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PIERRIE ROBIN SYNDROME - CASE REVIEW

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Abstract- A case study was carried out of 3 consecutive patients with the Pierre Robin Sequence diagnosed with significant upper airway obstruction at birth with retrognathia, micrognathia, U shaped palate and glossoptosis; followed till three months of their age, treated successfully by conservative prone position management only.

Index Terms- Pierre Robin sequence, upper airway obstruction, prone position treatment.

I. INTRODUCTION

Pierre Robin syndrome is a congenital abnormality characterized by the presence of a combination of mandibular hypoplasia (micrognathia or small jaw), glossoptosis (leading to airway obstruction) and, often, labio palatine clefting.
Over the years there have been several names given to the condition, including Pierre Robin Syndrome, Pierre Robin Triad, and Robin Anomalad. Based on varying features and causes of the condition, either "Robin Sequence" or "Robin Complex" may be an appropriate description for a specific patient. Pierre Robin was a French physician who first reported the of small lower jaw, cleft palate, and tongue displacement in 1923.

II. CASE HISTORY

2 female and 1 male infant were born at term presented with respiratory distress at birth referred to our department from neonatal intensive care unit. On examination patients were having smaller jaw which was slightly posteriorly placed (figure 3).patients were unable to protrude their tongues, with tongue falling back and U-shaped palate (figure 2) .On supine position patients were having noisy breathing with supraclavicular, suprasternal and intercoastal retractions showing laboured breathing with use of all accessory muscles of respiration with mild cyanosis, which was apparent immediately after birth. Oxymetry shows oxygen desaturation into the mid 80s and capillary pco_2 was increased. out of 3 babies two female babies were first child of their parents and one male baby was second but his sibling was not having the history of the same problem.
All three patients were treated successfully by conservative, prone or lateral position treatment with oxygen supplementation by mask. However, one of the three patients required nasopharyngeal tube placement for initial one week (figure 1) later on patient responded well to only prone or lateral position with oxygen supplementation by mask. In all 3 patients feeding was done by nasogastic tubes for initial one month, later on breast feeding was started. The mothers were advised to pump breast milk to aid their infants in breast feeding. Bottle feeding was accomplished by using a long cross cut nipple and with slight pressure at the angle of the mandible with infant in the prone or side lying position .At the end of three months all three patients were normal, with normal weight for age with normal breast feeding with no choking spells and no distress on supine position.
Parents were explained the probable risk of having the same disease in their next babies.

III. DISCUSSION

Pierre Robin Sequence or Complex (pronounced "Roban") is the name given to a birth condition that involves the lower jaw being either small in size (micrognathia) or set back from the upper jaw (retrognathia). As a result, the tongue tends to be displaced back towards the throat, where it can fall back and obstruct the airway (glossoptosis). Most infants, but not all, will also have a cleft palate deformity either involving soft palate i.e U-shaped or involving both soft and hard palate i.e V-shaped (figure 4), but none will have a cleft lip.

PRS is not generally diagnosed with ultrasound before birth because a profile view of the fetus is difficult to achieve.(19) An autosomal recessive disorder, the incidence of PRS is reported to be 1 in 80000 to 1 in 200000 live births.(20)

A greater incidence in girls is attributed to the fact that the palate takes approximately 1 week longer to fuse completely in girls. It is important to understand that Robin Sequence/Complex can occur by itself (described as "isolated") or as a feature of another syndrome. Parents who have had one child with isolated Robin Sequence probably have between a 1-5% chance of having another child with this condition. There have not yet been enough large-scale studies to make more accurate predictions.When Robin Sequence/Complex is observed in patients with Stickler Syndrome, Velocardiofacial Syndrome, or Treacher Collins Syndrome, genetic/chromosomal factors will influence whether more affected children will be born. Robin Sequence/Complex also occurs in children with environmentally-induced ("teratogenic") syndromes such as Fetal Alcohol Syndrome and Fetal Hydantoin Syndrome. It is extremely important that an infant born with Robin Sequence/Complex be evaluated by a geneticist, who will thoroughly investigate the possibility of an associated syndrome.

The pathogenesis of the syndrome is attributed to mechanical compression of the mandible, genetic growth disturbance, teratogen exposure, and growth arrest due to an in utero insult. The basic cause appears to be the failure of the lower jaw to develop normally before birth. At about 7-10 weeks into a pregnancy, the lower jaw grows rapidly, allowing the tongue to descend from between the two halvesof the palate. If, for some reason, the lower jaw does not grow properly, the tongue can prevent the palate from closing, resulting in a cleft palate. It has been postulated that when the mandible is too small it fails to accommodate the descent of the tongue into the mouth; consequently, the tongue remains positioned between the sides of the developing palate, thus preventing complete fusion. . The small or displaced lower jaw also causes the tongue to be positioned at the back of the mouth, possibly causing breathing difficulty at birth. This "sequencing" of events is the reason why the condition has been classified as a deformation sequence. For some patients, these physical characteristics may result from another syndrome or chromosomal condition.

In our case, marked micrognathia is associated with reduction of the buccal space and glossoptosis. As usual in this malformation, the tongue size is normal but buccolingual disproportion is due to micrognathia, increasing the glossoptosis.High thin palate is evidenced by nuclear magnetic resonance. In Pierre Robin sequence, the neurodevelopmental outcome is normal(4).

In several cases in which mandibular hypoplasia is prominent, the tongue tends to encroach on the airway leading to breathing and feeding difficulties, bouts of cyanosis, and chocking spells,as mentioned by Robin(16) to whom the first use of the term "glossoptosis" is attributed. Airway obstruction, and related hypoxia, carries a high mortality risk in Pierre Robin sequence. Children with craniofacial anomalies may present as obstructive sleep apnoea syndrome even in the first weeks of life(9,11,13,14,18).

A history of repetitive broncopneumonia in the first months of life suggests it is related to swallowing impairment. The level of obstruction in the pharynx, appeared as the narrowing of the posterior air space and evidenced in the cephalometrics. This technique provides useful information on bony and soft tissue abnormalities of the upper airways in obstructive sleep apnoea syndrome patients(1,8,10,15,17).Polysomnography determined the obstructive nature of the apneas, quantified its severity and the secondary systemic impairment. This information is fundamental in the evaluation of the Pierre Robin sequence apneas(3,5). Apneas in these patients are predominantly obstructive(3,5,6,11,12).

Patients with Pierre Robin sequence and related craniofacial anomalies who present with respiratory sleep symptoms and daytime sleepiness should be completely worked out including: radiologic,polysomnography and multiple sleep latency test(MSLT) evaluation in order to determine the sites and severity of obstruction,as well as systemic impairment(2,3,6,7). Such evaluation should guide treatment planning**.** Robin Sequence/Complex, like most birth defects, varies in severity from child to child. Some children may have more problems than others.

Problems in breathing and feeding in early infancy are the most common. Parents need to know how to position the infant in order to minimize problems (i.e., not placing the infant on his or her back). For severely affected children, positioning alone may not be sufficient, and the pediatrician may recommend specially-designed devices to protect the airway and facilitate feeding The sequelae associated with the syndrome may range from respiratory difficulties (obstructive sleep apnea), nutritional difficulties(regurgitation, gastroesophageal reflux), speech and hearing problems (otitis media, difficulty in speech), crowding of the dentition, and facial asymmetry.

Feeding difficulties arise because the presence of cleft prevents creation of the negative intraoral pressure necessary to withdraw milk from the breast or the bottle. A small jaw and posterior placement of the tongue further impedes effective mechanical sucking movements and chronic airway obstruction further increases the work of breathing.To counter the feeding difficulties, the mothers may be advised to pump breast milk to aid their infants in breast feeding. Bottle feeding can be accomplished by using a long cross cut nipple and with slight pressure at the angle of the mandible with infant in the prone or side lying position. The compression of the nipple against the bone allows greater expression of milk. Frequent burping is also recommended. Some children who have severe breathing problems may require a surgical procedure to make satisfactory breathing possible.The pediatrician or ear, nose, and throat specialist will also carefully monitor the baby for ear disease. Virtually all children with cleft palate are prone to build-up of fluid behind the eardrum. The placement of ventilation tubes in the eardrums may be recommended to reduce fluid build-up. Since ear infections can cause temporary hearing loss that can affect speech and language development, the infant's hearing should also be monitored from early infancy by an audiologist.

The rate of catch-up growth of the jaw is dependent on the cause of the PRS. A majority of children with isolated PRS achieve normal or near-normal mandibular size within a few years of birth(21).In many patients, the lower jaw (mandible) grows rapidly during the first year of life. In some children, the jaw may grow so quickly that by the time the child is approximately four to six years of age, the profile looks normal. Children who do not experience this "catch-up" growth may require surgery on their jaws. It is not fully understood why children's jaws grow at varying rates.The cleft palate, if present, needs to be surgically closed. The timing of the surgery depends on the child's individual growth and development, but it is generally done at 1 to 2 years of age. Because children with cleft palate are at higher risk for delayed or defective speech development, they should be monitored by a speech pathologist throughout their childhood.

Several methods of treating babies with Pierre Robin sequence have been described since the condition itself was first documented in 1923. The main aim of treatment has been to relieve upper airway obstruction. Treatment methods used range from positioning of the baby to invasive surgery. Treatment concentrates on the relief of airway obstruction with a nasopharyngeal airway (NPA) and nutritional support of the babies until they grew out of their respiratory and feeding difficulty.(22) .

Immediately after delivery, because of the micrognathia and, therefore, relative glossoptosis, many children have airway distress. This can require emergency treatment. As the body will always prioritize breathing over eating, many infants have difficulty in achieving adequate caloric intake. A cleft palate further adds to the feeding difficulties. The primary concern in airway compromise is

its life-threatening aspect. Most neonates have an isolated defect that is not part of a syndrome, for which the airway and feeding complications are usually greater. The great majority of neonates can be treated in the prone position (face down as in our case). Devices or procedures such as oral airways, palatal prostheses, continuous positive airway pressure or endotracheal intubation, mechanical ventilation, and tracheostomy can be avoided.

These neonates also need to be fed in a prone position, but they can be fed by mouth. Again, very few infants need long-term gavage feeding tubes or other devices. The vast majority of nonsyndromic neonates, those who are breathing without assistance and orally feeding while in a prone position, can be discharged home after a few days in the hospital. The mandible is expected to eventually grow; hence, the severe airway obstruction and the feeding issues are expected to decrease. The infant continues to need feeding and speech assessments, and breathing capacity needs to be monitored. Eventually, the cleft palate needs closure, and a long-term orthodontic care is required;

Secondary to the micrognathia/retrognathia, airway obstruction may be mild or severe. Severe obstruction may require immediate intervention with a very difficult intubation. A mild obstruction can normally be handled in a very conservative manner with positional changes. By putting a baby in the prone position, gravity pulls the tongue forward and results in a larger airway passage.

In patients who consistently maintain CO_2 levels above 50, a surgical procedure is appropriate. Three surgical procedures are used to treat Robin sequence: tongue-lip adhesion/glossopexy, tracheostomy, and distraction osteogenesis of the mandible.[23] Distraction osteogenesis is a relatively new technique for treating airway obstruction in Robin sequence. Mandibular distractional osteogenesis offers a definitive structural resolution of micrognathia.

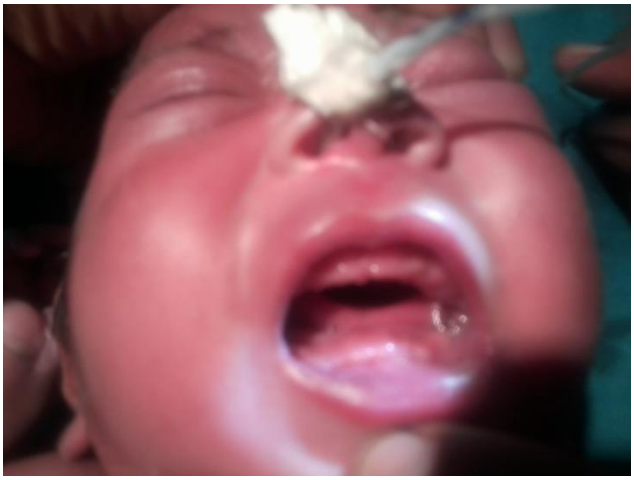

Figure1. Patient with nasopharyngeal airway

Figure2. Showing retrognathia and micrognathia

IV. CONCLUSION

Primary management of airway insufficiency in patients with PRS can be managed in a prone position mostly however some may require nasopharyngeal airway placement, prolonged intubation, tongue-lip adhesion, mandibular distraction osteogenesis and tracheostomy. The present study confirmed that proper conservative management can be used to manage most of the patients with PRS. However, early mandibular distraction should be considered when indicated in patients with respiratory insufficiency to avoid tracheostomies or successfully decannulating tracheostomies.Multidisciplinary care that includes a neonatologist, a neonatal nurse

specialist, members of the otorhinolaryngologist team, and the parents is the best approach in the complex care of neonates affected with Robin sequence and Robin complexes.

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