Anaesthetic considerations in Aarskog Scott Syndrome: A syndrome new to our understanding

ABSTRACT
Aarskog Scott syndrome is a rare genetic disorder characterised by facial, limb and genital abnormalities first described in 1970. Its evolving nature in terms of associated features and increased surgical interventions necessitates anaesthesiologists to have a thorough knowledge about this syndrome for a better preparedness. Although multiple case reports have been published in literature since its discovery, no case report regarding its anaesthetic considerations and challenges have been described in literature till now. We report challenges encountered and successful anaesthetic management of a seven-year-old girl with Aarskog Scott Syndrome posted for a corneal repair in view of traumatic corneal perforation.

Key words: Anesthesia; Aarskog syndrome; diaphragmatic eventration; female; Hernia; Hiatal; Scott syndrome

Introduction
Aarskog Scott syndrome is a rare genetic disorder first described by Dagfinn Aarskog (1970) characterised by unusual facies, short stature, limb and genital abnormalities.[1] Later, Charles Scott (1971) reported presence of similar findings along with ligament laxity, hyperextensible joints and flat feet, thus acquiring the name Aarskog Scott syndrome.[2]

Due to recent recognition, rarity and limited knowledge about this syndrome, very minimal has been described in literature about its anaesthetic concerns. To best of our knowledge, no article explaining its anaesthetic challenges and concerns have been described in the literature till now. Therefore, we elucidate challenges encountered and successful outcome of a 7-year-old girl with known case of AAS who presented to us for an emergency corneal perforation repair.

Case Report
A 7-year-old girl (15 kg), known case of AAS (FDG1 mutation as per documents) presented in emergency with traumatic corneal perforation and subsequently was posted for corneal repair surgery [Figure 1]. On examination, she had short stature (100 cm), clinodactyly, hypertelorism, high arched palate, macroglossia, irregular dentition, receding mandible, mild mental retardation and a 7 mm atrial septal defect (2D echo) [Figure 2]. There were no other fresh complaints and present biochemical investigations were within normal limit. She had taken her last meal 3 hours back, so we waited for the next 5 hours for her fasting status to complete. In past history, she had episodes of repeated pneumonias in her early childhood with repair of right-sided diaphragmatic eventration at 2 years of age. At present, she had no complaint of respiratory tract infections or signs of...
respiratory compromise and had good functional capacity as per parents (METS >4).

Surgery was planned under general anaesthesia with controlled ventilation after completing an adequate NPO status (8 hours) and keeping in mind the coexisting conditions of child. We had kept a difficult intubation cart ready. Induction of anaesthesia was done with inj. Fentanyl 30 mcg and inj. Propofol 30-40 mg intravenously. After ensuring an adequate bag and mask ventilation inj. vecuronium 1.5 mg was administered. We secured the airway with PLMA# 2 (second generation supraglottic device) and ensured an optimal ventilation. However, soon after securing airway food particles were encountered in orogastric tube inserted via gastric drain tube of PLMA. An immediate head low was done and a continuous suction was attached to orogastric tube to decompress gastric contents. Under continuous suction and direct vision using a videolaryngoscope PLMA was removed and trachea was secured with cuffed ETT #5. Patient’s vital remained stable and surgery was proceeded with following which child was extubated successfully. Intraoperative and postoperative period was also uneventful. On regular follow up in our hospital, child was later found to have a hiatus hernia.

Discussion

Aarskog Scott syndrome or faciodigitogenital syndrome is a complex developmental disorder characterised primarily by facial, skeletal and genital abnormalities. Typical clinical features include short stature, hyperflexible joints, maxillary and mandibular hypoplasia, teeth malocclusion, short and broad distal extremities, clinodactyly, shawl scrotum (males), clitoral hypertrophy (females) and delayed puberty. Additional features include round facies, hypertelorism, ptosis, downward slanting palpebral fissures, broad nasal bridge, palate abnormalities, hernias, contractures, kyphoscoliosis, heart defects and various ophthalmological abnormalities like ptosis, large cornea, nystagmus and ambyopia. The causative mutation responsible is FDG1 gene mutation (X-linked inheritance), which regulates cell growth and apoptosis resulting in anomalous embryonic development and abnormal musculoskeletal formation. However, several clinical and genetic heterogeneity have been reported.

As anaesthesiologists, we can encounter such children for sedation or anaesthesia in various surgical or technical procedures. Commonly performed surgeries are orthopaedic surgeries (contracture, scoliosis repair), dental surgeries, inguinal and umbilical hernia repair, cryptorchidism, eye surgeries, cleft palate repair and even cardiothoracic surgeries. Therefore, it is important to be familiar with anaesthetic considerations and unique challenges likely to be encountered in children with AAS.

Presence of abnormal facial features (mandibular hypoplasia, broad nasal bridge, abnormal dentition, macroglossia, high arched palate, cleft palate, short neck) and cervical spine abnormalities (odontoid hypoplasia, cervical vertebral defects, ligamentous laxity) with hyperextensible joints make these patients prone to a difficult airway situation. Therefore, excessive neck extension should be avoided during airway manipulation and a difficult airway cart should always be kept ready. Since the child was adequately fasting and nature of surgery allowed use of a SGD safely, we used PLMA avoiding undue neck manipulation.

Associated cardiac defects reported in these patients are pulmonary stenosis and ventricular septal defect. However,
our patient had an associated ASD, signifying that all patients should undergo a thorough cardiac evaluation prior to surgery. Positioning under anaesthesia is another concern in these patients due to presence of hyperextensible joints and hyperelastic skin making them more prone to joint dislocations and bruises. Presence of large corneas make them more prone to corneal injuries and thus need more careful positioning especially when made prone.

Patients with AAS usually have normal intelligence but mild to moderate mental retardation and mania have been reported previously. Also, these children are more prone to behavioural problems and attention deficit disorders necessitating use of an efficient premedication and effective post-operative pain management. Other CNS abnormalities involve associated seizures, necessitating knowledge of a detailed treatment history. Spine deformities like kyphoscoliosis with associated respiratory and cardiovascular changes should be evaluated prior to surgery. Altered ossification, abnormal bone age and musculoskeletal derangements result is altered respiratory mechanics in AAS. Therefore, chest and spine X-ray with PFTS should be done preoperatively when necessary. Some of the radiological features are presence of intervertebral disk calcification, vertebral anomalies and spinal bifida occulta in these patients. Administration of central neuraxial block (CNB) can be technically difficult and should be avoided or used with extreme caution and under complete supervision.

Our patient had an associated diaphragmatic eventration (successfully repaired) in her early childhood, which may be as result of poor muscle fibre development and insertion. No case with diagrammatic eventration have been reported in literature suggesting that detailed history and examination should be done to rule out other associated musculoskeletal abnormalities like pectus excavatum, genu recurvatum, hiatus hernias and other vertebral anomalies. We had an unanticipated and impending aspiration risk situation in present case despite adequate fasting status, which was explained later by an undiagnosed associated hiatus hernia. Thus, presence of gastroesophageal reflux (associated hiatus hernia or GE incompetence) should be evaluated preoperatively. A rapid sequence induction (RSI) should always be considered whenever an aspiration risk is anticipated. An aspiration prophylaxis is vital as premedication in these children.

Renal function should be evaluated due to associated urogenital anomalies and renal abnormalities (hypoplasia, cysts, etc). Liver function tests and coagulation profile should be done due to associated haemochromatosis, hepatomegaly and liver cirrhosis in this syndrome. Use of pharmacological agents is to be chosen as per associated features and biochemical investigations. However, drugs altering seizure threshold such as ketamine and tramadol should be used with extreme caution.

Therefore to conclude, this is a recent and evolving syndrome in terms of aetiology, prevalence, associated features and related difficulties. Due to more surgical and diagnostic interventions in recent times, these patients have increased in number coming for sedation or anaesthesia. Thus, early recognition of risk factors, thorough knowledge of the syndrome, better understanding of associated concerns and related complications can help anaesthesiologists to prepare themselves better for a smooth conduct of anaesthesia with a safe outcome.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal. The guardian understands that names and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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