Picture Stories

Pierre Robin sequence with congenital talipes equinovarus

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Pierre Robin sequence (PRS) is a heterogeneous birth defect with a prevalence of 1 per 8,500 to 1 per 20,000 live births[1,2]. The association of congenital talipes equinovarus (CTEV) is rare[3].

Case report

A 3 week old male neonate presented with respiratory distress and coarse crepitations in the chest. On examination he had micrognathia, glossoptosis and U shaped cleft palate (uvula was normal) which was suggestive of PRS (Figure 1). There was also CTEV present (Figure 2).

He was kept on nasal continuous positive airway pressure (CPAP) and injectable antibiotics and was nursed in the lateral position or prone position to combat recurrent airway obstruction because of downward displacement of tongue which in turn may be the result of abnormally small sized mandibular bone. Baby was kept on orogastric tube for feeding during the course.

He was born vaginally to a 28 years female and had a birth weight of 2.5 kg. He was dyspnoeic at birth with APGAR scores of 6 and 8 at 1 and 5 minutes respectively. Within 2 days he developed pneumonia which recurred after 3 days of treatment. In the 3 week duration he had a history of 4 such episodes. The neonate was unable to breast feed and so was on spoon feeding since birth. There was no family history of any male affected in previous 3 generations as well as siblings.

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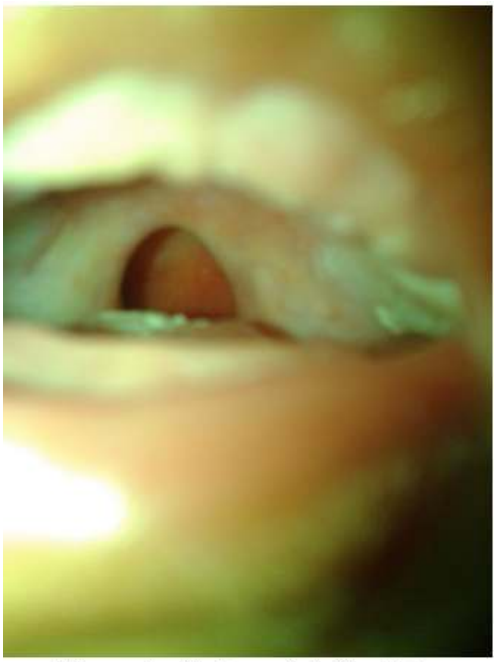

Figure 1: ***U shaped cleft palate***

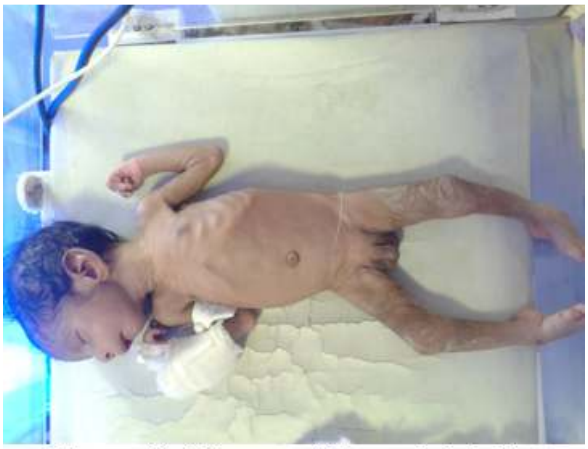

Figure 2: ***Micrognathia and club foot***

**Permission given by parents to publish photograph*

In addition to these findings, the neonate had CTEV having equinus deformity in the ankle joint, varus deformity in the hindfoot, adductus in forefoot and cavus in midfoot. This condition is a rare association

with PRS. To manage this deformity serial cast was applied using the Ponseti method. Testes were in the normal position and the genitals were normal and there were no other musculoskeletal defects. Umbilicus was centrally placed with no deformity. Cardiovascular examination, including echocardiography, was within normal limits and atrial and ventricular septa were intact. Urinary system and gastrointestinal system including liver were normal on ultrasonography.

The neonate was slowly transitioned from nasal CPAP to room air maintaining oxygen saturation within normal range and was nursed in prone or lateral position. While in supine position breathing was maintained through thrusting the jaw and oral airway. Antibiotics were discontinued after completion of the full course and the neonate was discharged after 25 days of treatment with the correction cast for CTEV.

Discussion

PRS is a heterogeneous birth defect with possible autosomal recessive inheritance. An X- linked variant has been reported involving cardiac malformation and clubfeet (TARP syndrome)[3]. Initially mandibular hypoplasia takes place between the 7th and 11th week of gestation. This leads to a tongue high in the oral cavity causing cleft in the palate preventing the closure of palatal shelves. This also explains the classical U shaped cleft palate and absence of associated cleft lip[4]. Oligohydramnios could play a role in the aetiology since lack of amniotic fluid could cause deformation of chin and subsequent impaction of tongue between the palatal shelves as well as CTEV simultaneously.

A clinical array based comparative genome based hybridisation (aCGH) data were used to define 2.08 Mb minimum region of overlap among 4 de novo deletion and one mother to son inherited deletion associated with at least one component of PRS. Deletion of 2.08 Mb on 5q23 region causes a clinically recognisable subtype of PRS. A possible region for CTEV is distinct and telomeric to the PRS region. There may be combined deletion of the region leading to simultaneous presentation[5]. Thus, the typical presentation like in our case goes in favour of gene deletion which may suggest a syndromic presentation of PRS with CTEV.

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