International Journal of Pharmaceutical Sciences and Drug Analysis

E-ISSN: 2788-9254
P-ISSN: 2788-9246
Impact Factor (RJIF): 6.03
IJPSDA 2025; 5(2): 108-112
www.pharmacyjournal.info
Received: 03-08-2025
Accepted: 07-09-2025

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Understanding anencephaly: A rare congenital disorder

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DOI: https://www.doi.org/10.22271/27889246.2025.v5.i2b.137

Abstract
A significant amount of the brain, skull, and scalp are absent in anencephaly, a severe congenital neural tube defect (NTD). It arises when the anterior neural tube fails to close during the development of the embryo, usually during the first 23 to 26 days of pregnancy. Affected fetuses frequently die soon after birth or are stillborn, making the syndrome consistently deadly. Maternal folate deficiency, poorly managed diabetes, exposure to specific drugs or chemicals, and a family history of NTDs are among the environmental and genetic risk factors linked to anencephaly. Ultrasonography is frequently used to make the diagnosis during pregnancy, and high maternal serum alpha-fetoprotein (AFP) values are used to confirm it Anencephaly has no known cure or effective therapy, despite advancements in prenatal care. Its incidence has been greatly decreased by preventative measures, including taking enough folic acid supplements before getting pregnant and during the first trimester of pregnancy.

Keywords: Neural-tube defect, anencephaly, folate, diagnosis ultrasound, stillborn

Introduction
Anencephaly, a neural tube defect caused by the failure of normal tube closure at the cranial end of the 4-week-old embryo. Globally, it is thought to happen in 1 in 1000 births. When compared to other European nations, Ireland has historically had a comparatively high incidence of neural tube defects. However, from 1980 to 2007, the prevalence of anencephaly in Ireland was 6.41 per 10,000 births, which did not differ significantly from other European registries (Dolk, 2005) [1].

After inborn heart defects, neural tube defects (NTDs) are regarded as the second most devastating congenital malformations of the central nervous system (CNS) and the most prevalent [2]. The primary cause of this abnormality is thought to be non-spontaneous neural tube closure between the third and fourth weeks of intrauterine growth. The majority of these impairments are thought to be caused by a multifactorial inheritance, which is the result of interactions between many genes and environmental variables [3]. According to studies, members of the immediate family are more vulnerable than others; if a child is born with NTD, the likelihood of recurrence in subsequent pregnancies is 25-50 times higher than in cases that do not occur [4, 5, 6]. Neural tube abnormalities are also predisposed by diabetes mellitus, the use of valproic acid to treat epilepsy during pregnancy, obesity, zinc insufficiency hyperthermia, and folate deficiency [7, 8]. NTD affects roughly 1 in 1000 live births on average, while this varies widely by region. The incidence of NTD is normally around 1 in 1000 live births, though it varies significantly in different geographic areas [9]. Pathologically, neural tube defects can range from a tiny, straightforward opening in the vertebrae's posterior canal to the complete neural tube failing to close, which results in craniorachischisis, the most severe kind of abnormality the most common conditions are encephalocele, spina bifida, and anencephaly [10]. The absence of the brain's hemispheres and cranial arch is a deadly congenital deformity known as anencephaly [11]. The most prevalent CNS illness in the West is anencephaly, which affects women 37 times more often than it does males [12]. While neonates with spina bifida and encephalocele need special medical attention and surgery to survive, babies with such deformities typically pass away at birth or soon after [13]. In contrast to Spina bifida (7%) and encephalocele (46%), the prevalence of anencephaly mortality (100%) is substantially greater [14]. As a result, anencephaly is seen as a burdensome public health issue that can result in a substantial loss of human resources

[15].Infants with anencephaly are characterized by a big tongue, bulging eyes, a short neck, and a frog-like look [16]. Other anatomical abnormalities, such as cleft lip, cleft palate, clubfoot, and omphalocele, are connected to anencephaly in approximately 12% of cases [17]. The first congenital abnormality identified by ultrasound was anencephaly, which can be identified between weeks 12 and 13 of pregnancy. Preventive methods include managing known risk factors and providing couples with medical advice regarding pregnancy termination Anencephaly is a complex process influenced by genes and a wide range of other environmental factors, as previous research has shown. According to current research, folic acid supplementation before and during the first three months of pregnancy as well as up to 12 weeks of gestation can significantly prevent anencephaly and lower its prevalence by 50-70% [18]. To lower the incidence of neural tube abnormalities, the National Research Council, the Food and Nutrition Council of the Institute of Medicine, and the U.S. Public Health Service advise all women of reproductive age to take 0.4 mg of folic acid daily [19, 20]. An estimated 300,000 newborns are born with neural tube defects each year, which causes 8.6 million lifetime impairments and 88,000 fatalities [21]. The prevalence of anencephaly varies geographically and throughout time. For example, it was estimated that the prevalence of this abnormality was 12 per 10,000 newborns in northern Iran between 1998 and 2005 [22], but it was 2.81 per 10,000 births in Texas, USA, between 1999 and 2003 [23]. According to data gathered from (EUROCAT) member nations between 2000 and 2010, the prevalence of anencephaly was calculated to be 3.52 per 10,000 newborns [24]. Given that anencephaly is the most severe kind of neural tube defect, its negative impact on the number and quality of patients' and parents' lives, as well as the significant health, psychological, social, and financial costs to both the individual and society, it is crucial to accurately identify patients in order to plan health care services and put preventive measures in place. Furthermore, the current study was carried out using a systematic review and meta-analysis to provide insight into the prevalence, incidence, and mortality of anencephaly globally due to a variety of statistics on the condition's prevalence and the lack of a thorough investigation that can analyze the results of these studies.

Fig 1: Anencephaly

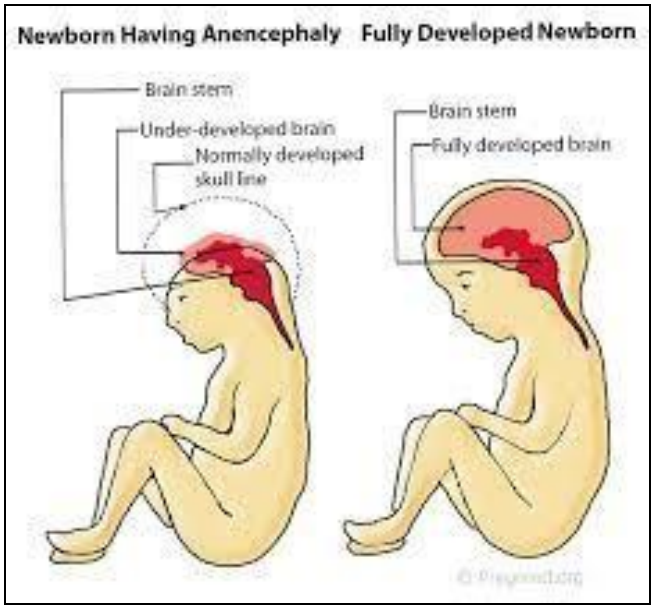

Fig 2: Differ in anencephaly

When the upper portion of the neural tube fails to shut during fetal development, it can result in anencephaly, a kind of neural tube defect (NTD). Although there are various forms of anencephaly, they are typically categorized according to the severity of abnormalities in the brain and skull.

Anencephaly Types

1. **Meroanencephaly:** A form of anencephaly in which there may be some brain tissue present but missing portions of the brain and skull.
2. **Holoanencephaly:** The more severe kind of anencephaly known as holoanencephaly occurs when the complete brain and skull are either missing or significantly undeveloped.

Objective

Prevention

1. **Reducing Risk:** To avoid anencephaly, identify and address risk factors, such as a folate deficit.
2. **Encouraging Healthy Lifestyle:** Supporting healthy practices such as taking supplements of folic acid, eating a balanced diet, and taking care of chronic illnesses.

Early Identification

1. **Prenatal Screening:** To identify anencephaly early in pregnancy, prenatal screening procedures such as ultrasonography and MSAFP are used.
2. **Accurate Diagnosis:** Using fetal imaging and other diagnostic techniques to provide an accurate diagnosis.

Assistance and Attention

1. Providing consolation and assistance to infants born with anencephaly is known as palliative care.
2. **Family Support:** Providing anencephaly-affected families with resources, counseling, and emotional support.

Research and Education

1. **Understanding Causes:** Research is being done to gain a better understanding of the risk factors and causes of anencephaly.

2. **Increasing Public Awareness:** Teaching people about anencephaly, how to prevent it, and how to manage it.

These goals are to lower the prevalence of anencephaly, enhance diagnosis and detection, and assist impacted families.

Symptom

Anencephaly is a severe congenital condition, and symptoms are frequently noticeable at birth or during a prenatal ultrasound. These are a few signs:

Symptoms of pregnancy

1. **Abnormal Ultrasound Findings:** A prenatal ultrasound may reveal a brain and skull that are absent or severely underdeveloped.
2. **Elevated Maternal Serum Alpha-fetoprotein (MSAFP):** Anencephaly or other open neural tube defects may be indicated by elevated MSAFP levels.

Birth-related symptoms

1. **Absent or undeveloped Brain:** The baby's brain may be badly undeveloped at birth, or it may be missing a sizable chunk.
2. **Absent or malformed Skull:** The skull may be malformed or absent entirely or in part.
3. **Facial Abnormalities:** Underdeveloped or deformed facial characteristics are possible.
4. **Breathing and Feeding Issues:** Because of brainstem malfunction, babies with anencephaly may have trouble breathing and feeding.

Prognosis

1. **Poor Prognosis:** Infants with anencephaly usually do not live long after birth, and the condition is frequently deadly.
2. **Palliative Care:** This type of care focuses on giving the baby and family support and comfort.

Families may make educated decisions regarding treatment and get ready for the birth with the aid of early detection and diagnosis.

Stage of occurring

Anencephaly develops during the following stages of embryonic development:

1. The neural tube forms and closes during the neurulation stage, which happens 23-26 days after conception.
2. Neural tube formation: When the upper portion of the neural tube does not close correctly, anencephaly results, which causes the brain and skull to either not develop at all or to develop very poorly.

The Critical Phase Three to four weeks following fertilization is the essential time for neural tube creation and closure. Folic acid supplementation is essential during this period to avoid neural tube problems like as anencephaly.

Diagnosis

Diagnosis of anencephaly can be made by

1. **Prenatal Ultrasound:** By demonstrating the absence or underdevelopment of the brain and skull, a thorough ultrasound examination can identify anencephaly.
2. **Maternal Serum Alpha-fetoprotein (MSAFP) Screening:** Anencephaly or other open neural tube defects may be indicated by elevated MSAFP values.
3. **Prenatal MRI:** A prenatal MRI can assist diagnose anencephaly by providing fine-grained pictures of the unborn brain.
4. **Postnatal Examination:** A physical examination and imaging tests (such as CT scans and X-rays) can confirm the diagnosis after birth.

When the diagnosis was made

Prenatal diagnosis: usually takes place between weeks 16 and 20 of pregnancy, in the second trimester.

Postnatal diagnosis: Takes place after delivery, frequently right after birth.

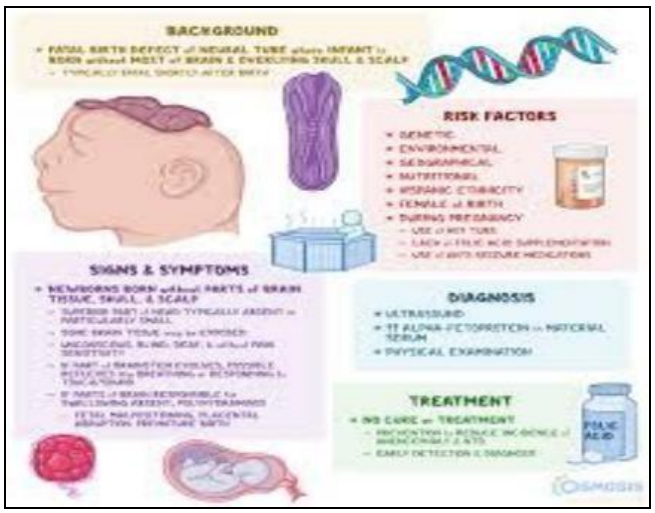

Fig 3: Anencephaly: Overall view

Treatment

Treatment of Anencephaly

Palliative Care: Since anencephaly is frequently deadly, this involves comforting and supporting the newborn after birth.
Prenatal Care: To guarantee the greatest result, the pregnancy is closely monitored and managed.

Assistance for Families: Providing resources, counseling, and emotional support to families impacted by anencephaly.

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