

Dr.Violeta Zanaj, Klementina Vogli

Congenital diseases and health care for patients with congenital diseases

Introduction: Congenital anomalies are also known as congenital defects or malformations. They may appear as congenital structural or functional defects that occur during intrauterine life and can be identified before birth, at birth, or later in life. Among the most common anomalies reported by public health structures are: defects affecting the cardiovascular, musculoskeletal, gastrointestinal (including oral cavity), genital and nervous system.

Purpose: It consists in identifying anomalies in pregnancy and caring for them by becoming familiar with the factors that may lead to the creation of these anomalies. To treat these anomalies in pregnancy and continue their treatment later. Preventing them, when possible, by diagnostic tests and analysis from the beginning of pregnancy.

Material and method: 1. To identify the factors that cause these anomalies. 2. Informing the patient about risk factors. 3. Prevention and screening of these anomalies through tests and analysis. 4. Care for these patients on the basis of a descriptive diagnosis. 5. Care and supervision of the emotional side of these patients, the way they experience this period.

Keywords: Congenital diseases, risk factors, prevention tests, nursing care, treatment of anomalies

Congenital diseases

Also referred to as congenital abnormalities, defects, or malformations, are conditions that arise during intrauterine life. These abnormalities may be structural or functional and can be detected before birth, at birth, or later in life. Such defects often result in poor health at birth and are considered significant contributors to death, chronic illness, and varying degrees of disability.

The most common congenital defects include:

- Cardiovascular system defects
- Lip and palate defects
- Down Syndrome (Trisomy 21)
- Neural tube defects, such as spina bifida

Congenital defects are categorized into four main types:

- Genetic factors
- Environmental factors
- Multifactorial factors (a combination of genetic and environmental influences)
- Defects with unknown causes

Risk Factors

Congenital anomalies can occur in any pregnancy, though certain factors increase the risk. Pregnant individuals, or those who could become pregnant, should be aware of these risk factors:

Lack of folic acid: Folic acid is crucial in preventing neural tube defects (NTDs), and it's recommended that pregnant individuals or those planning to conceive take 400 micrograms daily. However, according to the Centers for Disease Control and Prevention (CDC), only 2 out of 5 people of childbearing age follow this recommendation.[1]

Drinking alcohol: Consuming alcohol during pregnancy can result in a variety of issues, including fetal alcohol spectrum disorder (FASD), which is associated with intellectual and developmental disabilities (IDD), physical impairments, and behavioral problems. No level of alcohol consumption during pregnancy is considered safe.[2]

Smoking cigarettes: Smoking during pregnancy can lead to a range of complications, such as lung issues like asthma, and it is strongly linked to congenital anomalies like cleft lip and cleft palate. [3]

Using drugs: Drug use during pregnancy raises the risk of congenital anomalies, including IDD and behavioral problems. It can also increase the risk of pregnancy loss or stillbirth.[4]

Medication use: Some medications can cause congenital anomalies when taken during pregnancy. A notable example is thalidomide, which was once prescribed for morning sickness but caused severe birth defects. Although tighter regulations have been put in place since, many medications used during pregnancy have not been thoroughly tested for safety. Pregnant individuals should consult with their healthcare provider regarding all medications and supplements they take.[5]

Infections: Certain infections during pregnancy increase the risk of congenital anomalies. Examples include cytomegalovirus (CMV), toxoplasmosis (spread through contact with cat feces or raw meat), and Zika virus, which is linked to microcephaly, a condition in which the brain and skull are abnormally small.

Obesity or uncontrolled diabetes: Research indicates that maternal obesity increases the risk of heart defects and NTDs. Obesity is also linked to developmental delays in children. Uncontrolled diabetes, whether pre-existing or gestational, can lead to complications such as large birth weight, breathing issues, and other poor health outcomes.[6]

Environmental exposure: Exposure to harmful substances can also raise the risk of congenital anomalies. Pregnant individuals who are exposed to high levels of radiation (such as through cancer treatments) or certain chemicals may be at higher risk of having a child with congenital defects.[7]

Being aware of these risk factors can help individuals take steps to reduce the chances of congenital anomalies in their pregnancies.

Diagnosis of congenital anomalies

The diagnosis of congenital anomalies depends on the specific condition and the parts or systems of the body affected.

Many structural problems, such as club foot or cleft palate, are diagnosed through a physical examination of the baby immediately after birth. For other conditions, newborn screening or prenatal testing is necessary to detect and diagnose potential issues.

This information primarily focuses on structural congenital anomalies, their causes, prevention, and treatments. Functional and developmental congenital anomalies, on the other hand, are covered in greater detail in topics related to intellectual and developmental disabilities and condition-specific discussions.

✓ **Newborn Screening:**

Newborn screening is a process that tests an infant's blood for various health conditions, including many congenital anomalies. While newborn screening does not provide a

definitive diagnosis, it helps detect potential problems early on. This allows for timely diagnosis and treatment, potentially preventing lifelong complications.

Newborn screening typically includes tests for hearing issues, as well as pulse oximetry, which measures the baby's pulse rate and blood oxygen levels to identify critical congenital heart defects.

Infants at higher risk for specific conditions—such as those with a family history of certain diseases—can undergo additional testing at birth to identify and address these conditions early. For example, newborn screening has been successful in detecting cases of Menkes disease, enabling treatment to begin before significant health problems develop. [8]

✓ **Prenatal Screening**

During pregnancy, individuals undergo routine tests, such as blood and urine tests, to monitor for conditions like diabetes, infections, and pregnancy-related disorders such as preeclampsia. Blood tests also measure the levels of specific substances that can indicate the risk of certain chromosomal disorders and neural tube defects in the fetus. Ultrasound screenings, which use sound waves to create images, allow healthcare providers to observe the developing fetus and detect some congenital anomalies, such as spina bifida.

Healthcare providers often recommend additional prenatal tests for certain pregnant individuals, particularly those over the age of 35 or with a family history of specific conditions. Early prenatal detection helps doctors begin treatment as soon as possible for some congenital anomalies.

✓ **Noninvasive Prenatal Testing (NIPT)**

NIPT is not a routine prenatal test, but it is used when a routine screening indicates that the fetus may have a chromosomal disorder, such as an extra or missing chromosome. This includes conditions like Down syndrome, Patau syndrome, and Edwards syndrome.

NIPT works by analyzing the placental DNA found in the mother's blood, eliminating the need for cell samples from inside the womb.

At present, NIPT is generally recommended for high-risk pregnancies. However, it does not detect open neural tube defects, nor does it predict complications that may arise later in pregnancy. [9]

✓ **Amniocentesis**

Amniocentesis is a test typically performed to check if a fetus has a genetic disorder. During the procedure, a healthcare provider uses a long needle to extract a small amount of amniotic fluid from the womb. This fluid contains cells with genetic material that is identical to that of the fetus. The cells are then cultured in a laboratory, where their genetic material is examined for any abnormalities. Amniocentesis can detect congenital anomalies such as Down syndrome and certain types of muscular dystrophy.

While amniocentesis is a valuable diagnostic tool, there is a small risk of pregnancy loss associated with the procedure. Therefore, it's important for pregnant individuals to discuss the benefits and risks with their healthcare provider before deciding whether to undergo the test. [10]

✓ **Chorionic Villus Sampling (CVS)**

Chorionic Villus Sampling (CVS) is a test that involves extracting cells from the placenta to determine if the fetus has a genetic disorder. A healthcare provider uses a long needle to obtain cells from the chorionic villi, which are tissues in the placenta that provide nourishment to the fetus. The genetic material in these cells is identical to that of the fetus.

Like amniocentesis, CVS is used to test for chromosomal disorders and other genetic issues. CVS can be performed earlier in pregnancy than amniocentesis, but it carries a slightly higher risk of miscarriage. Pregnant individuals considering CVS should discuss the test and its associated risks with their healthcare provider. [11]

Prevention of congenital abnormalities

Not all health conditions that persist from birth can be prevented, but you can reduce your baby's risk by:

- Maintaining a healthy weight
- Avoiding smoking and drug use before pregnancy
- Avoiding alcohol and other drugs during pregnancy
- Taking folate and iodine supplements—it's important to start folate one month before trying to conceive
- Ensuring any medications, you take are safe during pregnancy
- Getting vaccinated against rubella and chickenpox if you're not already immune

If there's a genetic disorder in your family, it's advisable to undergo genetic testing before attempting to conceive. [12]

Treatment

The treatment of congenital disorders depends on the specific condition, its severity, and the affected organs or systems. While some congenital disorders can be managed with medication or surgery, others may require ongoing care and support. Here are some general approaches for treating congenital disorders:

Medical Treatment

- Some congenital conditions can be managed with medication to control symptoms or prevent complications. For example, hormone replacement therapy may be used for conditions like hypothyroidism or growth hormone deficiency.

- For metabolic disorders, specialized diets or enzyme replacement therapies may be prescribed.

Surgery

- Certain structural congenital anomalies, such as heart defects or cleft lip/palate, may require surgery to correct or improve function.
- In some cases, surgery may be necessary to treat congenital malformations that affect vital organs, such as the kidneys, spine (e.g., spina bifida), or digestive system.

Therapies

- For developmental and intellectual disabilities associated with congenital disorders, various therapies can help improve functioning. This may include physical therapy, occupational therapy, speech therapy, and behavioral therapy.
- Early intervention programs are crucial in managing developmental delays, and they help children reach their full potential.

Genetic Counseling

- For genetic disorders, genetic counseling can provide families with information about the condition, inheritance patterns, and the potential for future children to be affected.
- In some cases, prenatal interventions or pre-implantation genetic testing may be options for families planning future pregnancies.

Supportive Care

- Some congenital conditions may not have a cure but can be managed with supportive care, such as pain management, assistive devices, and lifestyle adaptations to improve quality of life.
- For conditions like Down syndrome or cerebral palsy, families often receive a combination of medical support and community resources to help with daily living and social integration.

Monitoring and Ongoing Care

- Children with congenital disorders may need regular monitoring and follow-up appointments with specialists to ensure proper growth and development and to manage any complications that arise over time.

Conclusion

Congenital diseases, though diverse in their types and causes, represent a significant challenge in maternal and child health. Early identification through prenatal screening, genetic counseling, and diagnostic tests plays a crucial role in managing and preventing many of these conditions. While not all congenital disorders can be prevented or cured, early interventions, such as medication, surgery, therapy, and ongoing supportive care, can greatly improve outcomes for affected individuals. A proactive approach—maintaining a healthy lifestyle, seeking regular medical advice, and addressing risk factors before and during pregnancy—remains essential in reducing the risk of congenital anomalies. By fostering awareness and offering timely treatment, healthcare providers can support families in managing congenital disorders and enhancing the quality of life for both parents and children.

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