Indian Journal of Medical Case Reports ISSN: 2319–3832(Online)
An Open Access, Online International Journal Available at http://www. cibtech.org/jcr.htm
2015 Vol. 4 (2) April-June, pp.23-25/Yudhishtir et al.
Case Report

A RARE CASE OF ASPHYXIATIING THORACIC DYSTROPHY WITH BLEEDING DISORDER

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ABSTRACT
Jeune syndrome or asphyxiating thoracic dystrophy is rare multisystem autosomal recessive skeletal dysplasia characterized by a narrow thorax, short limbs, and renalhepatic, pancreatic and retinal abnormalities with multiorgan involvement (Jeune *et al.*, 1955). We are reporting a rare case of asphyxiating thoracic dystrophy with bleeding disorder.

Keywords: *Asphyxiatiing Thoracic Dystrophy, Bleeding Disorder*

INTRODUCTION
Asphyxiating thoracic dystrophy (Jeune syndrome) is characterized by a small, narrow chest and variable limb shortness. The diagnosis is based on clinical and radiologic findings.

The small, narrow thorax often results in respiratory distress and recurrent respiratory infections in the neonatal period and infancy.

Often (60–80%), the outcome of ATD is said to be fatal in early childhood (Morgan *et al.*, 2003; O'Connor *et al.*, 2008). Associated congenital abnormalities are postaxial polydactyly of both hands and/or feet.

Occurs in1:100000births. Wedescribe one such case with diagnosis based on clinical and radiographic findings.

Key factors in the prenatal diagnosis are an abnormal small thorax, short limbs, polyhydramnios, and unidentifiable fetal respiratory movements (Chen *et al.*, 1996).

CASES
We had a case in our hospital of a single live male preterm baby born by LSCS to G2L1 mother, with indication of polyhydraminos with flat NST.

At birth child was limp RR: Nil, HR<60, color: cyanosis, baby shifted to NICU and ventilated for the same.

On examination, HR 140/MIN, PP: well felt, CFT <3 sec, NO spontaneous respiratory effort. On head to toe examination, there were hypertelorism, edema of both palms and soles, short stuby fingers and in flexed position, undescended testis, petechial rash were evolving on chest and upper limbs, tight skin. Baby was kept on ventilator support.

X ray showed narrow chest cavity, CT showed: bell shaped thorax, with minimal ascities in abdomen and hypoxic changes in brain. PT APTT was prolonged on two occasions with increase petechial rash.

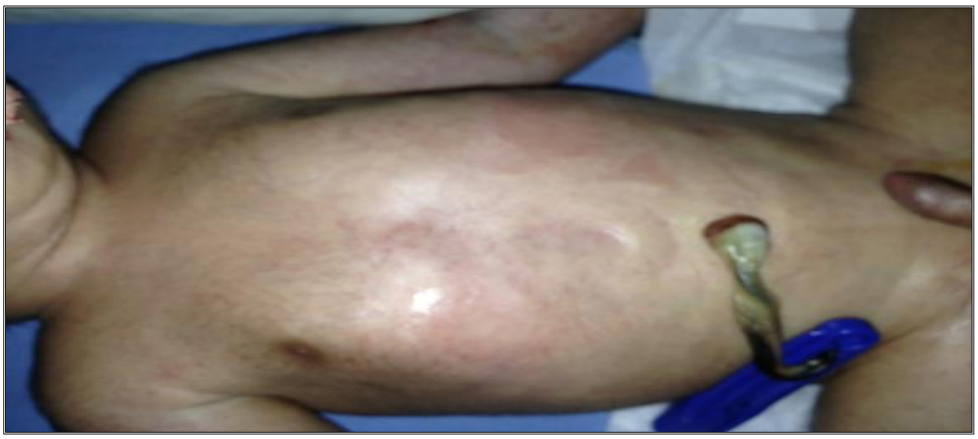
Other investigations were within normal limit. ABG showed sever metabolic acidosis with hypocarbia.

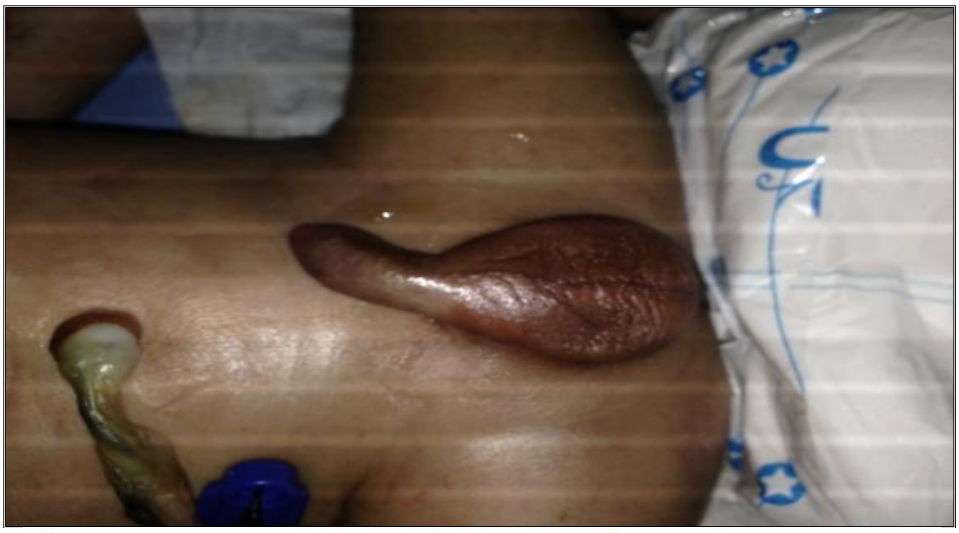
During the course of NICU stay baby developed petechia (bleeding diathesis) suspecting coagulation defects.

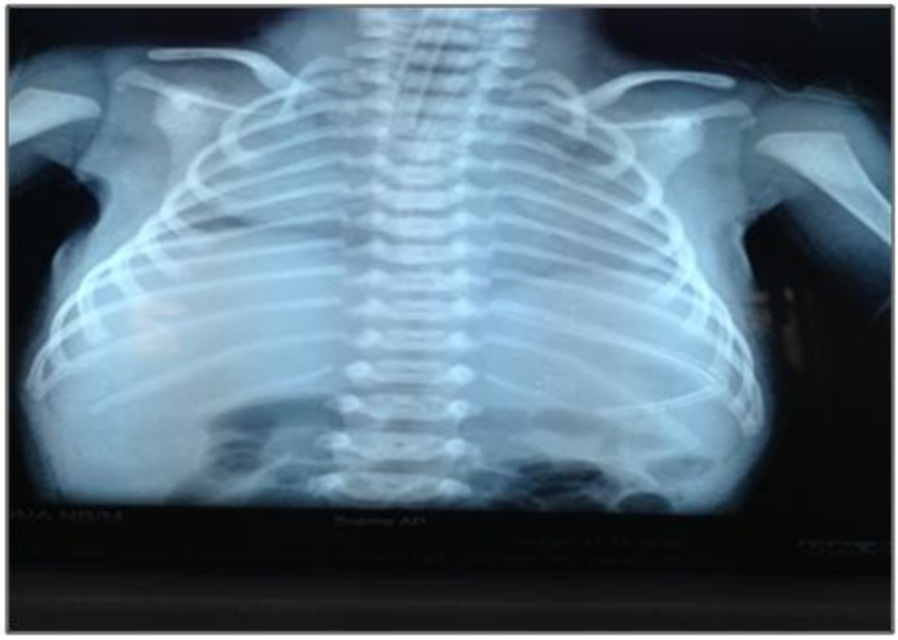
Inotropes, antibiotics were added over the period of stay. Fresh frozen plasma transfusion was advised. In view of financial constraints baby was discharged against medical advice by father.

Case Report

Pictures Showing Tight Skin, with Undescended Testes and Petechia







DISCUSSION

Asphyxiating thoracic dysplasia is also known as jeune syndrome is rare type of skeletal dysplasia, characterized by long narrow thoracic cavity, tight skin and characteristic skeletal features. It is thought to carry an autosomal recessive inheritance.

A locus has been identified on chromosome 15q13, while recently mutations were found in IFT80gene. In small subset of ATD there were no extra skeletal manifestations (IFT80, 2007).

Clinically characterized by narrow chest and variable short limbs, tight skin and polydactyly. Complications including pulmonary hypoplasia, hepatic cirrhosis, and renal failure. Prognosis is variable. Those who survive there can be improvement in thoracic deformity. Most mortality is due to respiratory compromise (Matt and Yuranga *et al.*,).

Jeune syndrome is sometimes compatible with life, although respiratory failure and infections are often fatal during infancy. The severity of thoracic constriction widely varies. For those patients who survive infancy, the thorax tends to revert to normal with improving respiratory function. This suggests that the lungs have a normal growth potential and the respiratory problems are secondary to restricted rib cage deformity. It is important to establish a correct diagnosis both in severe and mild forms since it might recur within family (De Vries *et al.,* 2010).

ATD is a genetically heterogeneous multi-organ disease with variable expression, predominantly affecting the thoracic cage with respiratory complications as the main problem. Children often need respiratory assistance during their first years, but the respiratory problems decrease with age (Beales *et al.,* 2007).

Conclusion

Jeune syndrome is a very rare case, with autosomal recessive trait, it can be diagnosed with clinical and radiological features. Although multi organ involvement is common, but association with bleeding disorders at birth is quite a rare occasion.

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