

## **Craniorachischisis Totalis: First Case Reported in Egypt**

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### **Abstract**

Craniospinal rachischisis is a rare and severe form of neural tube defects (NTDs), fatal and described as anencephaly. With a bony defect of the spine and exposure of neural tissue to amniotic fluid. With thorough review in PubMed, the prenatal diagnosis with characteristics sonographic images of this anomaly have not been previously published in Egypt. Here I present prenatal sonographic images of this anomaly detected during 22 weeks of gestation through two-dimension ultrasound images showed anencephaly, spina bifida, we describe the patient with ultrasonographic and post termination imaging appearance of craniospinal rachischisis totalis, detected antenatally at 22 weeks of gestation, and confirmed after post mortem examination of abortus parts of pregnancy. The patient spontaneous abortion of pregnancy and delivered a fetus with anencephaly and rachischisis totalis as described in utero imaging ultrasound findings.

**Keywords:** *Craniospinal rachischisis; Neural Tube Defects (NTDs); Anencephaly*

### **Introduction**

Craniorachischisis is a rare and severe form of NTD that results when the neural tube fails to close normally in the primary neurulation/dorsal induction stage of the central nervous system development, during the 3<sup>rd</sup> - 4<sup>th</sup> week of gestation, leading to fetal loss and stillbirth [1-3].

The failure of closure of cranial pore and parts of neural tube produces the two severe forms of NTDs, namely, the anencephaly and spinal rachischisis totalis. Ultrasound has established itself as a sensitive and accepted modality of choice for the prenatal detection of all congenital anomalies, including the NTDs [4]. Recent studies have shown the complementary NTDs are one of the few congenital defects amenable to preventive measures. Maternal folic acid supplementation significantly reduces the number of NTDs including lethal defects such as acrania, exencephaly, anencephaly, and rachischisis, with various studies showing 35% - 75% reduction in NTDs by maternal folic acid ingestion [5]. I present one case of 22 weeks of gestation who came for routine antenatal care. With examination obstetric scan and diagnosed anencephaly and craniorachischisis that was confirmed after spontaneous abortion after few days of diagnosis.

### **Case Report**

19-year-old primigravida presented at 22 weeks of gestation for her routine antenatal examination. This pregnancy was a spontaneous conception, confirmed by ultrasonographic examination at 8 weeks with ultrasonographic scan measures. no history teratogenic exposure during pregnancy. Once diagnosed pregnancy at 8 weeks at first antenatal visit, she started taking oral folic acid (0.5 mg). There was no history of chronic illness, hypertension, diabetes mellitus, thyroid disorders, psychiatric disorders or chronic intake of medications. There is family history of diabetes, hypertension, there is no family history of multiple pregnancies, or congenital anomalies.

Ultrasonography (USG) evaluation revealed a single live fetus Intra uterine of unstable lie, average amniotic fluid. Fetal heart rate was 170 bpm. Placenta was posterior. Congenital abnormalities detected mainly in fetal skull and spine. Abnormal skull with absent Fetal calvarium, the brain tissue appeared deformed within amniotic fluid (exencephaly), and the orbits were prominent. Abnormal fetal spine of absence of vertebral posterior elements with splaying of the lamina at all levels and exposing the spinal cord to amniotic cavity (Figure 1 and 2).



**Figure 1:** Transabdominal ultrasonography image showing absence of posterior elements with splaying of lamina and exposure of spinal cord to the amniotic fluid and associated with pleural effusion.



**Figure 2:** Transabdominal ultrasonography image showing absence of posterior elements with splaying of lamina and exposure of spinal cord to the amniotic fluid.

With sagittal view of the spine showed loss of the normal thoracic kyphotic curvature (Figure 5). These findings were consistent with craniospinal rachischisis totalis (Figure 7). The absence of calvarium with deformed brain tissue directly exposed to amniotic cavity suggestive of exencephaly (Figure 3 and 4). These findings confirmed a diagnosis of craniospinal rachischisis totalis. The prognosis was explained to the patient.



**Figure 3:** Transabdominal ultrasonography image showing absence calvaria with deformed brain tissue.



**Figure 4:** Transabdominal ultrasonography image showing absence calvaria with deformed brain tissue.

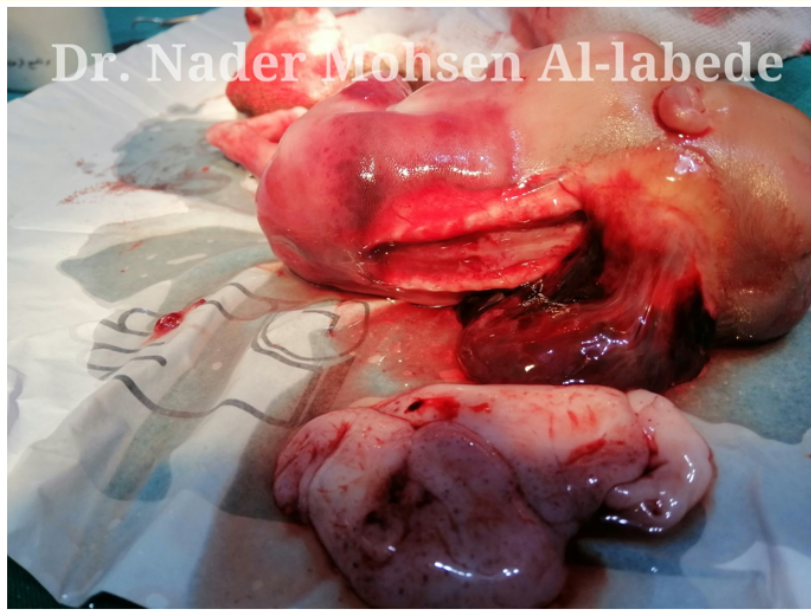


**Figure 5:** Spine showed loss of the normal thoracic kyphotic curvature.

Within few days spontaneous abortion with delivery of a fetus with exencephaly and spinal rachischisis confirming the in utero imaging findings (Figure 6-7).



**Figure 6:** Short neck, ears and orbits were prominent.



**Figure 7:** Postmortem showing craniospinal rachischisis totalis.c

## Discussion

NTDs are common, congenital malformations resulting from complete or partial failure of the neural tube to close in the developing embryo, preventing muscle and bone from growing around the gap. Clinically, there are two types of neural tube disorders open and closed types. Among these, closed NTDs affect mainly the spinal cord without significant cranial involvement. However, open NTDs often have cranial involvement. Depending on the location and severity, open NTDs can be classified as myelomeningocele, myeloschisis, or rachischisis. Most severe of these conditions is rachischisis which is incompatible with life. A PubMed search for rachischisis totalis revealed only six such reports [6-10]. Anencephaly can be easily detected by ultrasound in first trimester the characteristic appearances are the "Mickey Mouse" sign in early pregnancy and the "frog eyes" sign in the second trimester [11].

Craniospinal rachischisis totalis being a lethal congenital malformation necessitates early termination of the pregnancy. This is associated with significant morbidity in the women of reproductive age group. In addition, there is a risk of recurrence of NTDs in the subsequent pregnancies.

## Conclusion

Rachischisis totalis and anencephaly is a rare fetal anomaly this case is the first case reported in Egypt. This anomaly is not compatible with life and the diagnosis is important and extremely challenging, thus high lighting the importance of a thorough ultrasound examination. Termination of pregnancy is recommended upon diagnosis, with a detailed consultation followed by close-up monitoring for future pregnancies. I described the first case of prenatal sonographic images of anencephaly and rachischisis totalis in Egypt. Awareness of these rare associations will avoid missed diagnoses and help in prenatal counselling, thus high lighting the importance of a thorough ultrasound examination.

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