

# Palatoplasty in a patient with Seckel syndrome

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Website:

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DOI:

10.4103/2231-0746.95324

Quick Response Code:



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## ABSTRACT

We report a rare case of a patient of Seckel syndrome having cleft palate born to nonconsanguineous married parents. This 8-year-old male child underwent successful palatoplasty under general anesthesia. Till date there are approximately 60 cases of Seckel syndrome reported in the literature. The syndrome which has autosomal recessive inheritance is characterized by Intra Uterine Growth Retardation, microcephaly, dwarfism, and bird-like face. The associated features of the syndrome and technical details of surgery and anesthesia are discussed.

**Keywords:** Cleft palate, palatoplasty, Seckel syndrome

## INTRODUCTION

Seckel syndrome first described in 1960<sup>[1]</sup> (also known as bird-headed dwarfism, microcephalicprimordial dwarfism, nanocephalic dwarfism, and Seckel type dwarfism) is an autosomal recessive disorder<sup>[2]</sup> found in consanguineous marriages<sup>[3,4]</sup> and is characterized by severe Intra Uterine Growth Retardation (IUGR), postnatal growth retardation, mental retardation, bird-like face, and retrognathia. Its incidence is less than 1 in 10,000 live births with 25% chances of recurrence in subsequent siblings.<sup>[2,5,6]</sup> Nearly 60 cases have been reported till date with only three cases having being administered general anesthesia (GA). We report the first successful palatoplasty done under GA in a child with Seckel syndrome.

## CASE REPORT

An 8-year-old male child was presented to our outpatient department with complaints of a wide cleft palate causing poor feeding, repeated upper respiratory infections, and inability to verbalize. The parents of the child expressed their concerns for an early surgical correction.

On detailed history, the parents revealed that he was conceived by Intra Uterine Insemination (IUI, donor husband) of nonconsanguineous parents. The father has had infertility from

severe oligospermia and the mother had hyperprolactinemia for which they had undergone treatment 1½ years earlier. Mother also gave history of hypertension, diabetes mellitus and severe oligohydramnios, during the gestational period. Antenatal Intra Uterine Growth Retardation (IUGR) has been noticed in all trimesters. Born at 34 weeks of gestation by normal vaginal delivery with birth weight of 930 g, he cried immediately after birth. He was kept on ventilator for almost 1 month for lung hypoplasia. His developmental milestones were slightly delayed. A history of uneventful dental restoration under sedation or GA in another tertiary care hospital was present. General physical and systemic examination showed evidence of characteristic features of Seckel syndrome, i.e., microcephaly, retrognathia, bird-like face [Figure 1], large eyes, small low set ears, dental caries, long neck, clinodactyly, bilateral elbow contracture, left undescended testis, and urinary and fecal incontinence because of small spina bifida, high arch palate with incomplete cleft of palate [Figure 2]. His weight was 9.5 kg and height was 100 cm [Figure 3].

Detailed investigations showed normal hematological profiles with no abnormality on echocardiography. Chest x-ray showed a tubular cardiac shadow with downward slanting rib cage and normal lung fields. His other two female siblings, also conceived by IUI, showed normal development with no evidence of Seckel syndrome. The patient was scheduled for a pushback palatoplasty under GA and hence an informed parental consent was taken



**Figure 1:** Typical features of Seckel syndrome



**Figure 2:** Showing cleft palate



**Figure 3:** Height of child



**Figure 4:** Postoperative picture

both by the anesthesiologists and surgical team.

#### Operative Details

GA was administered using inhalational induction with sevoflurane in titrated doses while preserving spontaneous respiration in view of an anticipated difficult airway. A fully equipped difficult airway cart was kept ready. After achieving IV cannulation using 24G cannula, and ascertaining chest rise with bag mask ventilation, endotracheal intubation was facilitated using a short-acting neuromuscular blocking agent (NMB) with uncuffed RAE tube size 4.5 mm, after initial failed attempt with tube size 6.0 mm due to narrowing of subglottic region. Anesthesia was maintained with oxygen and air along with titrated doses of sevoflurane and intermittent doses of atracurium. Monitoring in the form of ECG, heart rate, NIBP, EtCO<sub>2</sub>, and temperature was instituted.

Pushback palatoplasty was done for incomplete cleft palate [Figure 4]. The mucoperiosteal flaps were thin and hypoplastic. The lateral alveolar grooves were packed with collagen sheet for postoperative hemostasis. The intraoperative period was uneventful. The total operative time was 75 minutes. Postoperatively, patient was kept in intensive care for monitoring of any apneic spells, respiratory failure, or oral bleed.

#### DISCUSSION

Initially known as nanocephalic, this bird-headed dwarfism was described by Seckel as Seckel syndrome in 1960.<sup>[1]</sup> The incidence of this syndrome is less than 1 in 10,000.<sup>[2,5,6]</sup> Krishna *et al.* in 1994 and Shanske *et al.* in 1997 characterized this disorder as autosomal recessive, resulting from consanguineous marriages.<sup>[3,4]</sup> This disorder has a wide range of phenotypic features such as hydrocephalus, intrauterine and postnatal growth retardation, mental retardation, typical beak-like triangular nose, and skeleton and dentition immaturity as described by Majeswski *et al.* and Goecke *et al.*<sup>[5]</sup> A total of 60 cases have been reported since 1960, out of which only 19 have been described of having classical Seckel syndrome by Sugio *et al.*<sup>[7]</sup> in 1993. Most recently, the locus of this syndrome has been mapped to chromosome 3q22.1-q24 by Goodship *et al.*<sup>[8]</sup> The dentition has been described sporadically, focusing on absence of teeth and atrophy or hypoplasia of the enamel by Gellies.<sup>[9]</sup> Kjaer<sup>[10]</sup> discussed that more pronounced orthopedic and dental abnormality is found in female children when compared with male. Till date only two adults<sup>[7]</sup> and one child<sup>[11]</sup> of Seckel syndrome have been administered GA for abdominal, intracranial, and cleft lip surgery, respectively. Although search of the literature did not reveal any such child undergoing cleft palate surgery, cleft lip surgeries have been

reported in the past.<sup>[12]</sup> This patient had quite a few variations from typical, classical Seckel syndrome as he was born to nonconsanguineous married parents after IUI. He had borderline intelligent quotient with nearly normal comprehensive and nonverbal expressive language. He had no strabismus or cataract of eyes and also had normal hematological and nutritional profile<sup>[11]</sup> because of which palatoplasty could be planned. The veins were very fragile causing difficulty in intravenous canula insertion. The hypoplastic palatal tissue was difficult to handle during surgery. The patient had no postoperative complications such as apneic spells or intraoral bleed as reported in literature,<sup>[11]</sup> and the palatal healing was normal on follow-up.

In conclusion, we recommend that such patients should be thoroughly evaluated preoperatively to rule out any cardiac or other systemic problem. Anesthetic problems because of difficult airway, long neck, narrow trachea, and fragile veins should be anticipated,<sup>[12]</sup> and emergency measures should be made ready for any eventuality, as were done in our case by the anesthesia team. The possibility of hematological abnormalities such as anemia, pancytopenia, and leukemia should be preoperatively evaluated<sup>[13]</sup> along with nutritional assessment. Careful handling of hypoplastic tissue of the palate should be done during surgery. The hemostatic technique should be very accurate in these patients. Because these patients may have a normal life span,<sup>[14]</sup> the parents should be motivated for aggressive postoperative speech therapy, so that these patients could have normal speech and better quality of life.

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**Cite this article as:** Garg R, Uppal S, Mittal R, Grewal A, Sood D, Shah S. Palatoplasty in a patient with Seckel syndrome. *Ann Maxillofac Surg* 2012;2:63-5.

**Source of Support:** Nil, **Conflict of Interest:** None declared.

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