CONDITION

Anencephaly

Learn how anencephaly develops, how it is diagnosed during pregnancy, what care options are available, and how families can prepare and find support.

WHAT IT IS
Severe birth defect of the brain and skull

BODY SYSTEM
Nervous system

MAIN SYMPTOMS
Absent scalp and skull, exposed brain tissue, unusual facial or eye features

WHAT CAUSES IT
Usually unknown, incomplete neural tube closure

HOW IT IS DIAGNOSED
Detailed prenatal ultrasound

MAIN TREATMENT
Comfort-focused palliative care

CAN IT BE CURED?
No, missing structures cannot be rebuilt

IS IT AN EMERGENCY?
Sometimes, with severe vaginal bleeding

LONG-TERM OUTLOOK
Death before birth or shortly after birth

Overview

Anencephaly is a severe birth defect affecting the brain and skull. It happens when the upper part of the neural tube does not close early in pregnancy. The neural tube is the structure that later develops into the brain and spinal cord.

Because the neural tube closes during the first month after conception, anencephaly develops before many people know they are pregnant. Parts of the brain, skull, and scalp do not form. Some brainstem structures may remain, so an affected baby may have a heartbeat, breathe, move, or show reflexes. However, the parts of the brain needed for awareness and long-term survival are profoundly underdeveloped.

Anencephaly is considered life-limiting. Affected pregnancies may end in miscarriage or stillbirth. Babies born alive usually survive for only a short time, commonly hours or days. There

is no treatment that can rebuild the missing brain and skull structures.

A diagnosis can be overwhelming. Parents may face decisions about continuing the pregnancy, ending the pregnancy where that option is available, and planning comfort-focused care. A maternal-fetal medicine specialist, genetic counselor, palliative care team, and the family's usual obstetric clinician can provide information and support without assuming what choice is right for the family.

Symptoms

Anencephaly does not usually cause specific symptoms in the pregnant person early in pregnancy. It is most often found through prenatal screening or ultrasound.

Ultrasound may show that much of the skull and brain has not developed. The appearance can change with gestational age because exposed brain tissue may break down as the pregnancy progresses. An expert ultrasound can help distinguish anencephaly from other problems involving the skull or brain.

Later in pregnancy, some people develop polyhydramnios, meaning there is too much amniotic fluid around the baby. This may occur because the baby cannot swallow and regulate the fluid normally. Polyhydramnios can cause rapid abdominal enlargement, discomfort, shortness of breath, contractions, or early breaking of the waters.

Fetal movement may still be felt because movement and reflexes can arise from the spinal cord and remaining brain structures. Movement does not mean that the missing parts of the brain have developed.

After birth, visible findings may include:

The clinical team can explain what to expect while protecting the baby's comfort and the family's privacy.

- Absence of much of the scalp and skull over the upper head
- Exposed or incompletely developed brain tissue
- Unusual facial or eye features related to the absent skull
- Reflex movements, breathing, or responses to touch

Causes and risk factors

Anencephaly results from incomplete closure of the upper neural tube. This process happens

very early in embryonic development. In most cases, there is no single identifiable cause.

Risk is thought to reflect a combination of genetic susceptibility, nutrition, health conditions, medicines, and environmental factors. Having a risk factor does not mean anencephaly will occur, and many affected pregnancies occur without a known risk factor.

Factors associated with a higher chance of neural tube defects include:

Anencephaly is not caused by ordinary exercise, sexual activity, routine work, or a parent's thoughts or emotions. Most parents could not have known it was developing or prevented it through their actions after learning about the pregnancy.

Folic acid, the supplemental form of folate, lowers the chance of neural tube defects but cannot prevent every case. People who could become pregnant are generally advised to take a daily supplement containing folic acid. Someone with a previous neural tube defect pregnancy may be advised to take a higher amount before conception and during early pregnancy. That plan should be made with a clinician because high-dose supplementation is not appropriate for everyone without medical guidance.

Medicines should not be stopped because of pregnancy concerns without speaking with the prescribing clinician. Suddenly stopping an anti-seizure or other essential medicine can be dangerous. A preconception visit allows time to review safer options when needed.

- A previous pregnancy affected by anencephaly or another neural tube defect
- A personal or family history of neural tube defects
- Not getting enough folate, a B vitamin, around conception
- Diabetes that was present before pregnancy, especially when blood glucose is not well controlled
- Certain anti-seizure medicines, particularly valproate
- High body temperature early in pregnancy, such as from a significant fever or prolonged extreme heat exposure
- Some genetic or chromosome conditions, especially when other fetal abnormalities are present

Diagnosis

Anencephaly can often be diagnosed by prenatal ultrasound. It may be visible during a first-trimester scan, but it is also commonly identified during the detailed anatomy ultrasound performed in the second trimester.

A maternal blood screening test may show a high level of alpha-fetoprotein, or AFP. AFP is a protein made by the developing baby. An open neural tube defect can allow more AFP to enter the amniotic fluid and the pregnant person's blood. A high AFP result is not a diagnosis. Dates that are different from expected, multiple pregnancy, and other conditions can also raise the result.

Cell-free DNA screening, sometimes called noninvasive prenatal testing, primarily evaluates the chance of selected chromosome conditions. It does not replace ultrasound or AFP-based screening for open neural tube defects.

When anencephaly is suspected, the next steps may include:

Amniocentesis is not always necessary when ultrasound findings are clear. Genetic testing may be more useful if additional abnormalities are present or if the diagnosis is uncertain.

Parents should have an opportunity to review the findings before making irreversible decisions. They may ask the specialist to explain which brain and skull structures are absent, how certain the diagnosis is, and whether any other abnormalities were seen.

- A detailed ultrasound with a maternal-fetal medicine specialist
- Review of the pregnancy dates and all fetal structures
- A second expert opinion if findings are uncertain
- Amniocentesis in selected cases to measure substances in amniotic fluid or perform genetic testing
- Genetic counseling to discuss possible causes and the chance of recurrence

Treatment and management

There is no surgery, medicine, or fetal treatment that can correct anencephaly. Care therefore focuses on the pregnant person's health, informed decision-making, and the baby's comfort if the pregnancy continues.

Depending on gestational age, personal values, health needs, and local availability, parents may consider ending the pregnancy or continuing it. The medical team should explain the expected course and available options in a clear, nonjudgmental way.

For a continuing pregnancy, routine prenatal care is still important. Additional visits may be needed if polyhydramnios, contractions, or other complications develop. Severe excess fluid can sometimes be managed by a maternal-fetal medicine team when it causes significant symptoms.

Perinatal palliative care can begin before birth. This is specialized care for a baby with a

life-limiting condition and the baby's family. It does not mean that care stops. It shifts the goal toward comfort, time together, and support that matches the family's wishes.

A written birth plan may address:

Vaginal birth is often possible. Cesarean delivery generally does not change the baby's underlying outcome, but it may be needed for the pregnant person's safety or considered in response to individual goals and circumstances.

If the baby is born alive, the newborn team can provide warmth, skin-to-skin contact, and treatment for signs of discomfort. Invasive resuscitation cannot replace the missing brain structures. Decisions about interventions should be discussed before labor when possible, while recognizing that plans can be revised.

Organ donation is sometimes raised by families, but opportunities are limited and depend on medical, legal, and logistical requirements. A transplant or donation organization can provide case-specific information without promising that donation will be possible.

- The preferred place and timing of delivery when choices are available
- Whether fetal heart-rate monitoring during labor would change care
- When cesarean delivery would or would not be considered
- Comfort measures after birth
- Feeding, holding, bathing, photographs, keepsakes, or spiritual practices
- Which relatives or support people should be present
- Autopsy, placental examination, genetic testing, or donation questions
- Bereavement, lactation, and postpartum support

Complications

For the baby, anencephaly leads to death before birth or a short time after birth. The exact timing cannot always be predicted from ultrasound.

Pregnancy complications may include excess amniotic fluid, preterm contractions, early rupture of membranes, difficulty breathing from abdominal pressure, or an abnormal fetal position. Delivery may not follow the expected pattern because the fetal head does not have its usual shape.

The emotional effects can begin at diagnosis and continue long after the pregnancy ends. Parents and other family members may experience grief, anxiety, sleep problems, depression, or traumatic stress. Partners may grieve differently. Support can include counseling, faith or

cultural care, pregnancy-loss groups, and clinicians trained in perinatal bereavement.

A history of anencephaly also raises the chance of a neural tube defect in a future pregnancy compared with someone without that history. It does not mean that another pregnancy will be affected.

Follow up

During a continuing pregnancy, follow-up is used to monitor the pregnant person's health, discuss developing symptoms, and review the birth and comfort-care plan. Families can request another meeting if their goals change or if new ultrasound findings appear.

Contact the maternity team promptly for possible labor, leaking fluid, fever, worsening shortness of breath, a marked change in fetal movement, or symptoms from rapidly increasing abdominal size. These concerns need timely assessment but do not always represent an emergency.

Seek emergency care for severe vaginal bleeding, fainting, a seizure, severe trouble breathing, chest pain, or severe headache with vision changes. After delivery or a pregnancy-ending procedure, emergency assessment is also appropriate for very heavy bleeding, loss of consciousness, or severe breathing difficulty.

A postpartum visit should address physical recovery, blood pressure when relevant, bleeding, breast milk production, contraception if desired, and emotional health. Parents should seek urgent mental health help if grief is accompanied by thoughts of self-harm, feeling unable to remain safe, or thoughts of harming someone else.

Before another pregnancy, a preconception appointment is recommended. The clinician can review folic acid supplementation, medicines, diabetes management, family history, and the plan for early screening. Genetic counseling may help place recurrence risk in context. No screening strategy can guarantee an unaffected pregnancy, but early ultrasound and appropriate blood screening can provide information sooner.

Sources

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