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Case Series

Rare case series: iniencephaly, conjoined twins and anencephaly

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ABSTRACT

A discovery is said to be an accident meeting a prepared mind. This quote is true when we happen to see certain rare cases which we could not diagnose unless we are aware. The aim of this presentation is to bring attention of cases like Iniencephaly, conjoined twins, Anencephaly to our medical fraternity and the need to focus still more on health need of the women in reproductive age group in particular. This is also to highlight the benefits of periconceptional Folic acid supplementation and need for intense antenatal surveillance programmes and proper genetic counselling to affected parents.

Keywords: Iniencephaly, Anencephaly, Neural tube defects, Conjoined twins, Siamese twins, Folic acid supplementation

INTRODUCTION

Neural tube defects are the most common anomalies which occur during early developmental stages in pregnancy, as early as 21 to 28 dayspostconception.[1]

Certain neural tube defects are easily diagnosed in first trimester by ultrasound. Here we are presenting three cases, iniencephaly, anencephaly and conjoined twins.

CASE SERIES

Case 1

A 24 years old primigravida of non-consanguineous marriage, 24 weeks gestation presented to us for routine antenatal ultrasound scan. This foetus of 24 weeks was diagnosed as iniencephaly apertus.

The mother was counselled about the condition of the fetus and the termination of pregnancy was done.

Case 2

A 26 years old primigravida, of non-consanguineous marriage, 16 weeks gestational age was referred for obstetric ultrasound.

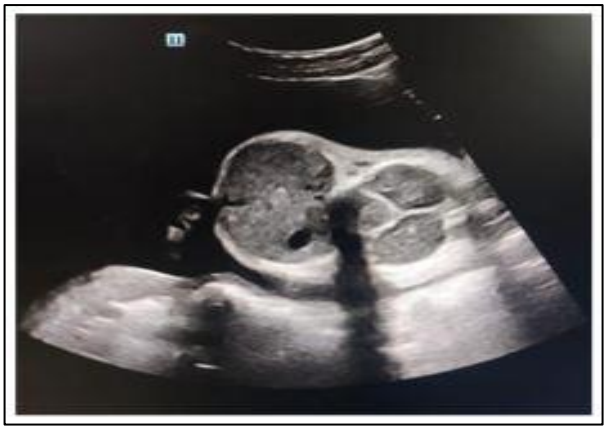

Figure 1: Fixed retroflexion of head. No separate neck.

She has no significant medical history like chronic illness or on any antiepileptic medications. She is underweight for her age.

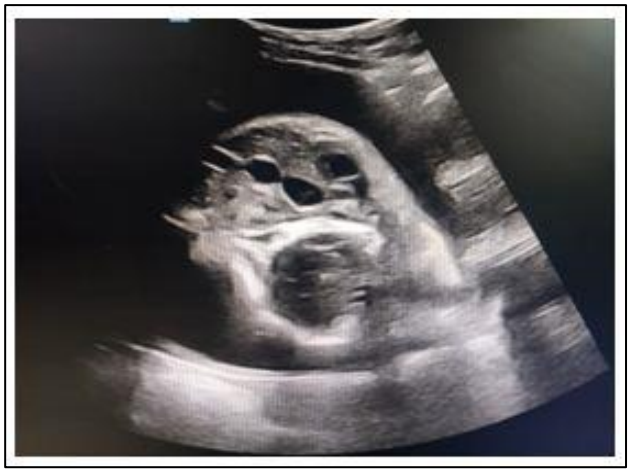

Figure 2: Stargaze posture -abnormal hyperextension of head over cervical spine.

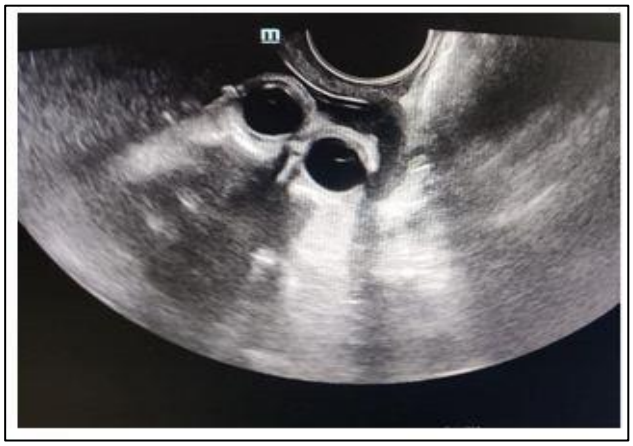

Figure 3: Hypotelorism, proboscis.

Figure 4: Stargaze posture with meningocele of expelled fetus.

Case 3

A 25 years old, unbooked primigravida of 25 weeks gestation came to us for routine antenatal scan. She had no prior ultrasound scans done elsewhere. She has no history of consanguinity. She has no history of chronic illness.

Termination of pregnancy was done by induction.

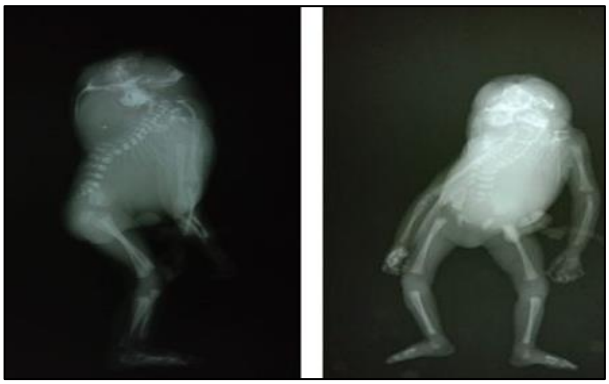

Figure 5: Babygram of the same expelled fetus.

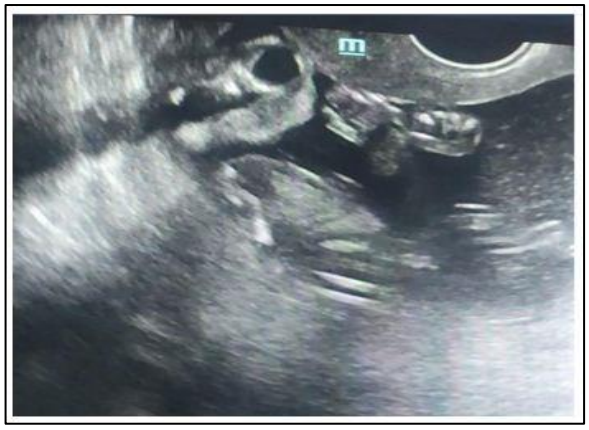

Figure 6: Frog eye sign. USG showed fetus of 16weeks gestation with absent cranium and exposed brain tissue.

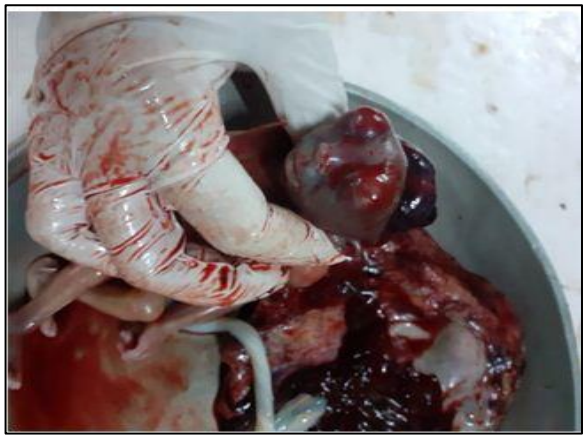

Figure 7: Pregnancy was terminated by induction.

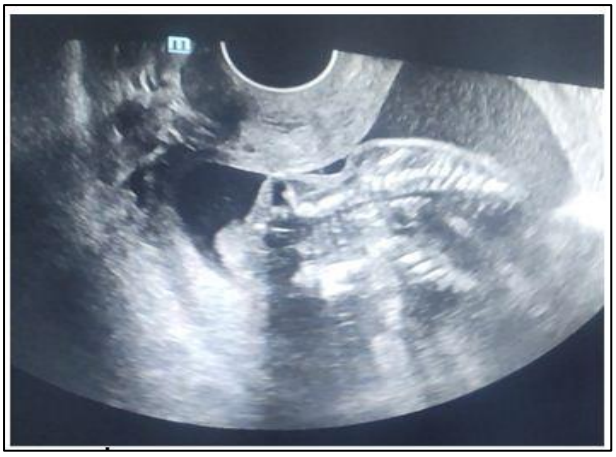

Figure 8: Craniothoraco Abdominopagus.

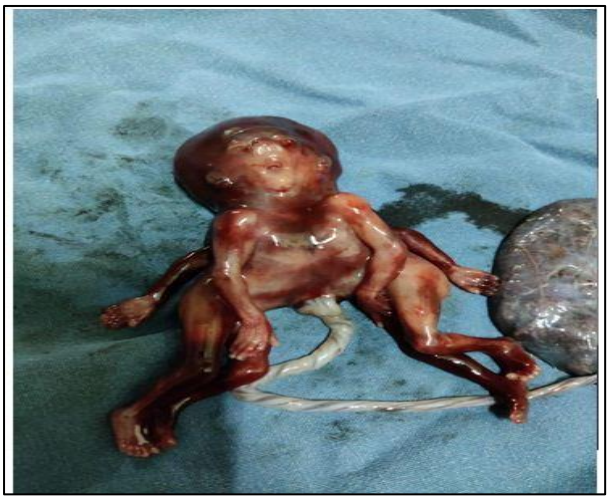

Figure 9: Expelled craniothoracoabdominophagic fetus.

DISCUSSION

Iniencephaly[2-14]

Iniencephaly is an uncommon NTD. Inion means nape of neck, occiput in Greek. The first case was reported in Saint-Hilare in 1836. Incidence is 0.1-10 in 10000 pregnancies. Earlier, embryology of neural tube closure was described as cranially upwards from cervical region and then caudally. But recent studies say that closure occurs at multiple sites: mid-cervical region, between prosencephalon and mesencephalon, stomodeum, caudal end of rhombencephalon and caudal end of neural tube.

Closure defects at mid-cervical and caudal end of neural tube is now proposed to be the cause of iniencephaly. Aetiology is unknown, several genetic and environmental factors are included. Numerous studies show that Folic acid 4mg daily reduces the risk.

The salient features are hyperextension of head on cervical spine, occipital bone and cervical vertebrae defect. Iniencephaly is incompatible with life and is lethal. Genetic counselling should be offered to those as there is high suspicion of chromosomal anomalies in non isolated iniencephaly and craniorachischisis.

There are two types:

Iniencephaly Apertus: there is associated meningocele.

Iniencephaly Clausus: there is associated spinal defect.

Differential diagnosis includes physiological cervical hyperextension. Here no occipital or cervical vertebrae defect will be seen

Anencephaly with retroflexion: Anencephaly will not have skin covering or cervical vertebrae defect unlike Iniencephaly Apertus. The Calvarium is absent in Anencephaly.

Klippel Feil Syndrome: There is failure of segmentation defect in cervical vertebrae and is treated surgically. Retroflexion of head will not be seen in KFS.

Clue to USG diagnosis: Abnormal hyperextension of head with short and deformed spine.

Anencephaly[15-22]

Anencephaly is cranial neural tube closure defect. It is 100% preventable with periconceptional Folic acid supplementation. UK and Hungarian studies showed that periconceptional Folic acid supplementation has significantly reduced the incidence of NTD in babies. Genetic research shows that MTHFR gene encodes the enzyme involved in intracellular metabolism of Folic acid. Polymorphism of this gene causes decreased enzymatic activity which is also dependent on plasma Folate levels. Various other aetiologies include maternal hyperthermia, Valproate use during pregnancy for epilepsy, certain essential nutrients excess and deficiency, chronic maternal diseases like diabetes mellitus.

Differential diagnosis

Amniotic band syndrome

There will be limb, body wall and spinal abnormalities due to amputation. This differential diagnosis is based on the symmetry of cranial defects which occurs in almost all Anencephaly fetuses.

Learning points

Periconceptional Folic acid supplementation before pregnancy and upto 12 weeks of pregnancy significantly reduces NTD, and 100% of anencephaly fetuses. Early diagnosis of anencephaly in first trimester and Iniencephalyis essential to plan safe delivery.

Conjoined twins/Siamese twins[23-27]

Conjoined twins arise from late twinning of single zygote. They usually share single placenta and Umbilical cord. They are classified according to their site of fusion.

Thoracopagus: common.

Omphalopagus: abdomen.

Craniopagus: head.

Cephalopagus: face.

Ischiopagus: pelvis.

Pyopagus: sacrum.

Two postulated theories include:

Fission theory: There is incomplete split of fertilised egg causing incomplete separation of embryonic mass after 12 days of fertilisation.

Fusion theory: Like-stem cells of two fetuses fuse after complete separation of fertilised egg.

Conjoined twins are generally not compatible with life. Appropriate thorough radiological investigations should be carried out in centres where safe obstetric and paediatric surgical facilities are available for safe separation. In such centres, elective caesarian section is planned at 36-38 weeks of pregnancy.

Fetal echo is mandatory in thoracic fusion. MRI and CT is for assessment of bone and anatomical details, viscera and vascular sharing details. Contrast radiography is done for gastrointestinal and urogenital tracts evaluation. Angiography is to assess the vascular supply.

Management

Once diagnosed, they are categorised into 3 categories. No surgical intervention when there is no possibility of creating single functioning heart due to cardiac fusion. Emergency separation in case of death of one of the twins /dying one twin /lethal anomaly of left untreated. Planned, precise surgical mapping when infant's condition is stable.

Usually separation is planned around 3 months of age. In developing countries like India, where optimum separation /surgical facilities are not at ease, parents should be counselled about the outcomes and if termination is opted, it should be done as early as possible.

Diagnostic clues

Two fetuses facing each other with constant relative position of fetusesover time, head and other body parts persistently at the same level.

CONCLUSION

Antenatal USG plays a crucial role in determining the survival potential of fetusat risk. Though there is a popular teaching that rare presentation of common diseases is more important than common presentation of rare cases, radiologists should be aware of certain rare anomaliesto minimise the complications if undiagnosed or diagnosed at later stages of pregnancy. Women of reproductive age group should be offered Folic acid supplementation to reduce the incidence of preventable neural tube defects.

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