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Case Study

A Case Report on Anencephaly

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I N F O

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A B S T R A C T

A newborn with anencephaly does not have parts of the brain or skull. It is a significant birth condition. A particular sort of Neural Tube Defect (NTD) is present. The neural tube helps form the baby's brain and skull (upper segment of the neural tube), spinal cord, and back bones (lower half of the neural tube), as it grows and closes.When the top portion of the neural tube does not completely seal, anencephaly occurs. As a consequence, many babies are born without the cerebrum, which is the part of the brain responsible for thought and coordination, or the front portion of the brain (forebrain). Brain tissue that is not covered by bone or skin is commonly exposed. We are presenting one such case with 23 weeks of gestation and an anencephaly baby with cardiac activity.

Keywords: Anencephaly, Neural Tube Defect, Ultrasound, Foetal Magnetic Resonance Imaging (MRI), Amniocentesis

Introduction

Anencephaly is a brain malformation that occurs at birth. It occurs when a newborn's brain, skull, and scalp do not develop normally during pregnancy.[1] Babies with this condition die within a few hours or days of birth.[2] Taking the proper dose of folic acid before and throughout pregnancy may reduce the likelihood of having a newborn with anencephaly.[1]

Anencephaly affects around one out of every 5,000 to 10,000 newborns, and it affects more girls than boys. The majority of pregnancies involving anencephaly end in miscarriage or stillbirth.Women, who have already had a child with a Neural Tube Defect (NTD), such as spina bifida, are more likely to have another child with anencephaly.[1,3] This condition does not appear to be a hereditary disorder (passed down in families).[2] In a vast majority of instances, there is no family history of the condition.[1] Female foetuses make up around 70% of anencephalic foetuses. It is more common among first-time moms, as well as in young and elderly mothers. There may be genetic and environmental components (multifactorial) associated with this condition.

Diagnosis

In the early part of the pregnancy, alpha - fetoprotein levels in the amniotic fluid are utilised to establish the diagnosis. Sonography is a diagnostic tool. Around 10-week-old babies show (a) no cranial vault and (b) angiomatous brain tissue. Even with hydramnios, making a diagnosis in the second half of pregnancy might be difficult because of the complication. Suspicion is raised when the foetal head may be felt on palpating the abdomen. The facial presentation diagnosis is established even during an internal examination. Sonography is used for confirmation.[4]

Complications include (1) hydramnios (70%), (2) malpresentation (breech or face), (3) premature labour, particularly when related to hydramnios, (4) postmaturity tendency, (5) shoulder dystocia, and (6) obstruction of labour if the head and shoulders attempt to engage simultaneously due to a short neck.

Prevention

Pregnancy counselling is essential. The incidence of NTD has been greatly decreased by folic acid treatment starting one month before conception and continuing until roughly

12 weeks of pregnancy (85%). The suggested daily dosage is 4 mg. In a second pregnancy, there is a 2% chance of recurrence.[4]

Case Presentation

The primary health centre recommended a 26-year-old female patient to the tertiary health centre. She was Gravida 2, Parity 1, Live 1, with 23 weeks of gestation and an anencephaly baby with cardiac activity. The diagnosis was confirmed by ultrasonography and was expected to be above 96% with a gross congenital anomaly.

The mother was informed of the baby's condition and agreed to the induction of labour. The family gave the consent for do-not-resuscitate code. The mother was brought into the labour and delivery area. After 4 hours of induction of labour, she started having labour pains. Her general condition was good and she was well-hydrated. Her period of gestation was 27 weeks, blood pressure was 110/70, and pulse was 84/minute. She delivered a baby girl weighing 495 kg with moderate anaemia. The baby did not cry after birth. Heart rate was 46 beats/minute and respiratory rate was 29/minute. When the infant was born, she had a severe congenital abnormality. The baby expired after 1 hour of birth.

Discussion

Anencephaly is a common and fatal neural tube defect which occurs due to the defective closure of the rostral pore of neural tube.[5] It can be diagnosed by ultrasonography and increased levels of alpha-fetoprotein. The incidence of anencephaly has been reported to show variations among different geographical regions.[6] The lower incidence reported in India may be due to early termination of diagnosed foetuses, which reduces the prevalence at birth, and changes in the level of ANC.[7] It has been reported that anencephaly occurs more frequently in female than in male foetuses.[8,9]

A vast majority of anencephaly babies die before birth, and the pregnancy results in a miscarriage.[1,2] Babies born with anencephaly usually die within a few days or weeks.[2,10,11] Usually, anencephalic babies are unconscious, blind, and deaf; parents may be concerned that their baby is upset or unhappy,[2] but these babies cannot feel pain.[4,12] However, their brainstem may respond to some reflexes, such as breathing, sound, and touch.[13]

Maternal exposure to hyperthermia around the time of anterior neuropore closure is significantly related to anencephaly.[14] The exact causes of anencephaly are not known. However, the interaction of genetic and environmental factors is considered to be implicated. Some of its causes, such as genes which provide instructions for making a protein involved in processing the vitamin folate, have been identified.[15]

Anencephaly is a deadly disorder that makes living a lengthy life impossible. Anencephalic infants are thought to die shortly after birth,[3] according to long-held beliefs. Anencephaly is deadly during the first year of life, according to numerous pieces of research. Foetal mortality was found to be 100% during the first few days to weeks of life in other studies as well.[1,11,16]

Conclusion

Anencephaly is a gross congenital anomaly. It is a type of neural tube defect that results from a failure of the anterior (rostral) portion of the embryonic neural tube (anterior neuropore) to close properly. The deformity stems from inadequate brain tissue and skull vault development, yet the face section is normal. The prognosis of anencephaly is very poor. Termination of pregnancy could be the best option but, due to various cultural and religious beliefs, it is not practised in India. Moreover, in India, awareness regarding lethal defects is very low among the population. As no curative treatment is available so creating awareness should be the key component in the prevention of such problems.

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