Anaesthetic Management of a Child with Progeria

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ABSTRACT

Introduction: Hutchinson-Gilford Progeria Syndrome (HGPS) is an extremely rare genetic disorder (incidence ~1 in 8 million live births) of accelerated aging in children ending in premature death at an average of thirteen years secondary to atherosclerotic heart disease

Case Report: We are reporting a case of progeria for elective circumcision and rectal prolapse repair under general anaesthesia. Children with progeria present many challenges to the anaesthesiologist such as a potential difficult airway, potential for perioperative myocardial ischemia and haemodynamic instability due to coexistent cardiac disease, risk for perioperative hypothermia and hypoglycemia, risk of pressure sores and skin avulsion and significant osteopenia predisposing them to pathological fractures and pressure sores with surgical positioning. Despite the appearance of advanced age, emotionally and developmentally these patients are still children and interaction with them at the appropriate chronologic developmental stage is essential.

Conclusion: This case highlights the implications on anaesthetic management for a patient with progeria

Keywords: anaesthesia, Hutchinson–Gilford Progeria Syndrome

INTRODUCTION

Hutchinson–Gilford progeria syndrome (HGPS) is an extremely rare genetic disorder pre-dominantly affecting male caucasians (incidence ~1 in 8 million live births). The term progeria originated from Greek and means "prematurely old". Hutchinson[1] and Gilford[2] first described the disease in 1886 and 1904 respectively in England.

It is characterized by pre-mature ageing, severe growth failure (average height ~100 cm, average weight ≤12-15 kg), short life span (mean survival age ~13 years) and very early onset atherosclerosis manifesting as cerebrovascular and coronary artery disease.[3] Atherosclerosis of the cerebral vasculature can predispose progeria patients to intra-cranial hematomas even with seemingly minor trauma.[4] The appearance and growth of the child are usually normal during the first year of life and the clinical picture is typical, rarely requiring differential diagnosis with other diseases. Alopecia, exophthalmos, pinched nose, thin lips, craniofacial disproportion, micrognathia, scleroderma-like skin, hypotrichosis, absent eyebrows and eyelashes, nail dystrophy, delayed and abnormal dentition are common findings but mental development is preserved[5]

Although progeria is a rare condition, these patients often require surgery at some time during their short lives. We report here one such child with progeria who underwent circumcision and repair of rectal prolapse under general anesthesia.

CASE REPORT

A 2 year old male child, born of non-consanguinous marriage was diagnosed as case of progeria at 3 months of age. He was brought to hospital by parents with complaints of straining and pain while urination since 2 months and rectal prolapse since a month. He was posted for elective circumcision and rectal prolapse repair. The child had a full term normal vaginal delivery and had cried immediately after birth. He was immunized till date. Pre-operative evaluation revealed a history of slow growth, progressively appearing features of ageing and normal intellectual development. On examination the patient had typical manifestations of progeria; a small thin built (weight ~ 6 kg), pinched nose with small nares, depressed nasal bridge, dry stretched-out and wrinkled skin, prominent scalp veins, alopecia, sunken eyes without eyebrows and eyelashes, few missing teeth, a 2.5 cm inter-incisor gap, restricted neck movements, knee and elbow deformities with limited movements at both joints and normal mentation. There were no other positive findings on general and systemic examination. The presence of a disproportionately large head, micrognathia, and class-3 Mallampati airway score was suggestive of a difficult airway. Before the start of anesthesia, a well-equipped intubation cart and facilities for emergency tracheostomy were kept ready in anticipation of a difficult tracheal intubation. Intubation via fibreoptic bronchoscopy was not planned as pediatric fibreoptic bronchoscope was not available. Table 1 shows the laboratory investigations. ECG showed sinus tachycardia and right ventricular hypertrophy (normal for the age group).

Hb	12.3 gm%
PCV	34.8%
Platelet count	310,000/cmm
WBC count	6380/cmm
Serum Creatinine	0.3 mg/dl

Table-1: Laboratory investigations

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System involved	Abnormalities
1] Craniofacial	craniofacial disproportion, prominent eyes, beak-like nose ('plucked bird appearance'), protruding ears with no earlobes, thin lips, delayed closure of fontanelles and sutures, stiff neck, decreased mobility of temporo-mandibular joint, micrognathia, hypoplastic mandible, high arched palate, narrow glottis, abnormal dentition, alopecia, prominent scalp veins
2] Cardiac and cerebro- vascular	atherosclerosis, hypertension, carotid and cerebral aneurysms, coronary artery disease, heart failure
3] Skin and skeletal	decreased sub-cutaneous fat, inelastic and wrinkled skin, skeletal dysplasia and hypoplasia, osteoarthritis, wide shuffling gait ('horse riding' stance)
4] Multi-systemic involvement	Diabetes mellitus, restrictive lung disease, vision loss, hearing loss, peptic ulcer disease, absent sexual maturation.

Table-2:

Chest X ray showed deviation of trachea to the right side. 2D Echocardiography was normal. He was taking regular tablet aspirin12 mg once a day which was stopped 7 days prior to date of surgery.

After checking consent and adequate fasting, the child was premedicated with oral midazolam 0.5mg/kg and atropine 0.02mg/kg one hour prior to the surgery. Routine monitoring done with pulse oximetry, non-invasive blood pressure, ecg, end tidal carbon dioxide, oesophageal temperature probe and precordial sthethoscope. The patient was preoxygenated with 100% oxygen with 2% sevoflurane for 3 minutes. Intravenous access established with 22 gauge iv cannula. General anesthesia was induced with intravenous Fentanyl 2mcg/kg, Propofol 2mg/kg and Atracurium 0.5mg/kg. Direct laryngoscopy showed a Cormack Lehane grade 4 view. With hooking and BURP maneuver (backward upward and rightward pressure on larynx) the view improved to Cormack Lehane grade 1. Endotracheal intubation was done using oral uncuffed endotracheal tube with ID 4mm. Ringers lactate with 2.5% dextrose was used as the maintenance fluid at 100ml/hr for first hour and 60ml/hr for second hour. Total duration of the surgery was 90 minutes. The haemodynamics were well maintained throughout the procedure. Intravenous Ondansetron 0.1mg/kg was given 20-30 minutes before extubation. The patient was reversed with Neostigmine 0.05mg/kg and Glycopyrrolate 8ug/kg and subsequently extubated. He was monitored in the recovery room for 30 min and then shifted to the ward. Intraoperative blood loss was 20 ml.

DISCUSSION

The origin of progeria is linked to a genetic alteration in the LMNA gene (producer of the lamin A protein) located on chromosome 1, responsible for maintaining the architecture of the cell nucleus. It is suggested that the absence of that protein makes the nucleus of the cells unstable and leads to consequent premature aging.[6] The most common mutation in patients with progeria is located at codon 608 (G608G). Recently, several studies have been conducted using inhibitors of the genetic alteration for the purpose of establishing a cure and providing better quality of life for affected patients.[7]

Children with progeria are normal at birth and characteristic craniofacial, skeletal and skin abnormalities, and systemic diseases start manifesting by 1-2 years of age. Clinical features and physiological changes in patients with progeria[8] are summarised below in table 2:

Stroke, myocardial infarction and congestive cardiac failure are major causes

of death in these patients.[3] Difficulty in securing the airway, as faced by us in this patient, is a commonly reported complication in patients with progeria, attributed to the presence of multiple craniofacial abnormalities like, micro-gnathia, mandibular and maxillary hypoplasia, large head, short stiff neck, and poor dentition.[3,9] Anticipation of this problem and adequate intra-operative preparedness is necessary to avoid failed intubation catastrophes. Preservation of spontaneous breathing until control of airway is established and the use of special intubation aids like laryngeal mask airways, fibreoptic bronchoscopes, bougies, etc., and methods like awake intubation, blind intubation, emergency tracheostomy, etc., are some of the recommended management methods.[3,9,10] Difficult patient-positioning and increased susceptibility to pressure injuries and skin avulsions due to skeletal and skin abnormalities, potential for peri-operative myocardial ischemia or hemodynamic instability due to co-existent cardiac disease, and requirement for modified anesthesia protocols in keeping with the physiological changes and multi-systemic derangements of old age are some of the other perioperative concerns in progeria patients.[3]

CONCLUSION

The anaesthetic management for patients with progeria needs modified anesthesia protocols keeping in mind the physiological changes and multi-systemic derangements of old age.

These include factors like difficult airway due to craniofacial abnormalities, difficulties in patient positioning, risk of pressure sores, pathological fractures, skin avulsion, potential for perioperative myocardial ischemia and haemodynamic instability due to coexistent cardiac disease. Despite the appearance of advanced age, emotionally and developmentally these patients are still children and interaction with them at the appropriate chronologic developmental stage is essential

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