Anaesthetic management of Mowat-Wilson syndrome

Sir,

First described by D Mowat and M Wilson in 1998, Mowat–Wilson syndrome (MWS) is a very rare genetic condition having multiple facial, gastrointestinal and neurologic features.[1,2]

Our patient, an 18-month-old female child, weighing 6 kg was admitted for repeated episodes of cough, cold, fever, breathlessness with global developmental delay since birth. She was born of non-consanguineous marriage and full-term caesarean section for breech and polyhydramnios.

She was drowsy, hypotonic, hyporeflexic with no social smile, finger grasp or speech. Air entry was decreased on the left with bilateral crepitations. Following facial features were noted: Microcephaly, plagiocephaly, prominent forehead, light coloured sparse hair, medial flaring of eyebrows, prominent eyelashes, depressed nasal bridge, pear shaped nasal tip, inverted V-shaped thick upper lip, everted lower lip, open mouth, pointed chin, malformed and low set ears. She also had mildly everted nipples and proximally placed great toes, clinodactyly 4th toe and camptodactyly.

Haemoglobin concentration was 9 g %. Chest X-ray revealed hyperinflation and bilateral haziness. Arterial blood gas analysis showed hypoxia and hypoxaemia. Magnetic resonance imaging of the brain showed microcephaly and agenesis of corpus callosum. There were increased protein and decreased sugar levels in cerebrospinal fluid. Auditory evoked potential study showed moderate hearing loss in the right ear. Cytogenetic study showed normal female karyotype. Computed tomography of thorax showed patchy areas of consolidation, in multiple segments (suggestive of aspiration pneumonitis) and rounded well-defined density of 2.8 cm × 2.6 cm involving left para-oesophageal area. After above physical features and investigations, diagnosis of MWS was made. Possibilities of bronchogenic cyst or oesophageal duplication cyst were noted which was planned to be removed surgically. Subsequently, anaesthesia team was involved.

After pre-operative stabilisation and written informed consent, general anaesthesia was planned. Intra-operative monitoring included electrocardiogram, pulse oximetry, temperature, end-tidal CO_2 and neuromuscular monitoring. Premedication included injection glycopyrrolate 30 μg and injection hydrocortisone 15 mg. After careful positioning of patient's head and pre-oxygenation with 100% O_2 for 5 min; sevoflurane was started and increased up to 6%. With altered facial features, ventilation with bag and mask, laryngoscopy and the visualisation of vocal cords proved difficult. She was intubated with difficulty using a Portex™ endotracheal tube uncuffed number 3.5 using a small sized stylet. Intravenous fentanyl 6 μg was used for analgesia. Sevoflurane 2% was used as maintenance agent with

oxygen and nitrous oxide in remaining gas (50:50) and atracurium. In supine position, the oesophageal duplication cyst was located, ligated and excised. Blood loss was 20 cc. Reversal of anaesthesia and extubation was uneventful. Paracetamol suppository 180 mg was placed.

MWS is a very rare wide spectrum of clinically heterogeneous features suggestive of neurocristopathies at cephalic, cardiac and vagal levels.[1] This leads to a variety of facial, intestinal and neurological features that are not common in all cases.[2] With only about 200 cases reported, prevalence is about one per 70,000 live births.[3] The inheritance is autosomal dominant.[3] The mutation in the ZEB2 gene on chromosome 2q22 leads to the production of an altered transcription factor resulting in abnormal DNA and protein synthesis at multiple organ system level.[3,4] Hirschsprung's disease is a feature in two-thirds of cases; its absence may still support diagnosis based on other features.[5] Central nervous system (CNS) features include microcephaly and agenesis of corpus callosum.[6] Management is supportive, symptomatic and prognosis is generally poor.[5] In our patient, we tried to prevent repeated lower respiratory tract infection (LRTI) by taking the risk of surgery to remove the possible source; which was possibly aspiration from oesophageal duplication cyst.

A multidisciplinary approach including paediatric surgeon, paediatrician, genetic consultant and paediatric anaesthesiologist was developed. Pre-operative stabilisation included blood transfusion for anaemia and antibiotics for LRTI. Nebulisation and supplemental oxygen corrected hypoxia. Recurrent aspiration pneumonitis was prevented by treatment with antacids and insertion of Ryle's tube. For possible malignant hyperthermia risk, cold ice packs, anti-arrhythmics were kept ready. Omission of succinylcholine, adequate monitoring of electrocardiogram, temperature and neuromuscular status was done for the same. Difficult intubation cart was kept ready. Pre-operative sedation was avoided considering the risk of respiratory and CNS depression. Other rationale behind the omission of succinylcholine was the presence of hypotonia, dystonia, risk of long QT syndrome, hyperkalemia and malignant hyperthermia.[7] Use of non-depolarising muscle relaxant was also avoided initially in view of difficult intubation. Due to its rapid induction and recovery, safety in the presence of agenesis of corpus callosum, and less incidence of malignant hyperthermia, sevoflurane was used.[8]

Thus, the main anaesthetic challenges in MWS are a risk of malignant hyperthermia, difficult intubation, and possibility of difficulty in weaning, LRTI and anaemia. Thorough knowledge of this very rare syndrome and a multidisciplinary approach is necessary for successful outcome. The patient's parents did not consent of publication of images but consented for publication of the case.

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Conflicts of interest

There are no conflicts of interest.

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