, West Indies. Fax: 977-6160, e-mail: mdscarlett@yahoo.com

geal airway. The size 3 Rendell-Baker-Soucek paediatric facemask was used since the appropriate size laryngeal mask airway (LMA) was not available. Analgesia was provided with intravenous pethidine 5 mg boluses (given twice), paracetamol 500 mg per rectum and infiltration of the wounds with 3 mls of 0.25% bupivacaine solution. Warm drapes and intravenous dextrose solution were used to maintain normal temperature and blood glucose level (7.5 mmol/L) during surgery. Recovery was uneventful and he was discharged from hospital the following day.

DISCUSSION

The pre-anaesthetic preparation for these patients include a thorough "work-up" as they may present with various abnormalities including congenital heart disease (22), urogenital (23, 24), orthopaedic (25), ocular (26), dental anomalies (2) and various endocrinopathies (6). Appropriate measures must be taken to deal with any condition that could impact negatively in the peri-operative period. These include: a possible difficult airway (maintaining patency and tracheal intubation), congenital heart disease and the various endocrinopathies. Assessment of the patient's airway is crucial in order to determine the ease or difficulty of airway management including tracheal intubation and appropriate preparations must be made. The facial, mandibular and buccal manifestations caused anaesthetic concerns because of the possibility of difficulty with airway management.

Preoperative administration of an antisialagogue agent (eg glycopyrronium, atropine or hyoscine) will decrease secretions in the airways and aid visibility (27). This is beneficial in patients for whom multiple attempts at laryngoscopy and tracheal intubation are anticipated or fiber-optic assisted tracheal intubation is planned. The decision to administer a sedative agent must be determined individually and avoided if the airway will be compromised.

Minimum apparatus that should be available for difficult airway management includes several sizes of face masks, oro-pharyngeal airways, laryngoscope blades, tracheal tubes (smaller than the expected diameter for age), a gum elastic bougie, laryngeal mask airways (LMA) and fiber-optic equipment. Experienced anaesthetist/s should also be involved in the management of such patients (27).

Inhalational induction of anaesthesia using sevoflurane is ideal since it does not cause airway irritation and has a quick onset of action (28). If intravenous induction of anaesthesia is chosen, the induction agent must be carefully titrated and long acting muscle relaxants avoided for tracheal intubation since they may have a prolonged duration of action in these patients. Severe hypoxia could occur if tracheal intubation is unsuccessful and it becomes difficult to maintain the airway *via* "bag-and-mask." The facial abnormalities in the patient presented, made it somewhat difficult to get a good seal even with the appropriate size facemask. The calculated dose of any neuromuscular blocking agent will appear small in these patients because of their small muscle mass, but that

dose may cause profound relaxation. The nerve stimulator should therefore be used to monitor neuromuscular blockade if muscle relaxant is necessary. Recovery from inhalational anaesthetic agent is usually quick, as seen in the patient presented. The "wash-out" of the volatile anaesthetic agents occurs at a rapid rate due to the lack of significant fat for uptake during maintenance anaesthesia, and the subsequent release into the circulation during emergence.

Hypoglycaemia is the most common endocrinopathy reported and occurs in a large percentage of neonates but decreases in frequency by the age of four years. One theory for the cause of hypoglycaemia is that it is due to the rapid depletion of limited hepatic glycogen stores, especially in small-for-gestational-age neonates. The large cranial-to-body mass ratio may also lead to a disproportionately increased utilization of glucose (29). Hypoglycaemia was found to be directly responsible for a variety of symptomatology including seizures (30), tachycardia, excessive sweating (31) and periodic episodes of severe weakness which were relieved by food (32). The possibility of abnormal glucose homeostasis due to the prolonged fasting period and the peri-operative stress was anticipated, and hence assessed for and prevented. Unexplained tachycardia, diaphoresis or excessive somnolence after anaesthetic emergence must prompt measurement of serum glucose.

These patients, especially infants are prone to hypothermia and especially in the cold operating room. Normal infants are already more susceptible to hypothermia as compared to adults due to their large surface-to-weight ratio, a smaller insulating mass and a greater tendency toward radiative thermal loss, most notably *via* the head. This is made worse by the abnormally increased cranial-to-body mass ratio and the lack of muscle mass and subcutaneous fat in patients with RSS. Care must therefore be taken to ensure a thermally controlled environment and to prevent excessive heat loss. These can be achieved by warming the operating room, respiratory gases, drapes, intravenous fluids and irrigating solutions, wrapping and insulative guarding of the head and extremities of the patient. These and other measures taken, ensured an uneventful peri-operative experience in this patient.

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