OPEN ACCESS Freely available online

Cell & Developmental Biology

Opinion Article

Cellular Processes Underlying the Mechanisms of Birth Defects

Zainul Gree Tran*

Department of Cellular Biophysics, Max Planck Institute for Medical Research, Heidelberg, Germany

DESCRIPTION

Birth defects, also known as congenital anomalies or congenital disorders, are structural or functional abnormalities that are present at birth. These defects can affect various parts of the body, including organs, limbs, and systems. Birth defects can range from mild to severe, and their causes are often complex, involving genetic, environmental, or a combination of factors. Understanding the cell biology underlying these birth defects is significant for both diagnosis and potential therapeutic interventions. Advances in genetics and molecular biology have contributed to our knowledge of these conditions and may lead to improved treatments and prevention strategies in the future.

Chromosomal abnormalities

Down syndrome, it is one of the most common genetic disorders, occurring in approximately 1 in every 700 live births. This additional genetic material disrupts normal cell division during early embryonic development, resulting in characteristic physical and cognitive features. Down syndrome is typically caused by a nondisjunction event during cell division, where either the egg or sperm cell fails to separate its chromosomes properly. As a result, the fertilized egg ends up with three copies of chromosome 21 instead of the usual two.

Individuals with Down syndrome often share certain physical features, including almond-shaped eyes, a flat facial profile, a protruding tongue, and a single crease across the palm of the hand (known as a simian crease). They may also have low muscle tone (hypotonia) and a higher risk of certain medical conditions. Down syndrome is associated with intellectual and developmental disabilities of varying degrees. Individuals with Down syndrome may have delays in speech and language development, as well as challenges with fine and gross motor skills. Early intervention and educational support can greatly improve the quality of life and opportunities for individuals with Down syndrome.

People with Down syndrome are at an increased risk for certain medical conditions, including congenital heart defects, respiratory issues, gastrointestinal problems, and thyroid disorders. Regular medical check-ups and monitoring are essential for their well-being.

Neural tube defects

Neural Tube Defects (NTDs) are a group of congenital birth defects that involve abnormalities in the development of the neural tube, which eventually forms the brain and spinal cord in the developing embryo. These defects occur during early embryonic development, often before a woman even knows she is pregnant. NTDs can have serious and lifelong consequences for affected individuals.

Types of neural tube defects:

- Spina bifida, it occurs when the neural tube does not close properly, leading to a gap in the spinal column. There are several subtypes of spina bifida, with varying degrees of severity.
- Anencephaly is a severe NTD in which a major portion of the brain and skull do not develop. Infants with anencephaly are typically stillborn or survive only briefly after birth.
- Encephalocele is a rare NTD where a portion of the brain protrudes through an opening in the skull. The exposed brain tissue is covered by a thin membrane.

The exact causes of NTDs are not fully understood, but they are thought to involve a combination of genetic, environmental, and nutritional factors. Adequate folic acid intake before and during pregnancy is important for preventing NTDs. Women of childbearing age are often advised to take folic acid supplements or consume foods fortified with folic acid to reduce the risk of NTDs. NTDs can sometimes be detected during prenatal screening through procedures like maternal serum screening and ultrasound. A definitive diagnosis may require further testing, such as amniocentesis or chorionic villus sampling.

Metabolic disorders

Phenylketonuria (PKU) is a rare genetic disorder that affects the body's ability to metabolize an amino acid called phenylalanine (Phe). PKU is caused by mutations in the *PAH* gene, which is responsible for producing an enzyme called phenylalanine

Correspondence to: Zainul Gree Tran, Department of Cellular Biophysics, Max Planck Institute for Medical Research, Heidelberg, Germany, E-mail: zgreetran@gbg.de

Received: 17-Oct-2023, Manuscript No. CDB-23-27470; **Editor assigned:** 20-Oct-2023, PreQC No. CDB-23-27470 (PQ); **Reviewed:** 03-Nov-2023, QC No. CDB-23-27470; **Revised:** 10-Nov-2023, Manuscript No. CDB-23-27470 (R);**Published:** 17-Nov-2023, DOI: 10.35248/2168-9296.23.12.309.

Citation: Tran ZG (2023) Cellular Processes Underlying the Mechanisms of Birth Defects. Cell Dev Biol. 12:309.

hydroxylase. This enzyme is necessary to break down phenylalanine into other compounds that the body can use.

PKU is inherited in an autosomal recessive manner, meaning that a child must inherit two mutated copies of the *PAH* gene (one from each parent) to develop the condition. Individuals with one mutated copy are carriers but do not typically exhibit symptoms. In individuals with PKU, phenylalanine cannot be properly metabolized due to the lack of functional phenylalanine hydroxylase. As a result, phenylalanine accumulates in the bloodstream, leading to elevated blood phenylalanine levels, which can be toxic to the brain. High levels of phenylalanine in the blood can lead to intellectual and developmental disabilities if left untreated. This is because phenylalanine interferes with normal brain development, especially during the early years of life.

PKU is typically diagnosed shortly after birth through newborn screening programs in many countries. A small blood sample is collected from a newborn's heel to measure phenylalanine levels. Early detection and intervention are important to prevent intellectual and developmental disabilities. Pregnant women with PKU need to maintain strict dietary control because high phenylalanine levels during pregnancy can harm the developing fetus. Specialized care and monitoring are essential to ensure a healthy pregnancy and fetus. The primary treatment for PKU involves a strict, lifelong low-phenylalanine diet. This diet limits the intake of high-phenylalanine foods, such as certain proteins found in meat, dairy products, and some grains. Special medical foods and formula supplements low in phenylalanine may also be required.

CONCLUSION

Treatment and management of birth defects often involve a multidisciplinary approach, including surgery, medical interventions, therapy, and on-going care. The objective is to improve the quality of life and outcomes for affected individuals, enabling them to reach their full potential. Early prenatal screening and diagnostic tests, as well as genetic counseling, play a significant role in identifying and addressing birth defects. For some conditions, such as neural tube defects and certain genetic disorders, preventive measures like folic acid supplementation have proven effective in reducing the risk of these defects.