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RESEARCH ARTICLE

RIGHT SIDED DIAPHRAGMATIC HERNIA WITH HOMOLATERAL LIMB REDUCTION

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ABSTRACT

Amelia is a rare disorder refer to complete absence of skeletal part of arm or leg that occur when limb formation process is either interrupted or prevented during development of embryo. Amelia with diaphragmatic defects in a newborn is rare entity. We report a rare case of newborn with congenital malformation of Amelia associated with congenital diaphragmatic hernia, its manifestation and fetal outcome.

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INTRODUCTION

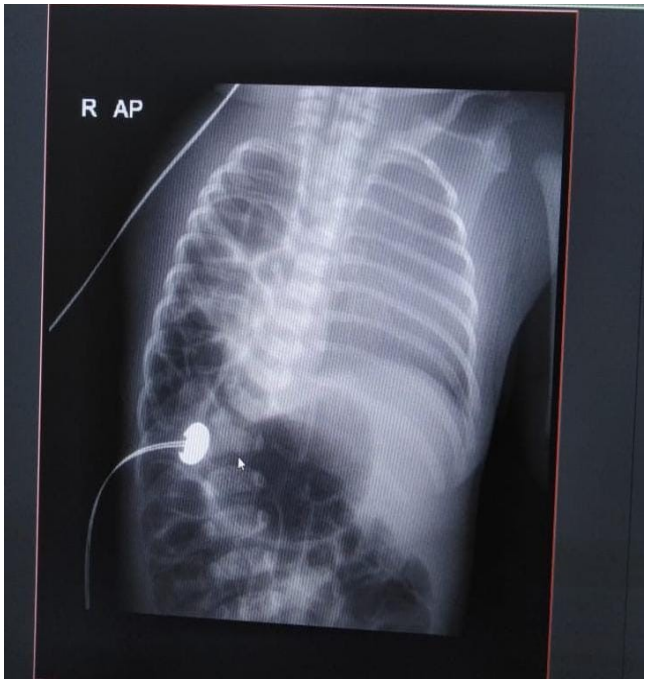
Amelia is defined as a congenital anomaly which is characterized by complete absence of the skeletal parts of a limb[1].It is a very rare condition with an incidence of 0.053 to 0.095 in 10,000 live births.[2-3] Amelia can present as an isolated defect or associated with other malformations, particularly abdominal wall defect, renal anomalies, cleft lip and or cleft palate, heart anomalies, defect in neural tube and diaphragm[4-6]. Etiology and mode of inheritance are still not clear and it is thought to be a sporadic anomaly. Causes such as exposure to thalidomide, alcohol, vascular compromise by amniotic bands and maternal diabetes have been reported in some limb deficiency cases.[7-10] We are reporting a case of female baby born live but unfortunately expired at 48hours of life.

CASE REPORT

A female, full term baby (38+3wk), appropriate for gestational age(2.500kg) born to 23 years old primigravida mother. Baby was delivered by assisted vacuum delivery for fetal distress. Baby did not cry immediately after birth, required positive pressure ventilation for 30secs. There was no improvement in breathing and saturations so baby was intubated.

At birth, the absence of right upper limb was noted. Baby was admitted to NICU for perinatal asphyxia with hypoxia and Amelia for detailed evaluation. Baby was born out of a non-consanguineous marriage. Mother received iron and folic acid supplements during pregnancy and there was no history of teratogenic drug exposure. There were 4 antenatal visits and ultrasound was done at 14, 20 and at 37weeks and no congenital malformation was documented in any scan. Triple marker studies were normal. In NICU, baby received mechanical ventilation requiring 100% oxygen and very high pressures. Pre and post ductal saturation of more than 5% and maximum saturation achieved was 88%. On detailed examination, head circumference and length were 34cm and 48cm respectively. Face was normal. Baby had complete absence of right upper limb and clavicle and scapula could not be felt on right side. Scoliosis of the thoraco-lumbar spine was noted. Heart sound heard on far-left side of the chest and were distal, there was no organomegaly and genitals were normal. Urgent USG thorax to look for pneumothorax revealed liver and bowel in the right side of chest suggesting a right sided congenital diaphragmatic hernia (CDH). X-ray chest and abdomen showed complete absence of upper limb with absence of same sided scapula and clavicle with some gaseous shadows in right thorax with scoliosis of spine in the thoraco-lumbar region. Echocardiography reveled a complex congenital heart anomaly, situs-inversus levocardia with large ventricular septal defect and large atrial septal defect, three pulmonary veins with over-riding aorta, patent ductus arteriosus with

right to left shunting with moderate to severe pulmonary hypertension.



Detailed ultrasonography of chest and abdomen shows bowel loops on right side of thorax and upper pole of right kidney herniated into right side thorax, with some part of liver confirming right sided CDH. Spleen was not visualized and stomach was seen in the middle part of abdomen. Aorta and IVC were in reversed relationship. Neurosonogram showed normal brain parenchyma and ventricular system. Baby continued to have increasing desaturation despite of maximum ventilation settings, trial of high frequency ventilation and sildenafil infusion. Gradually baby's condition got worse and despite of all measures, baby died at 48 hours of life. Autopsy was offered but parents refused and genetic analysis was sent.

DISCUSSION

Congenital diaphragmatic hernia (CDH) is a structural birth defect characterized by protrusion of abdominal viscera into the thorax and it is usually an isolated nonsyndromic anomaly, it may be associated with other malformations or may occur as part of a well-defined syndrome.[11-13]There are four malformations that preferentially in association with congenital diaphragmatic hernia[12] (a) polysplenia syndrome, (b) ipsilateral limb reduction, (c) omphalocele and (d) caudal dysgenesis with cost overtebral anomalies. In our patients there is right congenital diaphragmatic hernia with ipsilateral limb reduction (Amelia). During embryogenesis defect in neural crest development of cervical somites, leads to limb defect. The sensitive period for malformation of the arm (26 to 36 days of gestation) and for diaphragm (28 to 48 days) which both significantly overlap. During this critical period any injury to neural crest in the embryonic cervical region would result in developmental deficiency of the arm and diaphragm.[14] In our case there is complete absent of right upper limb and scapula with ipsilateral congenital diaphragmatic hernia with complex heart disease, though prenatal ultrasound did not reveal any gross anomalies, organ malformation has been reported to be frequently associated with case of amelia. In this case pregnancy and family history were noncontributory factors. Autopsy finding would have been helpful, but unfortunately, pathological examination could not be done.

CONCLUSION

Amelia a very rare disease may present unexpectedly in spite of screening ultrasounds. Genetic tests in index case may help in counselling family about the risk of recurrence.

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