Anencephaly

Anencephaly is a condition that prevents the normal development of the brain and the bones of the skull. This condition results when a structure called the neural tube fails to close during the first few weeks of embryonic development. The neural tube is a layer of cells that ultimately develops into the brain and spinal cord. Because anencephaly is caused by abnormalities of the neural tube, it is classified as a neural tube defect.

Because the neural tube fails to close properly, the developing brain and spinal cord are exposed to the amniotic fluid that surrounds the fetus in the womb. This exposure causes the nervous system tissue to break down (degenerate). As a result, people with anencephaly are missing large parts of the brain called the cerebrum and cerebellum. These brain regions are necessary for thinking, hearing, vision, emotion, and coordinating movement. The bones of the skull are also missing or incompletely formed.

Because these nervous system abnormalities are so severe, almost all babies with anencephaly die before birth or within a few hours or days after birth.

Frequency

Anencephaly is one of the most common types of neural tube defect, affecting about 1 in 1,000 pregnancies. However, most of these pregnancies end in miscarriage, so the prevalence of this condition in newborns is much lower. An estimated 1 in 10,000 infants in the United States is born with anencephaly.

Causes

Anencephaly is a complex condition that is likely caused by the interaction of multiple genetic and environmental factors. Some of these factors have been identified, but many remain unknown.

Changes in dozens of genes in individuals with anencephaly and in their mothers may influence the risk of developing this type of neural tube defect. The best-studied of these genes is *MTHFR*, which provides instructions for making a protein that is involved in processing the vitamin folate (also called vitamin B9). A shortage (deficiency) of this vitamin is an established risk factor for neural tube defects. Changes in other genes related to folate processing and genes involved in the development of the neural tube have also been studied as potential risk factors for anencephaly. However, none of these genes appears to play a major role in causing the condition.

Researchers have also examined environmental factors that could contribute to the risk of anencephaly. As mentioned above, folate deficiency appears to play a significant role. Studies have shown that women who take supplements containing folic acid (the

synthetic form of folate) before they get pregnant and very early in their pregnancy are significantly less likely to have a baby with a neural tube defect, including anencephaly. Other possible maternal risk factors for anencephaly include diabetes mellitus, obesity, exposure to high heat (such as a fever or use of a hot tub or sauna) in early pregnancy, and the use of certain anti-seizure medications during pregnancy. However, it is unclear how these factors may influence the risk of anencephaly.

Inheritance Pattern

Most cases of anencephaly are sporadic, which means they occur in people with no history of the disorder in their family. A small percentage of cases have been reported to run in families; however, the condition does not have a clear pattern of inheritance. For parents who have had a child with anencephaly, the risk of having another affected child is increased compared with the risk in the general population.

Other Names for This Condition

- anencephalia
- anencephalus
- aprosencephaly
- congenital absence of brain

Diagnosis & Management

Formal Diagnostic Criteria

- Society of Obstetricians and Gynaecologists of Canada: Prenatal screening, diagnosis, and pregnancy management of fetal neural tube defects http://www.jogc.com/article/S1701-2163(15)30444-8/pdf

Genetic Testing Information

- What is genetic testing? /primer/testing/genetictesting
- Genetic Testing Registry: Anencephalus https://www.ncbi.nlm.nih.gov/gtr/conditions/C0002902/
- Genetic Testing Registry: Neural tube defect https://www.ncbi.nlm.nih.gov/gtr/conditions/C3891448/
- Genetic Testing Registry: Neural tube defects, folate-sensitive https://www.ncbi.nlm.nih.gov/gtr/conditions/C1866558/

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov https://clinicaltrials.gov/ct2/results?cond=%22anencephaly%22

Other Diagnosis and Management Resources

- Children's Hospital of Philadelphia
  https://www.chop.edu/conditions-diseases/anencephaly

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Anencephaly
  https://medlineplus.gov/ency/article/001580.htm
- Encyclopedia: Folic Acid in Diet
  https://medlineplus.gov/ency/article/002408.htm
- Health Topic: Neural Tube Defects
  https://medlineplus.gov/neuraltubedefects.html

Genetic and Rare Diseases Information Center

- Anencephaly
  https://rarediseases.info.nih.gov/diseases/5808/anencephaly

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke
  https://www.ninds.nih.gov/Disorders/All-Disorders/Anencephaly-Information-Page
- Office of Dietary Supplements: Folate
  https://ods.od.nih.gov/factsheets/Folate-Consumer/

Educational Resources

- Boston Children's Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/a/anencephaly
- Centers for Disease Control and Prevention: Facts About Anencephaly
  https://www.cdc.gov/ncbddd/birthdefects/Anencephaly.html
- Centers for Disease Control and Prevention: Folic Acid
  https://www.cdc.gov/ncbddd/folicacid/index.html
- Centre for Genetics Education, New South Wales, Australia
  https://www.genetics.edu.au/publications-and-resources/facts-sheets/fact-sheet-60-neural-tube-defects
- MalaCards: anencephaly
  https://www.malacards.org/card/anencephaly

- Merck Manual Consumer Version: Neural Tube Defects and Spina Bifida
  https://www.merckmanuals.com/home/children-s-health-issues/birth-defects-of-the-brain-and-spinal-cord/neural-tube-defects-and-spina-bifida
- Orphanet: Isolated anencephaly/exencephaly
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1048

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/anencephaly/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/spinabif.html
- Share Pregnancy & Infant Loss Support
  http://nationalshare.org/
- The Compassionate Friends
  https://www.compassionatefriends.org/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Anencephaly%5BMAJR%5D%29+AND+%28anencephaly%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ANENCEPHALY
  http://omim.org/entry/206500
- NEURAL TUBE DEFECTS, FOLATE-SENSITIVE
  http://omim.org/entry/601634
- NEURAL TUBE DEFECTS, SUSCEPTIBILITY TO
  http://omim.org/entry/182940

Medical Genetics Database from MedGen

- Anencephalus
  https://www.ncbi.nlm.nih.gov/medgen/8068

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