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Challenging case of Pierre Robin Sequence

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CASE TITLE

Challenging case of Pierre Robin Sequence

CLINICAL HISTORY-

A single term male baby was delivered via LSCS in a tertiary care hospital. Maternal age 26 years with obstetrics score G2A1 and blood group O+. Baby conceived from a non consanguineous marriage. Uneventful antenatal history.

Birth weight 2.5kg

Blood group O+

APGAR score 8 & 9 at 1 and 5 minute respectively

Baby was admitted in NICU soon after birth and started feeds, as baby developed Respiratory distress,was referred to our hospital for further management.

On admission, baby was noticed to be dusky with tachypnoea, Retractions, and stridor, initially was nursed in prone position and as the distress worsened, was intubated and put on mechanical ventilator.

CLINICAL EXAMINATION

Heart Rate -138 bpm Respiratory Rate -68/min Spo2- 80% @ RA

Cry –weak Colour- dusky Tone-good

Head to toe Examination:

Baby was alert looking.

Hypoplastic mandible +

Small and posteriorly displaced lower jaw (micrognathia and retrognathia)

Short neck

Posterior position of tongue (glossoptosis)

Abnormal wide opening in the roof of the mouth (cleft palate)

Chest indrawing +

Stridor +

Systemic examination-

Per Abdomen- Soft. No organomegaly.

Cardiovascular System- S1 S2 heard and normal. No murmur.

Central Nervous System – Moderate activity. Newborn reflexes present

Respiratory System- Bilateral air entry present. Tachypnoea present, Stridor present. Pooling of secretions present. Bilateral intercostal and subcostal retractions seen.

INVESTIGATIONS-

Hb- 13.4g/dL

TLC- 12340 cells/cumm

PCV- 42.9%

RBC count- 4.03million/cu.mm

Platelet count- 2.81lakh/cumm

Blood group O+

CRP- 1.48mg/l

PBS Impression – Normocytic Normochromic Anaemia with neutrophilia.

Toxic granules in neutrophils present.

2D ECHO showed small 2.5mm Patent Foramen Ovale. No inotropic support required.

X-ray showed haziness in left middle lobe of lung.

Blood culture: negative

Urine culture: negative

Metabolic/Renal- parameters normal

DIAGNOSIS-

1. Pierre Robin Syndrome

2. Stickler Syndrome

3. Treacher -Collins Syndrome

4. Velo-cardio-facial Syndrome

TREATMENT AND FOLLOW UP

In view of persisting Respiratory distress, desaturation and cyanosis, baby was intubated and put on mechanical ventilation. Gradually was extubated to Nasal CPAP and eventually to oxygen.

Obturator plate was provided for the cleft palate by the dental department and feeds were started. Baby given trial off oxygen on day 21 of life and was nursed in prone position in room temperature. Sepsis was managed with antibiotics.

Temporary tongue tie was doneby the Oro-maxillofacial surgeon to prevent backward fall of tongue and as the distress improved, was nursed in supine position. Mother was encouraged to use a silicon nipple shield for feeding promote sucking and mandibular development.

Gastrostomy was done by Pediatric Surgical team to ensure growth by facilitating adequate nutrition. Dye study confirmed the position of G-tube and full feeds was achieved. The baby was started gaining weight and was discharged home.

After 4 days, baby was brought to ER with respiratory distress due to removal of tongue tie, due to repeated distress, parents were counselled about the risk involved and tracheostomy was done.

Parents were taught about the tracheostomy care and baby was discharged home on full feeds.

DISCUSSION:

Pierre Robin Sequence (PRS) occurs in 1/8500 to 1/14,000 births.

It is described as a condition present at birth, consisting of micrognathia (smaller than normal jaw) and glossoptosis (an abnormal posterior placement of the tongue), and cleft palate (an opening in the roof of mouth) which result in airway obstruction and feeding difficulties. [1][2]

PRS is a sequence rather than a syndrome where multiple anomalies result from a sequential chain of malformation. [3] [4] [5]

The small mandible is due to an inherent genetic problem or a deformational problem where intrauterine growth is restricted or mandibular positioning is altered.

Airway obstruction, is a result of the abnormal positioning of the tongue, which serves to occlude the nasal and oral pharynx on inspiration causing airway obstruction resulting in repeated oxygen desaturation, apnea, and cyanosis.

Infants struggle to breathe during feeds resulting in feeding difficulties. Gastroesophageal reflux and aspiration are common sequelae. The associated cleft palate prevents the formation of negative intraoral pressure, which is required to suck milk from the breast or bottle; the micrognathia and glossoptosis further impede mechanical sucking.

Poor caloric intake associated with reflux, difficulty in feeding and the increased respiratory effort driving increased energy expenditure, these infants often fail to thrive and are unable to gain weight during the early postnatal period.

Genetic basis:

The genetic causes for some of the isolated cases most commonly include mutations or deletions of parts of the DNA neighboring the *SOX9* gene (located in chromosome 17 (17q24)). This gene provides instructions for making a protein (protein SOX9) that plays an important role in the formation of many different tissues and organs during embryonic development. The SOX9 protein regulates the activity of other genes, especially those involved in the development of the skeleton, including the jaw.[2]

In about 37% of cases, Pierre Robin occurs as part of a syndrome with multiple malformations. Pierre Robin sequence has been reported as occurring in association with Stickler

syndrome (20%-25% of these cases), campomelic dysplasia, trisomy 11q syndrome, deletion 4q syndrome, CHARGE association, velo-cardio-facial syndrome, and Treacher-Collins syndrome.

Management of Pierre Robin Syndrome involves multidisciplinary approach. Treatment is focused on the specific needs of each patient, but may include surgery to assist with breathing and feeding modifications to prevent choking.

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