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BABY WITH LARGE HEAD, SHORT NECK WITH ABNORMAL RIBCAGE……..A CHALLENGE TO NEONATOLOGIST

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ABSTRACT

Hypochondroplasia or achondroplasia is a condition where all the cartilages are small and deformed leading to poor growth of future long and short bones sparing flat bones to some extent. Prominent forehead, midfacial hypoplasia with very short neck and disproportionate head circumference alongwith short tubular bones are main features of these short babies. Unexplained respiratory distress requiring ventilator since few hours of birth with normal lung parenchyma, echocardiography, and cranial USG compels neonatologist to think any ribcage anomaly. Narrow short ribs mimicking thoracic dystrophy contributes to increase work done and difficulty in breathing and feeding. In this case we share our experience and challenges faced by the neonatologist in NICU.

KEYWORDS: Hypochondroplasia, large head, respiratory distress, thoracic dystrophy to NICU.

INTRODUCTION

Achondroplasia is a condition where babies are born with large head, short neck, long trunk, midfacial hypoplasia and others.[1] These defects are due to chondrodysplasia or hypoplasia of cartilage. Narrow short ribs looking like thoracic dystrophy makes a pediatrician worried about first breath and after. Sometimes craniocervical junction stenosis makes respiratory distress prolonged and irreversible.[1,2] We report a case of similar nature.

Case

A large baby Fig:1, is delivered by LUCS with a birth weight of 3.6 kg to a short mother at term, baby developed respiratory distress immediately at birth for which she was admitted to neonatal intensive care unit of our hospital and put on nasal continuous positive airway pressure(CPAP). After six hours due to detoriation of oxygen saturation it was decided to put her on ventilator. But due to short neck Fig:2, two senior residents and one consultant failed to put a endotracheal tube(ET), finally anesthetist was called and ET was done.

After seven days of ventilation with much difficulty she was weaned from ventilator. Even after weaning she developed persistant respiratory distress needs pressure support by CPAP for next seven days. The baby was discharged at day 21 after training the mother on supervised expressed breast feeding plus top milk.

To evaluate the cause of respiratory distress chest x- ray Fig:3, was done which showed no parenchymal lesion but bony ribs are far away from sternum with suspected handle bar clavicle suggestive of thoracic dystrophy.[3] Ultrasonography of brain showed no hydrocephalus or any structural malformation. Routine blood, sepsis screen and echocardiography were within normal limit.

Figure 1.

Figure 2.

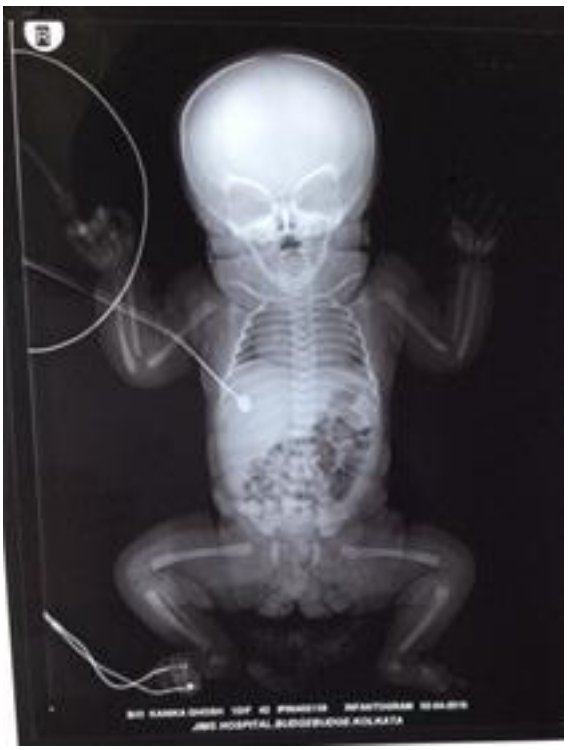

Figure 3.

DISCUSSION

Baby developed respiratory distress right from birth which needs exclusion of respiratory distress syndrome even in term baby or some congenital anomaly of heart, lung, thorax or central nervous system. We excluded all common causes by echocardiography, CXR, USG brain but expert radiologist opined for thoracic dystrophy from chest x-ray as a cause of persistant respiratory distress. For chest expansion we need bucket handle and pump handle movement of ribs, if ribs falls short of sternum due to aplasia or hypoplasia of chondrocytes overall movement of thorax is restricted leading to increase in respiratory rate.

Persistance of respiratory distress even after weaning from ventilator without any infection in a large baby needs exclusion of metabolic or CNS or other causes. Stenosis of craniocervical junction 2 is commonly seen in this kind of clinical phenotype which can contribute to irregular medullary respiration or distress but unfortunately father of the baby refused CT scan brain.

With watchful expectancy and conservative management respiratory distress settles and feeding was established. Initially nasogastric feeding followed by bottle, but direct breast feeding was not possible even if on 6 weeks follow up.

Achondroplasia is transmitted by autosomal dominant inheritance with variable penetrance[4], so all signs and symptoms of classical achondroplasia may not be present. In this case rhizomelic shortening of humerus and femur is not typical on radiology but frontal bossing, increased head circumference relative to long bone measurements, broad pelvis, squre ilia and narrow sacrosciatic notch Fig3, points towards some variety of hypochondrogenesis.[5] Mother of the baby is short with a height of 132cm and father is normal with 157cm.

So, when a large baby with short neck with or without achondroplastic phenotype is born, pediatrician or neonatologist should anticipate difficult intubation, look carefully for thoracic dystrophy and CT scan craniocervical junction brain to rule on stenosis. Lung hypoplasia, due to a restricted thoracic cage, is the major cause of death in infancy.

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