

# A diagnosed case of anencephaly and severe neural tube defect at 30 weeks, antenatal and intrapartum aspects

## Abstract

With routine prenatal diagnosis offered to pregnant women, cases of anencephaly are diagnosed and managed in the early stages of pregnancy. As cases of anencephaly have a lethal outcome, termination of pregnancy is offered. This has led to a lack of familiarity for clinicians on how to manage on-going anencephalic pregnancies from the obstetrics point of view. We present a case of late diagnosis of anencephalic pregnancy where the mother has opted to continue with the pregnancy.

**Keywords:** anencephaly, neural tube defect, spina bifida, antenatal care, labour

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**Abbreviations:** NTD, neural tube defects; BHCG, beta human chorionic gonadotropin; A&E, accident and emergency; ECV, external cephalic version

## Introduction

The global incidence of neural tube defects (NTDs) ranges from 1.0 to 10.0 per 1,000 births. The commonest forms of NTDs are Anencephaly and spina bifida.<sup>1</sup> Anencephaly is secondary to the failure of fusion of the cranial neural tube. It is fatal and leads to stillbirth or early neonatal death.<sup>2</sup> Nearly half of the live born anencephalic babies have a life expectancy of between a few minutes and one day. One in four live born anencephalic babies could live up to 10 days.<sup>3</sup> Risk factors of anencephaly include; social and epidemiological factors, maternal and paternal ages, hyperthermia during early pregnancy, diabetes or obesity, and maternal use of caffeine and medications during early pregnancy have been linked to increased incidence of the condition.<sup>4</sup> Folate deficiency is a known risk factor and folic acid supplements before and in early pregnancy results in a decreased risk of anencephaly.<sup>5</sup>

The introduction of antenatal screening has led to the early diagnosis of anencephalic cases either through the biochemical markers or on the ultrasound scan. The detection rate of anencephaly at 20 weeks anomaly scan is expected to be above 98%. Almost all cases of anencephaly are diagnosed and managed before viability.<sup>6</sup> We present a case of anencephalic fetus first diagnosed at between 29 weeks of gestation.

## Case presentation

A 20-year-old female patient was referred to the Accident and Emergency department (A&E) complaining of a non-specific abdominal pain. She gave a history of Irregular cycles over the last 9 months and missed periods for 7 weeks. Large abdominal girth was noted whereas routine blood results were normal; Free BHCG

was 86369U/L. Obstetric review and examination was performed in addition to a departmental ultrasound scan. The scan revealed a singleton intrauterine pregnancy at 29 weeks and 4 days of gestation based on the fetal biometry. Ultrasound findings were suggestive of Anencephaly. At that stage, the patient was happy to continue with the pregnancy.

A tertiary centre fetal medicine scan was performed the following day. This confirmed the presence of a severe neural tube defect starting at the level of the upper thorax with separation of the vertebral bodies (Figure 1). Fetal skull and brain could not be demonstrated and anencephaly was confirmed. The amniotic fluid volume was raised with an index of 34centimetres and the presenting part was cephalic. The lethal nature of the condition was explained to the patient and options were discussed. This included expectant management knowing the lethal outcome or to interrupt the pregnancy (potentially feticide however this would not be mandatory). The patient chose to interrupt the pregnancy without feticide; this was planned and performed at her regional Hospital. We were not aware of any attempts by the patient to terminate this pregnancy outside the national health system (NHS).



**Figure 1** Severe form of Neural tube defect starting at the upper thoracic level, with Anencephaly.

Other issues have been highlighted to the patient regarding the plan for delivery. The fact that the baby had no skull vault could lead to prolonged labour, dystocia with body part prolapse which might require surgical delivery including a caesarean section. This would be preferably avoided due to her young age and the poor outcome for the fetus. Additionally, spontaneous rupture of membranes with polyhydramnios carries a risk of placental abruption.

A plan was made to induce labour using mifepristone tablets orally (Progesterone receptor antagonist used to ripe the cervix and initiate labour). Aiming at decreasing the risk of surgical delivery and antepartum haemorrhage, External cephalic version (ECV) into a breech presentation and amnioreduction would be performed on the day of admission and before starting the misoprostol tablets.

The patient received the mifepristone and was admitted under observation waiting for the ECV and amnioreduction as planned. Several hours after admission, she progressed quickly into labour and had spontaneous rupture of the membranes. Spontaneous vaginal delivery of a stillborn baby took place with no maternal complications. Postnatal karyotyping showed a female with normal chromosomes.

## Discussion

This case describes the unusual encounter with an anencephalic fetus diagnosed late in pregnancy. On this occasion the pregnancy was unplanned and the patient's periods were irregular. The patient did not have the opportunity to receive the prophylactic dose of Folic acid as recommended by the national guidelines. One of the issues is that a number of viable born anencephalic fetuses would survive for hours up to a number of days. The debate would be whether to perform feticide or not before ending the pregnancy, in any case it would be important to offer this option to the patient and allow her to have an informed choice.

Since the introduction of the national fetal anomaly screening almost all cases of anencephaly are diagnosed and managed early before fetal viability due to the high detection rate on ultrasound scan. The Obstetric management of labour in pregnancies with anencephalic fetus at term or near term has become more challenging due to the lack of familiarity and clinical experience. The risks include polyhydramnios, placental abruption and antepartum haemorrhage following spontaneous rupture of membranes. Others include malpresentation, prolonged labour due to the absence of a presenting part in cephalic presentation and an increased risk of surgical management in the second stage of labour.

The delivery plan aimed at turning the fetus into breech, which would be a better presenting part on this occasion. Although there is generally a lack of fetal muscle tone in anencephalic fetuses, it is important to have a presenting part in the lower uterine segment that would promote cervical dilatation. Amnio drainage was to ensure the fetus would not turn back into a cephalic presentation and it reduces the risk of abruption.

The patient had presented in advanced labour with quick progress and good outcome, having said that it is important to perform a risk assessment with such rare clinical scenarios and have a plan in place beforehand to minimise the risk of complications.

## Acknowledgments

None.

## Conflicts of interest

The authors declare there is no conflict of interests.

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