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Rothmund–Thomson syndrome: anaesthesia considerations

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Rothmund–Thomson syndrome (RTS) or poikiloderma congenitale is a rare autosomal recessive disorder. Approximately 300 cases of this syndrome have been reported in the scientific literature worldwide. This study reports the case of an 11-year-old female child with RTS undergoing diagnostic oesophago-gastro-duodeno (OGD) scopy as a result of dysphagia to solids. Adequate knowledge of the condition is needed when planning anaesthesia in such a case as associated anomalies can interfere with anaesthesia management.

Keywords: anaesthesia management, Rothmund–Thomson syndrome

Introduction

Rothmund–Thomson syndrome (RTS) or poikiloderma congenitale is attributed to mutations of the *RECQL4* helicase gene on 8q24.¹ Approximately 300 cases of this syndrome have been reported in scientific literature worldwide.² Characteristic features include photosensitivity, poikilodermatous skin changes, juvenile cataracts, skeletal dysplasias, and haematological, gastrointestinal and genitourinary abnormalities (Figure 1). Patients also have increased likelihood of osteosarcoma and skin cancer.³

We successfully managed a known case of RTS undergoing oesophago-gastro-duodeno (OGD) scopy under general anaesthesia.

Case study

An 11-year-old child weighing 25 kg was admitted for OGD scopy. The patient was a known case of RTS diagnosed at one year of age. The patient gave a history of dysphagia to solids for the past two months. A history of prolonged bleeding from hypertrophic gums was present. A haematology opinion was sought to rule out any bleeding disorders. Her vitals were pulse rate 110/minute and blood pressure 100/60 mmHg. On physical examination hypopigmented and hyperpigmented skin patches were present. Examination of the oral cavity revealed gum hypertrophy and hypodontia. Airway examination did not reveal any other abnormality. The patient had short stature, measuring 115 cm, which is less than the second centile for her age. Eye examination showed the presence of telangiectasia. Systemic examination was unremarkable. Laboratory investigations including a haemogram, complete blood count, renal function tests and coagulation profile were within normal limits. Barium swallow revealed an upper oesophageal web with achalasia cardia. The patient was listed for OGD scopy.

Standard monitoring included electrocardiography, pulse oximetry, and non-invasive blood pressure and temperature monitoring. A 24G intravenous cannula was inserted and secured using non-adhesive dressing to prevent damage to the fragile skin. Injection of midazolam 0.05 mg/kg and injection of fentanyl 2 ug/kg were given for sedation. Diagnostic OGD scopy was done under total intravenous anaesthesia, using injection of propofol and ketamine and maintaining spontaneous ventilation. This revealed a structure in the upper oesophagus, for which

oesophageal dilatation was planned during the same anaesthesia. Subsequently the child was intubated with a 5.5 mm cuffed endotracheal tube (ETT) using injection of atracurium 0.5 mg/kg to prevent aspiration of blood. Intraoperatively 15 ml/kg Ringer's lactate was given to maintain hydration. Injection of ondansetron 0.1 mg/kg was given to prevent postoperative nausea and vomiting. The procedure lasted for 30 minutes. Subsequently the patient was reversed and extubated uneventfully.

Discussion

Rothmund–Thomson syndrome is a rare entity with wide variability in clinical expression. It was first described as an autosomal recessive skin condition by August von Rothmund in 1868. Matthew Sydney Thomson further described it in 1936. The syndrome is named after both of them. The primary defect is in the *RECQL4* helicase gene. The disease can involve multiple systems in the body.

Various manifestations reported include:^{4,5}

- Head, ENT, eye: microcephaly, juvenile cataracts, corneal dystrophy, saddle nose;
- Dental abnormalities: microdontia, hypodontia, ectopic eruption, dental caries and prognathism;
- Neuromuscular: may have mental retardation;
- Orthopaedic: Proportionately short stature, may have small hands and feet, hypoplastic to absent thumbs, syndactyly, club foot;
- Genitourinary/gastrointestinal: hypogonadism, cryptorchidism. Anteriorly placed anus or annular pancreas may be seen;
- Others: skin changes including irregular erythema progressing to poikiloderma mainly in sun-exposed areas. Anhydrosis, hyperkeratotic lesions, dysplastic nails, anaemia.

Intravenous access may be difficult due to poikilodermatous skin changes. The airway may be difficult due to characteristic facies with frontal bossing, saddle nose and micrognathia. Airway manipulation can lead to intra-oral bleeding due to hypodontia and gum hypertrophy. Utmost care should be taken during placement of the laryngeal mask airway or intubation with ETT as the teeth may be loose. Also ETT tubes of smaller size should



Figure 1: Physical appearance of the patient.

be kept ready due to the structure of the oesophagus; a larger tube may interfere with oesophageal dilatation. In view of possible skin problems, careful padding of pressure points should be done and the BP cuff should be lined with soft cotton.

Infants and young children can have gastrointestinal disturbances including diarrhoea and vomiting, which makes them susceptible to dehydration, electrolyte disturbances and malnutrition.

Serum electrolytes should therefore be monitored before elective surgery. Patients can also develop hypertension and nephropathy as complications. Adequate hydration must be ensured. Patients can develop hyperthermia due to hypohydrosis or anhydrosis.⁶ Thus, temperature monitoring becomes essential. Antibiotic coverage should be a priority as patients may be immunocompromised due to malnutrition. Short stature and bone deformities (abnormally formed, shortened or fused bones) can lead to difficulty in positioning. Anticholinergic drugs should be used cautiously as there may be an underlying glaucoma component. Also use of a drug such as suxamethonium, which raises intraocular pressure, should be weighed against the risk of a difficult airway. These procedures may be done in remote locations where all the necessary equipment to manage a difficult airway and experienced help may not be available. Haematological abnormalities ranging from anaemia and neutropenia to myelodysplasia may be present.⁶ Patients may require repeated anaesthesia exposure for multiple corrective surgeries, which can contribute to significant preoperative anxiety. A combination of pharmacological and non-pharmacological measures should be used for anxiolysis.

Conclusion

RTS is a rare syndrome with multiple ramifications due to multi-system involvement. Knowledge of the syndrome and its associated anomalies will help the anaesthetist to anticipate and plan the patient's management for a better perioperative outcome.

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