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Case Report

Congenital familial subglottic stenosis: A case series and review of literature

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ABSTRACT

Subglottic stenosis is a narrowing of the endolarynx and maybe classified as congenital (primary) or acquired (secondary). Congenital stenosis maybe caused by a small cricoid cartilage, thick submucosa or other laryngeal abnormalities and remains a well-known cause of stridor in infancy. It occurs sporadically and familial occurrence is rare. Our case series identifies three children with congenital subglottic stenosis born to consanguineous parents. Congenital subglottic stenosis in siblings of unrelated parents has been previously reported, but not in consanguineous parents indicating a strong genetic link. We recommend further genetic research to assess the mode of possible heritage in this disease.

1. Introduction

Subglottic stenosis is defined as a partial or complete cicatricial narrowing of the endolarynx [1]. This process may be congenital (primary) or acquired (secondary). Acquired subglottic stenosis (ASGS) is usually caused by soft-tissue stenosis due to trauma following prolonged endotracheal intubation. Iatrogenic injuries and external neck trauma account for most of the documented cases of ASGS [1].

Congenital stenosis is primarily a clinical endoscopic diagnosis. It may be caused by either a small cricoid cartilage or a thick submucosa secondary to failure of canalization of the subglottic lumen during the third month of gestation [2]. It may also be associated with other laryngeal anomalies. Congenital subglottic stenosis (CSGS) is a well-known cause of stridor in infancy and remains the third most common congenital laryngeal disorder after laryngomalacia and recurrent laryngeal nerve paralysis [3]. It usually occurs sporadically and familial occurrence is rare.

Currently little evidence exists to support a genetic basis for subglottic stenosis. Postulations have included factors such as individual susceptibility [4], inherent sequelae of prematurity such as an abnormal cartilage growth or development [5] or the priori presence of a congenitally small cricoid cartilage [1,6].

We report the unusual occurrence of congenital subglottic stenosis in three male infants, born to closely related consanguineous parents (Fig. 1). Case 1 was preterm (33 weeks), and had growth hormone deficiency. Cases 2 and 3 both had bilateral inguinal herniae, and congenital malformations; however all the children were male, had low birth weight, poor growth and failure to thrive with evidence of grade II to III congenital subglottic stenosis without prior history of intubation. We feel that this case series implicates a possible genetic aetiology for CSGS in children born to consanguineous parents.

2. Case series

2.1. Case 1

This male child was born at 33 weeks preterm by normal delivery and his birth weight was noted at 2.9 kg. He needed no resuscitation other than nasal continuous positive airway pressure and did not require ventilation or intubation. He was symptom free until the age of two years when the anaesthetist noticed subglottic stenosis during intubation for religious circumcision. The procedure was abandoned and postoperatively he developed harsh inspiratory stridor. He was referred to an otolaryngologist and a diagnosis of grade II congenital subglottic stenosis was made (Fig. 2). Intermittently he was having harsh inspiratory noise and mild recession without tracheal tug. Relevant medical history

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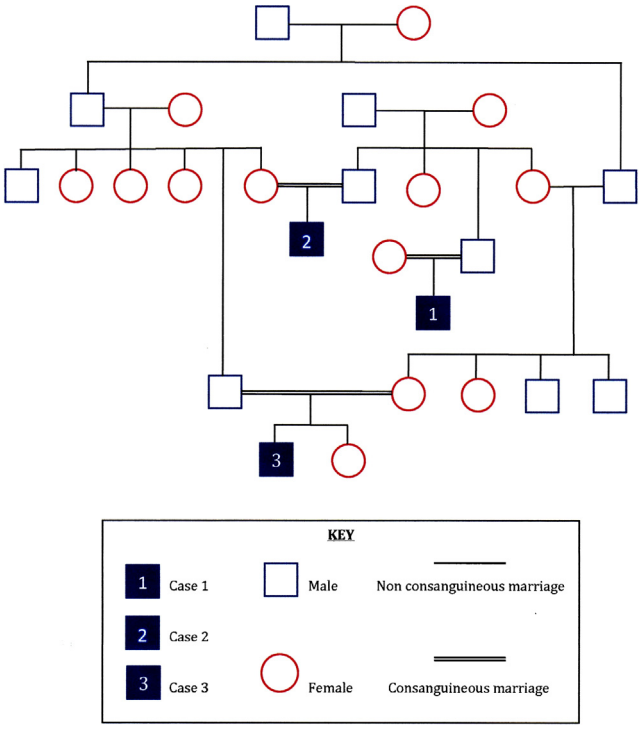

Fig. 1. Family tree with key.

included failure to thrive and rickets. He did not have any dysmorphic features and was developmentally normal. All blood tests including full blood count, U&E, liver function test, ferritin, antibodies and immunoglobulins, amino organic acids, intestinal biopsy, and ultrasound head were normal. He had normal dietary assessment with appropriate intake but with poor weight gain. He continued to be small for age and failure to thrive did not improve. At age of 35 months he was diagnosed with complete growth hormone deficiency and has been prescribed growth hormone injections.

He was managed conservatively until four years of age when he developed noisy breathing during upper respiratory infections. He underwent an adeno-tonsillectomy at 5 years for sleep disorder breathing; however, at 6 years 4 months he was still breathless on exertion and failing to thrive. A single stage anterior graft laryngotracheal reconstruction was carried out at the age of 7 years 4 months (Fig. 3). Post operatively he developed granulation tissue around the graft. This was managed conservatively. The shortness of breath on exertion abated following surgery and he was discharged one year after surgery.

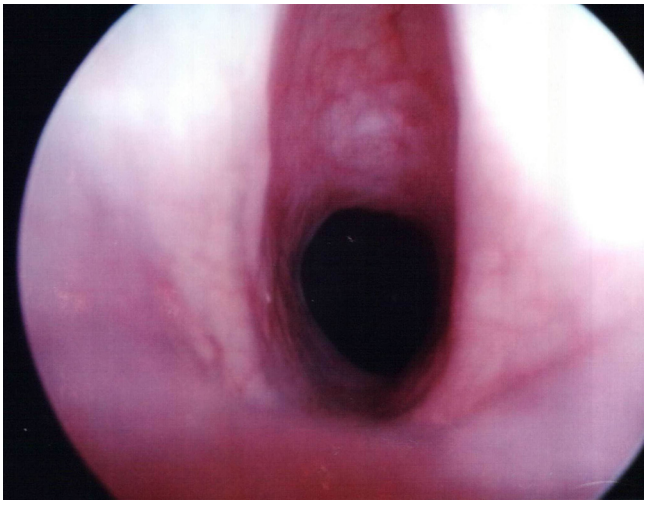

Fig. 2. Case 1: Pre operative grade II congenital subglottic stenosis.

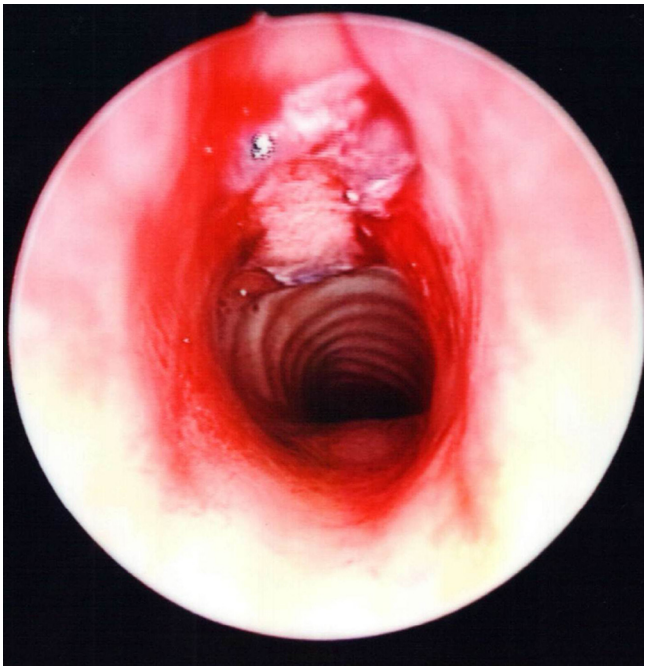

Fig. 3. Case 1: Post laryngotracheal reconstruction with anterior graft.

2.2. Case 2

This male child was born at 38 weeks by normal delivery with a birth weight of 2.58 kg. The antenatal and postnatal history was unremarkable and there was no history of previous intubation or special care unit admission. He had an emergency tracheostomy for failed intubation during congenital inguinal hernia repair and at the age of 5 months he eventually underwent a bilateral inguinal hernia repair and religious circumcision. He suffered with renal impairment and hypertension due to dysplastic kidneys and also suffered with poor weight gain. He had small stature and his growth was along 0.4th centile. He was diagnosed with grade III congenital subglottic stenosis (Fig. 4) and was subsequently managed with repeated balloon dilations. He underwent two stage laryngo-tracheal reconstruction (Fig. 5) with anterior and posterior rib graft at 33 months and is currently symptom free having been recently decannulated.

2.3. Case 3

This boy was born at 38 weeks by Caesarean section for poor fetal growth and maternal diabetes. He had noisy breathing from birth whilst feeding and was admitted at 3 months of age with an upper respiratory infection and worsening biphasic stridor. He was diagnosed with grade III congenital subglottic stenosis (Fig. 6), and subsequently had a tracheostomy. It was also noted that he had bilateral inguinal herniae and also facial dysmorphic features such as a broad based, upturned nose with a short philtrum. He was referred to the genetics department where his karyotype was normal male 46 xy. A Fish 22q11.2 test was normal. He too underwent two stage laryngo-tracheal reconstruction.