

Anencephaly with Cervical Rachischisis (Craniorachischisis) and Congenital Bronchogenic Cyst : An Autopsy Case Report of a Rare Association

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(Received:22/10/2014, Accepted: 16/11/2014)

Abstract :

Anencephaly is one of the commonest and lethal neural tube defects, which occurs as a result of defective closure of rostral pore of neural tube. Anencephaly can be associated with various other system anomalies. We present here a rare association of a congenital mediastinal bronchogenic cyst with anencephaly with cervical rachischisis (Craniorachischisis) noted in a recurrent gestation of anencephaly.

Key Words : Anencephaly, Craniorachischisis, Congenital bronchogenic cyst, Autopsy

Introduction

The spectrum of neural tube defects (NTDs) comprises of disorders related to the non-closure of the neural tube.¹ Anencephaly is one of the commonest and lethal neural tube defects which occurs as a result of defective closure of rostral pore of neural tube.² Reported prevalence of anencephaly was the highest at 2.1 (1.6-2.8) per 1000 births among NTDs in India.³ We present here a rare association of a congenital mediastinal bronchogenic cyst with anencephaly with cervical rachischisis (Craniorachischisis) noted in a recurrent gestation of anencephaly.

Case Report

A 26 yrs old lady, G₃P₁L₁A₁ at 18 wks gestational age underwent Medical Termination of Pregnancy (MTP)



Figure 1: Abortus



Fig 2: Rachischisis with exposed and flattened spinal cord

after an ultrasonographic diagnosis of anencephalic gestation was made. The first abortus was a medically terminated anencephaly and the next gestation ended in a normal live birth. The lady was a booked case and had regular antenatal check-ups during this gestation and was on iron-folate supplements after confirmation of pregnancy in the later part in the first trimester.

Autopsy was done after an adequate fixation of the internal organs 2 days after injecting 10% formalin to body cavities and cranium. Anthropometric measurements were noted soon after receipt of the body in the department. It was a 280 gram female abortus (Fig. 1) with 16 cm crown-heel length, 18 cm head circumference with the head showing a cystically enlarged swelling of the cranium covered by scalp with complete absence of calvarial bones. The defect extended posteriorly, in continuum to involve cervical vertebrae as rachischisis, exposing the spinal cord which appeared flattened (Fig 2). No retroflexion noted (which is a component of Iniencephaly). Dissection of the cystic swelling of the cranium confirmed the absence of cranial bones. Meninges were not well made out, and there was an angiomatic tissue with a small 1.2 cm chunk of grey-white brain tissue at the base of skull (Fig. 3) with disorganized anatomy on microscopy.

En masse removal of organs was done and the thoracic, abdominal and pelvic organs were evaluated grossly and histologically. A prominent dull-white unilocular smooth lined cyst (Fig. 4) with a clear fluid content, measuring 2x1x1 cm was noted on the right side of middle mediastinum with attachment to the carina, but no communication with the trachea. Microscopically (Fig. 5) the cyst was lined by pseudo-stratified ciliated columnar epithelium. The cyst wall was composed of smooth muscle and fibrous tissue. No fragments of

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Figure 3 : Angiomatous mass with a grey-white agenetic brain tissue at the base of skull



Figure 4 : Congenital bronchogenic cyst with a mass effect on postero-medial aspect of right lung (Shown are hilar surfaces of both the lungs)

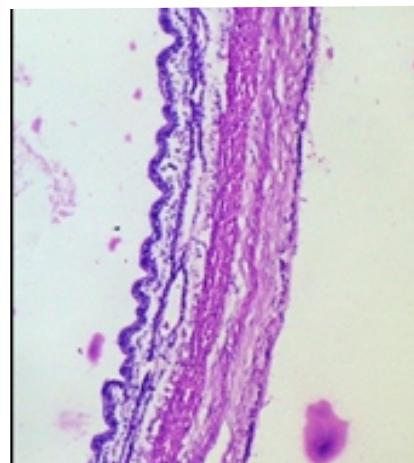


Figure 5 : Microscopy showing cyst lined by pseudostratified ciliated columnar epithelium

cartilage were noted. With these features, a diagnosis of 'Congenital bronchogenic cyst' was made.

The cyst had a mass effect in the mediastinum, compressing the right lung postero-medially. Except for this finding, both lungs were normal, with the right lung having two lobes and the left with three lobes. No abnormalities were noted in the cardiovascular, gastrointestinal and genito-urinary systems. Both adrenals and thymus were normal. No lower vertebral or skeletal abnormalities of extremities were noted.

Discussion

Neural tube defects are one of the most common structural congenital anomalies which include a spectrum of spina bifida, anencephaly (absence of brain calvaria, total or partial), encephalocele (herniation of brain and meninges through defect in calvaria), craniorachischisis (anencephaly associated with continuous bony defect of spine and exposure of neural tissue) and iniencephaly (dysraphism of occipital region accompanied by retroflexion of neck and trunk).⁴ NTDs overall have a female sex bias. Females more often than males tend to have craniorachischisis, spina bifida involving the thorax, the holoacrania form of anencephaly, anencephaly and cervical spina bifida and encephalocoeles, while males more often than females have spina bifida affecting the lower spine.⁵ Our case too is a female with craniorachischisis which obeys this trend.

This was a second conceptus with anencephaly of a consanguineously married couple. Kulkarni ML et al who have studied NTDs in the same geographic setting have concluded that there is a complex interaction of genetic and other factors in the aetiology of neural tube defects in highly inbred population of South India, with the genetic factors probably being the most important.⁶ The recurrence risk after the birth of an infant with anencephaly is noted to be 3-5%.⁷

Maternal folic acid insufficiency is a known predisposing environmental factor. Also, mutations of genes involved in folic acid metabolism are genetic risk factors for the occurrence of anencephaly.⁷ Mother was on folate regimen from the first antenatal visit in our case scenario. It is however essential that to have the desired effect, all women should receive folic acid before or immediately after conception. Unfortunately this isn't possible as most women are unaware of their pregnancy until after third week of conception, when folic acid supplements will be too late, since neural tube closes by 27th day.⁴

There are three types of anencephaly described: (1) meroanencephaly, where there is rudimentary brain tissue and partial formation of the cranium; (2) holoanencephaly, the most common type, in which the brain is completely absent, and (3) craniorachischisis, the most severe if the defect extends beyond the cranium.^{1,8} Our case clearly had a continuous calvarial defect extending down to have a cervical vertebral defect, thus it is a craniorachischisis.

In most instances, anencephaly occurs as an isolated birth defect with the other organs and tissues of the body forming correctly. In approximately 10% of cases, other malformations coexist with anencephaly.⁶ Neural tube defects may be associated with large thymus, small adrenal glands, hypoplastic lungs, cyclopia, syndactyly, absent radius and thumbs, club foot, imperforate anus, cleft palate, renal and cardiac anomalies.^{1,2,9} None of these associations were noted in our case, instead an interesting association of mediastinal bronchogenic cyst was noted. A case report of iniencephaly with congenital bronchogenic cyst has been reported in literature, but the same with craniorachischisis is a rarity.¹⁰ Significance of this case report is thus notable. Enigmas persist, however, regarding the relationship of NTDs to

malformations in organ systems widely disparate in developmental time and location.⁹

Folate supplementation to prevent recurrence, prenatal ultrasonogram and detection of elevated serum a-feto protein levels in early detection of NTD are important to reduce burden on parents.^{2,4,6,9} Counseling and advice on dietary supplementation and on starting folate regimen on planning next pregnancy were done in this case.

Conclusion

Anencephaly can be associated with various other system anomalies. Thus a thorough fetal autopsy is needed to document all associated anomalies, even if a second trimester 'anomaly ultrasound scan' might suffice for a diagnosis of anencephalic gestation.

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How to Cite this article :

Prashanth R, Shwetha S S, Rajashekhar KS, Raju VS, Hiremath SS. Anencephaly with Cervical Rachischisis (Craniorachischisis) and Congenital Bronchogenic Cyst: An Autopsy Case Report of a Rare Association. J Pub Health Med Res, 2014;2(2):61-3

Funding: Declared none

Conflict of interest: Declared none