

## Case Report



### Anencephaly: A case report highlighting the need for integrating Ayurvedic and modern approaches in Prenatal Care

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#### ABSTRACT:

**Introduction:** Anencephaly is a severe form of congenital defect. It has no effective treatment with a poor prognosis. The exact aetiology of anencephaly remains unclear. The causes of birth defects may be due to genetic and environmental factors, and can even result from multifactorial inheritance.

**Clinical findings:** A 22-year-old primigravida woman delivered a macerated stillborn male baby of 260 gm by vaginal route at 24 weeks of gestation. On examination of the newborn showed the absence of cranial vault, with exposed, malformed neural tissue replaced by an angiomatous mass and diagnosed as anencephaly. **Conclusion:** This case emphasizes the importance of early prenatal diagnosis, preconception education, and adoptable preventive measures to reduce the risk of neural tube defects. Genetic counselling should be offered, particularly for pregnancies that are at high risk or unplanned. Along with this, integrating Ayurvedic principles, such as *garbhini paricharya* and avoidance of *garbhopaghathakara* bhavas, with contemporary medical practices can provide a holistic approach to maternal care. Such integration may help to lower the incidence of congenital anomalies and promote healthy pregnancy and offspring.

**KEYWORDS:** Anencephaly, Preconception Education, Preventive Strategies, Prenatal Care, Ayurvedic & Modern Practices.

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## 1. INTRODUCTION

Anencephaly is a severe congenital defect characterized by the absence of a major portion of the brain, skull cap, and scalp. This is considered a type of neural tube defect (NTD). The development of the central nervous system (CNS) begins with the embryonic process of neurulation at the beginning of the third week of intrauterine life. Neurulation includes the formation of the neural plate, neural groove and neural folds. The lateral edges of neural folds approach the cervical region and proceed cranially and caudally, forming the neural tube. An upper expanded end of the neural tube forms the brain, and the lower tubular part forms the spinal cord. Till the fusion is complete, the cranial and caudal ends of the neural tube connect with the amniotic cavity. Before the complete closure of the neural tube, the cranial end is open, and it is closed approximately by the 25<sup>th</sup> day. If there is any distortion in the closure of the cranial end, it leads to a defect called anencephaly. [1]

Congenital anomalies share the major burden for stillbirths, infant mortality and morbidity. The prevalence of CNS defects is highest in cases of stillbirths. [2, 3] A meta-analysis study revealed the overall prevalence of NTD births was 4.1 per 1000 births in India. Among the NTDs, the prevalence of anencephaly was highest at 2.1 per 1000 births, followed by spina bifida at 1.9 per 1000 births. NTDs can be prevented by preconception folate supplements. [2, 3] Here, we are presenting a case of anencephaly and its risk factors, and the need for prenatal education to prevent untoward incidents.

## 2. CASE REPORT

A 22-year-old woman was admitted to a tertiary hospital for medical termination of pregnancy after a diagnosis of anencephaly fetus. She resides in a rural area and belongs to a low socioeconomic background. The patient is uneducated and works as a construction labourer. She has been in a second-degree consanguineous marriage for the past three years. She does not have any habits of chewing alcohol, tobacco, and pan-gutkha consumption. The patient presented with no significant clinical symptoms or complaints other than those associated with her pregnancy of 24 weeks of gestation. There was no history of fever, infection, or exposure to teratogens during the pregnancy. Routine antenatal care had been followed, and the diagnosis of anencephaly was made during a routine ultrasonography (USG) scan.

**Antenatal History:** She is a primigravida of 24 weeks of gestation as per her last menstrual date (LMP). Her menstrual cycle was normal with regular flow. The pregnancy was uneventful. No history of cardiac and renal diseases, gestational diabetes, pre-gestational diabetes, preeclampsia, and eclampsia. Mother claimed that she is not taking any medication other than folic acid and calcium. Neural tube defects and other congenital abnormalities were not reported in the family. The current conception was unplanned and occurred naturally. She is not taking any nutritional supplements before pregnancy.

**Physical examination and Investigations:** She was lean-built, weighing 50 kg with a height of 152 cm. On examination, she looked pale, and there was no

oedema. Her pulse and blood pressure were within normal range. Her haemoglobin was 9.9 g/dL with a B-negative blood group. Her random blood sugar was 120 mg/dL. Her urine, HIV, and HbSAG tests were found to be negative. A routine antenatal transabdominal USG was carried out at 20 weeks. The fetal anatomy revealed the absence of a cranial vault, suggestive of anencephaly. The spine appeared normal, with no evidence of spina bifida. The heart, lungs, kidneys and bladder appeared normal. Single intrauterine gestation with cephalic presentation was noted, corresponding to a gestational age of 20 weeks as per LMP. The estimated fetal weight was 190 gm according to fetal biometry. Anencephaly was diagnosed, which is incompatible with life. The patient was counselled regarding the condition and the available options. After considering the prognosis, the patient opted for medical termination of pregnancy. The termination was carried out following the standard protocol for pregnancies complicated by severe congenital anomalies.

**Findings:** She delivered a macerated stillborn male baby of 260 gm by vaginal route at 24 weeks of gestation. On examination of the newborn, the cranial vault was absent and nervous tissue was exposed with protruded eyes. The brain was malformed, degenerated and replaced by an angiomatous mass and diagnosed as anencephaly (Figures 1 & 2). The spine appeared normal. No craniofacial and other systemic congenital malformations were noticed. The umbilical cord shows one vein and two arteries.



**Figure 1 – Anterior view of an anencephalic baby**



**Figure 2 – Superior view of an anencephalic baby**

### 3. DISCUSSION

The occurrence of NTDs varies across different regions. In the US, it was seen lowest, 1 in 1000 births and highest, 1 in 200 births in China. [1] A meta-analysis study shows the pooled prevalence of congenital anomaly-affected births was 184.48 per 10,000 births in India. [2] The prevalence of CNS system defects was higher in stillbirths and musculoskeletal system defects in the case of live birth. [2, 4] A population-based study showed an alarming incidence of 7.48 per 1000 live births of NTDs in North India. [5]

Multiparity, elderly pregnant women, consanguinity and low birth weight are some other factors associated with congenital malformations. [4] Lower levels of folate,

vitamin B-12, with elevated homocysteine levels in pregnancy, increase the risk of having NTD babies. [6] Chronic maternal conditions like diabetes mellitus, use of valproate during pregnancy, maternal hyperthermia, deficiency and excess of certain nutrients are reported to increase the possibilities of occurrence of NTDs. [7]

Anencephaly is a fatal congenital anomaly with no effective treatment and a poor prognosis. Its exact cause remains unclear. Birth defects result from unknown aetiology and multifactorial inheritance in 70-75% of cases. Genetic factors, chromosomal abnormalities and environmental factors also contribute to its occurrence. Even though the embryo is well protected in the uterus, exposure to certain harmful environmental agents or teratogens accounts for nearly 7-10% of developmental disruptions. High doses of teratogens during the critical period of brain and other organogenesis (3-16 weeks of pregnancy) can lead to major birth defects [8]. Awareness of teratogen agents, which disrupt fetal development, offers the opportunity to prevent some birth defects. Human teratogen agents like cigarette smoking, alcohol consumption, cocaine and methadone affect foetal growth development, mental deficiency and diverse birth defects. Consuming oral contraception during an unrecognised pregnancy has been associated with VACTERL syndrome (vertebral, anal, cardiac, tracheal, oesophageal, renal & limb anomalies). Certain drugs, which cross the placental membrane and exhibit teratogenicity during pregnancy, such as antibiotics (streptomycin, tetracyclines), thalidomide, diazepam, oxazepam, acetaminophen (paracetamol), aspirin, warfarin, phenytoin and valproic

acid. Rubella virus, cytomegalovirus, herpes simplex virus, chickenpox, human immunodeficiency virus and treponema pallidum (syphilis) virus cause various birth defects. Pregnant ladies should avoid raw meat and close contact with domestic animals to reduce the risk of the toxoplasma parasite infection. Additionally, consuming fish with high levels of organic mercury may have increased the risk of neurological and behavioural disturbances in newborns. Some nutritional factors, such as long-term exposure to large doses of vitamin A, and maternal iodine deficiency, may also contribute to the origin of birth defects. [8] The prevention of NTDs using folic acid supplementation in the preconception period is well established. [2, 9] Congenital disorders involving genetic components have hardly any cure; hence, early diagnosis and preventive measures are of utmost importance. Most congenital anomalies are prenatally diagnosed by USG, amniocentesis, chorion villous biopsy, fetoscopy and estimation of maternal alpha-fetoprotein methods. [1]

The ancient Ayurvedic system categorizes congenital anomalies under *janmabala pravritta vyadhi* [10]. Acharya Charaka mentioned similar causative factors for *garbha vikritis* (fetal deformity) like: *beeja dusti* (deformed sperm & ovum or chromosomal defects), *atmakarma* (outcomes of previous life/unknown aetiology), *kaala* (adolescence or advanced maternal age), *ashaya dusti* (defects & infections in reproductive system & pathological maternal conditions affecting the uterus), and *matru ahara vihar* (maternal diet, conduct & exposure to teratogens). [10, 11] These etiological

concepts remarkably correspond to several risk factors recognized in contemporary medical science. [4, 6-8]

Ancient medicine also emphasizes the importance of *garbhini paricharya*, a structured antenatal care regimen that includes dietary regimen, lifestyle modifications and emotional regulations for pregnant women. During the first trimester of gestation, plain milk, or with honey or medicated sweetened milk, is recommended, as it provides optimal nourishment, promotes an anabolic effect and prevents dehydration due to nausea and vomiting. In the second trimester, due to the rapid growth of the fetus, it is important to add animal meat and black gram to meet the protein requirement, along with cooked shasti rice, butter and ghee. Foods such as pomegranate, spinach, beetroot, guava and Indian gooseberry are suggested to enhance the mother's haemoglobin levels. Ghee or rice medicated with *Gokshura (Tribulus Terrestris)* is advised in the sixth month. *Gokshura* has diuretic, anti-inflammatory and anthelmintic properties that reduce pedal oedema and urinary infections. During the third trimester, the increasing pressure of the gravid uterus leads to constipation in pregnant women. To alleviate symptoms of gas, constipation and facilitate the downward, outward movement of *vata dosha*, *ksheera yavagu* (milk gruel) processed with ghee, *asthapana basti* (medicated enema), and *anuvasana basti* (oil medicated enema with madhura rasa group of drugs) are recommended. Additionally, using medicated oil *pichu* (vaginal tampons) provides lubrication to the vaginal passage, aids in relaxation during labour and prevents perineal tear. [12, 13] Following the above-

mentioned dietary regimen aims to ensure optimal fetal development, maintain a healthy pregnancy and reduce the risk of congenital anomalies. [12, 13]

Furthermore, Ayurvedic text outlines several *garbhopaghathakara bhavas* (harmful influences on the fetus), which are considered potential risk factors for congenital anomalies. [10, 12] Pregnant women are advised to avoid activities such as strenuous physical exertion, violent acts, excessive squatting, suppression of natural urges (may have an adverse effect on the uterus), oversleeping, exposure to eclipse (harmful environmental radiation), consumption of alcohol, fish with high levels of mercury and excessive pork. [10, 12] Many of these factors are consistent with known teratogens in modern medicine. [8] Notably, Ayurveda also places significant emphasis on mental well-being during pregnancy, advocating the avoidance of negative emotions such as anger, grief and fear. These emotional disturbances can elevate stress and catecholamine levels, which may result in vasoconstriction, leading to fetal hypoxia, intrauterine growth retardation and adverse outcomes. [10, 14 &15]

#### 4. CONCLUSION

Congenital defects can be prevented to some extent by educating women about avoidable risk factors, such as the harmful effects of certain drugs, unhealthy habits, and exposure to environmental and chemical teratogens, which contribute to undue incidents in pregnancy. Women of reproductive age should be informed about the importance of folic acid supplementation during the preconception period. This case highlights the need for early prenatal diagnosis,

preconception education, and adoptable preventive measures to reduce the risk of neural tube defects. Genetic counseling should be offered, particularly for pregnancies that are high-risk or unplanned. Along with this, integrating the Ayurvedic principles such as *garbhini paricharya* (antenatal care regimen) and avoidance of *garbhopaghathakara bhavas* (harmful influences on fetus) with contemporary medical practices can provide a holistic approach to maternal care. Such integration may help to lower the incidence of congenital anomalies and promote healthy pregnancy and offspring.

**Declaration of Patient Consent** – The authors confirm that they have acquired a patient consent form, in which the patient or caregiver has granted permission for the publication of the case, including accompanying images and other clinical details, in the journal. The patient or caregiver acknowledges that their name and initials will not be disclosed, and sincere attempts will be undertaken to safeguard their identity. However, complete anonymity cannot be assured.

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