CONGENITAL MALFORMATIONS ASSOCIATED WITH INIENCEPHALY

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INTRODUCTION: Iniencephaly is a rare birth defect having retroflexion of the head combined with severe distortion of the spine. Based on the presence or absence of encephalocele, Lewis [1, 9] has classified iniencephaly into two groups - iniencephaly apertus and iniencephaly clausus.

Cardiovascular disorders, diaphragmatic hernias, and gastrointestinal malformations are other additional defects seen in these cases. Most of iniencephalic babies are stillborn or die soon after birth (2, 9); however the milder forms of iniencephaly cases are not fatal.

Here we present a rare case of iniencephaly clausus with review of literature.

CASE STUDY:

HISTORY: A 22 years old pregnant female, G2P1L0A1, came for routine antenatal scan at 21 weeks gestation.

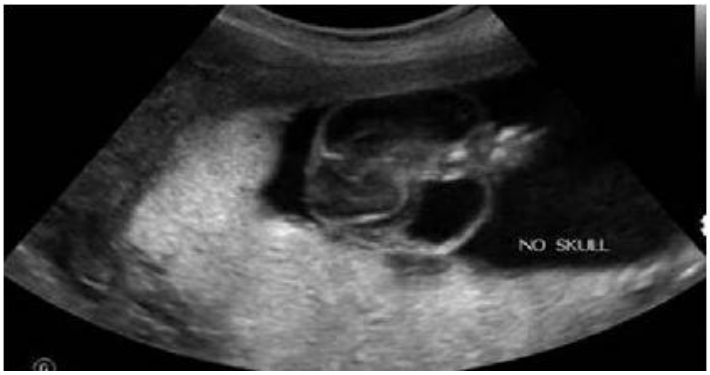
Results of a prior US examination at 7 weeks gestation confirmed a diamniotic dichorionic twin live intrauterine pregnancy corresponding to the gestation by last menstrual date.

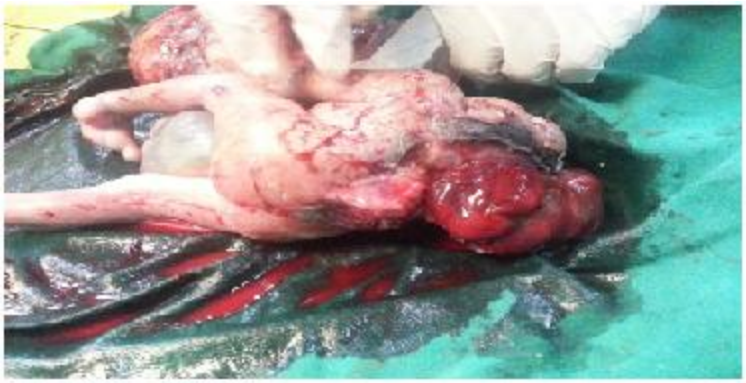
Her baby from previous pregnancy had meningomyelocele with deformity in lower limbs and died on 10th day of life in Neonatal Intensive Care Unit.

There were no intrauterine anomalies detected in one of the twin babies and is doing well post delivery.

The other foetus had multiple congenital anomalies.

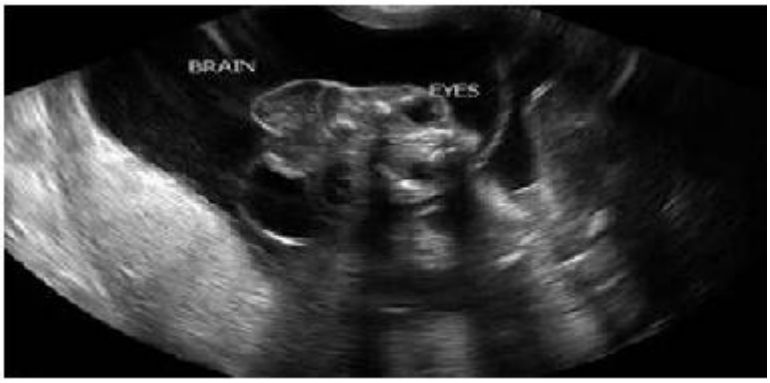
Ultrasound image shows absence of skull vault

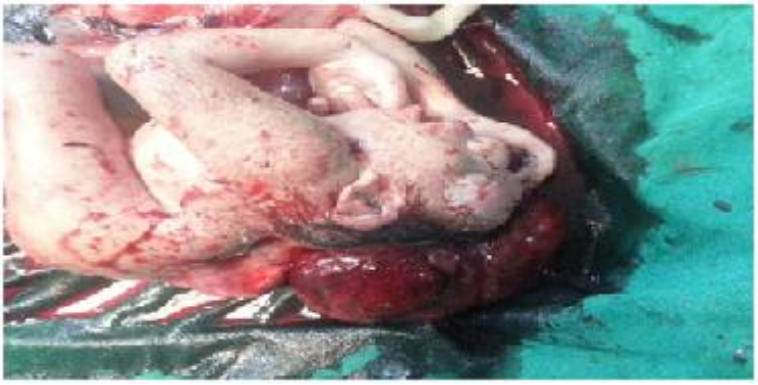




Gross specimen showing Spinal Rachischisis with retroflexion of neck

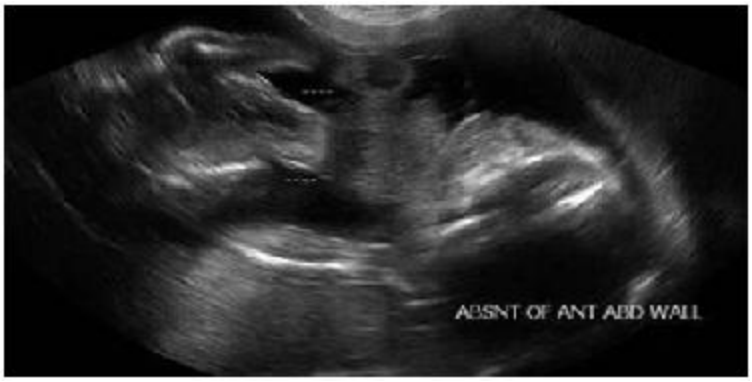
Ultrasound image shows absence of skull vault with brain matter lying in the amniotic fluid

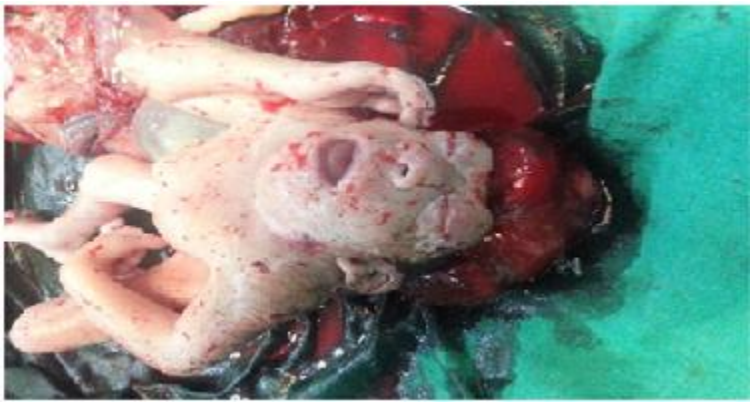




Gross specimen showing absent skull vault

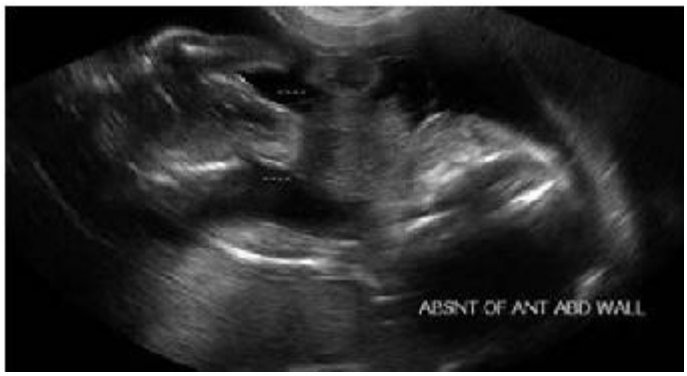
Ultrasound image showing anterior abdominal wall defect

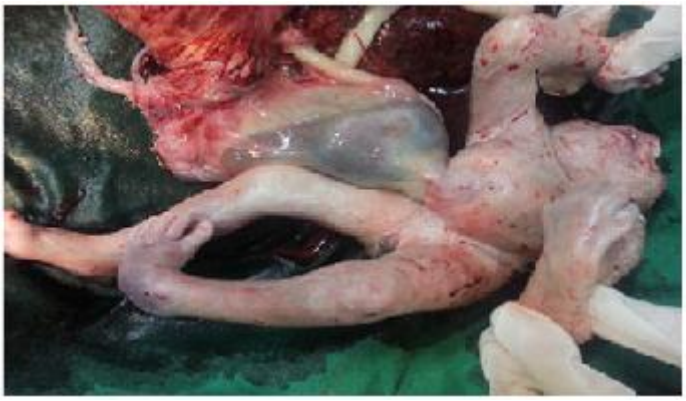




Gross specimen showing exaggerated cervico-thoracic spinal retroflexion with absence of skull vault

Ultrasound image showing defect of anterior abdominal wall with abdominal contents including the liver and heart herniating from the umbilical region





Gross specimen showing anterior abdominal wall defect and herniation

DISCUSSION: INIENCEPHALY is a rare neural tube defect that results in deficiency of occiput in the region of foramen magnum, spina bifida of considerable extent (up to spinal rachischisis) & cervico-thoracic spinal retroflexion (3, 5).

Iniencephaly is a developmental error occurring in early pregnancy prior to closure of cephalic neural folds at 24 days gestational age.

It is characterized by arrest as well as imperfect development of base of skull (portion of neural tube) and vertebral column (cervicothoracic region mainly).

At the early stage of embryo, paravertebral sclerotome (mesoderm) differentiates into two parts-

1] A ventral mass (forms the vertebral bodies, pedicles and their cranial homologous)

2] A dorsal mass (forms neural arches and vault bones of the cranium).

In iniencephaly, one or both of these masses are hypoplastic or ill developed. Incidence of Iniencephaly in India is 1:60,000 deliveries with the Female: Male ratio of 4.5:1 (6, 9).
In Simple Iniencephaly, the defect found in occipitocervical region indicates site 1 and probably site 4 closure defects.

In iniencephaly with anencephaly where vault and facial bones as well as lower vertebrae are involved, in addition to occipitocervical region, favours all 5 sites closure defect hypothesis.

Site 1 - Mid cervical.
Site 2 - Between prosencephalon and mesencephalon.
Site 3 - At stomodeum.
Site 4 - Caudal end of rhombencephalon.
Site 5 - Most caudal end of neural tube.

Exaggerated spinal retroflexion is due to absence of neural arches.

ANENCEPHALY is a rare neural tube defect that occurs when the head end of the neural tube fails to close, resulting in an absence of a major portion of the brain and skull.

Malformations/deformations and hypoplasia, for example, pulmonary hypoplasia, diaphragmatic hernia, omphalocele, etc. are due to lack of space (increased pressure) in thoracic and

abdominal cavity which is because of retroflexion and crowding of ribs (result of vertebral malformation).

Iniencephaly with anencephaly can be associated with other congenital malformations which are as follows:

Hydrops
Thymic hyperplasia
Diaphragmatic hernia/Eventration
CVS malformation
Pulmonary hypoplasia
Hepato-biliary malformation
Renal malformation
Adrenal hypoplasia &/or atrophy
GIT malformation
Omphalocele/Exomphalos
Inguinal hernia
Dandy Walker malformations

In our case, the congenital anomalies found in association with Iniencephaly and Anencephaly were Exomphalos Major (Ruptured) and Talipes Equino Varus.

Exomphalos literally translated from the Greek means 'outside the navel'. It is also called an omphalocele.

Omphalocele is a congenital abnormality in which the contents of the abdomen herniate into the umbilical cord through the umbilical ring. The viscera, which often include the liver, are covered by a thin membrane consisting of peritoneum and amnion.

A large defect may be associated with underdevelopment of the abdominal and thoracic cavities.

The incidence of exomphalos has increased slightly over the years and is approximately 1-2.5 in 5,000 births.

Epidemiological studies have found an association of iniencephaly and/or anencephaly with exomphalos in 5.2% cases with slight female preponderance.

Exomphalos results in a 4-12 cm abdominal wall defect which may be central, epigastric or hypogastric.

There are two varieties:

Exomphalos Minor - The sac is relatively small with the umbilicus attached to its summit.

Exomphalos Major - The sac is larger and the umbilicus is attached to its inferior aspect. It contains small and large bowel, and almost certainly, a portion of the liver.

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