CASE REPORT

Anencephaly with Cystic Hygroma : Antenatal Detection

Sudhaa Sharma*, Mandeep Singh**

Abstract

Anencephaly is the most common neural tube defect which is detected prenataly. We report here a rare combination of anencephaly with cystic hygroma.

Key Words

Anencephaly, Cystic hygroma.

Introduction

Anencephaly is the absence of cerebral hemispheres and cranial vault. There is absence of structures derived from forebrain and skull (1,2). The forebrain and midbrain are absent or replaced by rudimentary firbrovascular tissue with scattered islands of neural elements (referred to as the area cerebrovasculosa). The brainstem and cerebellum may be either spared or less severely involved (1). The facial bones and skull base are nearly normal in form, but parietal, frontal and squama of the temporal and occipital bones are present only as rudimentary fragments. It is a common congenital malformation and may be associated with anomalies like omphalocele, meningocele, spina bifida and polyhydramnios. Prognosis is poor and the condition is uniformaly fatal. Because the lesion is conspicious on sonography, the diagnosis is made in the second and third trimesters of pregnancy (3,4). We report here an unusual case of anencephaly associated with cystic hygroma detected on antenatal sonography.

Case Report

A twenty-six year old, second gravida reported for antenatal checkup in early third trimester of her pregnancy. Her general physical examination did not reveal any significant abnormality. On local examination, head of the fetus could not be palpated. Her routine laboratory tests of blood and urine were normal. Ultrasonographic examination showed a single live fetus, 30 weeks of gestational age, with absence of brain and carnial vault (Fig 1), prominent orbits at cranial end giving a "frog eye" appearance (Fig 2). A large multiseptate cystic mass was observed on the ventral aspect of the neck *i. e.* cystic hygroma (Fig 3). Keeping in view the poor prognosis of the fetus, parents decided to undergo for medical termination of pregnancy.

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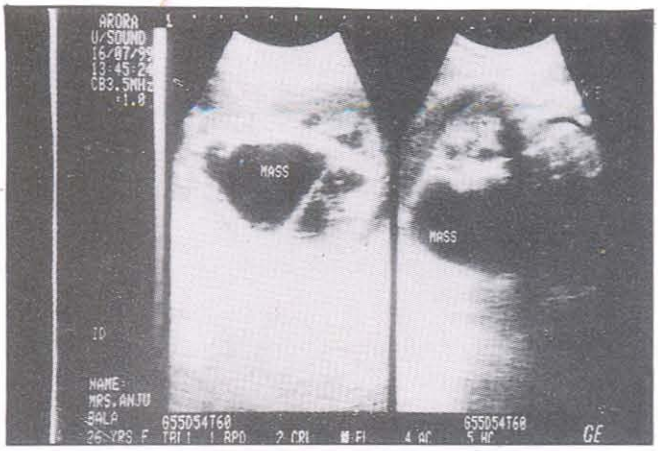

Fig. 1. Cranial end of the fetus showing absence of brain and bony cranial vault. Large cystic mass is noticed.

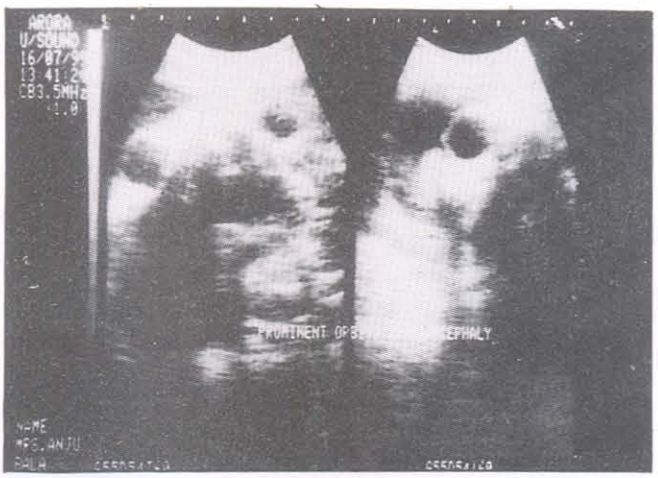

Fig. 2. Shows absence of cranial vault with prominent orbits – "Frog Eye" sign of Anencephaly.

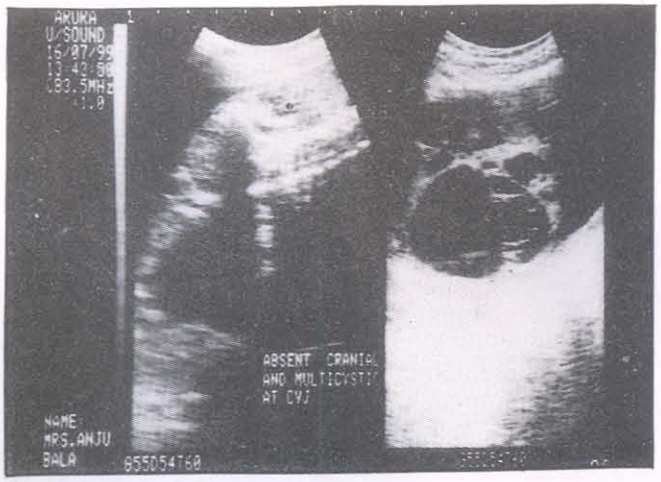

Fig. 3. Showing large multiseptate cystic mass ventral to craniovertebral junction of the fetus–cystic hygroma.

Discussion

Anencephaly results from the failure of closure of anterior neuropore, approximately at 24 days of fetal life. The severity of brain malformation associated with anencephaly, however, is variable. In the most severe form, holoanencephaly or holocrania, the brain is completely absent. In the mildest form, merocrania, a smaller defect in the rudimentary cranial vault is covered by area cerebrovasculosa (5). In all forms, the brain tissue that is present is abnormal (2,6,7), and the frontal bone is completely or nearly completely absent. Although, spinal defects may occur in association with anencephaly, the symmetric absence of bony skull vault is universal to anencephaly fetuses and represents the foundation of the sonography diagnosis. Epidemiology studies demonstrate a striking variation in prevalence rates. The highest incidence is in Great Britain and Ireland, and the lowest is in Asia, Africa and South America. Anencephaly occurs six times more frequent in whites than in blacks. Females are more often affected than males. Anencephaly is a lethal condition (8,9) and no specific treatment is available. Term infants with anencephaly who live for several days may respond to auditory, vestibular, and painful stimuli (10). Neuroendocrine defects are frequent, with failure of endocrine end-organ development secondary to a hypoplastic pituatary. Adrenal insufficiency may be associated with adrenocortical hypoplasia. The posterior pituatary is also hypoplastic and may cause clinical diabetes insipidus. The frequency with which polyhdramnios has been reported in association with anencephaly varies from 30% to 48% (1,7,11). But our patient had cystic hygroma with anencephaly. The term cystic hygroma is applied to a condition of marked lymphedema and lymphatic dilatation occuring

particularly in the cranial region during fetal life. It results from delayed communication of the juglar lymphatic sacs with the juglar veins which normally occurs at about 40 days of development. It is also believed that this cystic hygroma is a manifestation of "juglar lympatic obstruction sequence" (12). A high proportion of affected fetuses die in utero or shortly after birth. A cystic hygroma can be easily diagnosed in utero by ultrasonography, although, occasionally it may be difficult to distinguish it from encephalocele or other neural tube defects, haemangiomas and teratomas (13). An intact spinal column and lack of a solid component and presence of multiple septae in the mass aids in their differentiation. Cystic hygroma is often associated with chromosome anomalies like Turner's syndrome, hydrops or generalized edema.

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