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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.

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Congenital diseases

Congenital abnormalities 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. As a result of these defects, children are born with poor health, which is claimed to be in many cases important causes of death, chronic illness and disability of varying degrees.

❖ The most common defects are:

- a-Defects of the cardiovascular system
- b- Defects of the lip and palate
- c- Down Syndrome or Trisomy 21
- d-Neural tube defects such as spina bifida

❖ Inborn defects are divided into 4 main categories:

- 1-Genetic factor defects
- 2- Environmental factor defects
- 3-Multifactorial defects (a combination of the above two factors)
- 4-Defects with unknown factor

❖ Risk Risk factors

Although currently about 50% of all congenital abnormalities cannot be associated with a particular cause, there are some known causes or risk factors.

• Socio-economic and demographic factors

Congenital anomalies are more common among families and countries with limited resources, where women often lack access to adequate nutrition, may have increased exposure to agents or factors such as infections and alcohol that promote or increase the incidence of malnutrition. development of prenatal abnormalities. Moreover, advanced maternal age increases the risk of chromosomal abnormalities, including Down syndrome, while young maternal age increases the risk of some other congenital abnormalities.

• **Genetic factors**

Blood linkage (when both parents have blood ties) increases the prevalence of rare congenital abnormalities and nearly doubles the risk of infant deaths, intellectual disability and other abnormalities in the first cousins circle. Certain ethnic communities have a relatively high prevalence of rare genetic mutations, which lead to a higher risk of congenital abnormalities.

• **Infections**

Maternal infections such as syphilis and rubella are a significant cause of congenital abnormalities in low- and middle-income countries.

• **Breastfeeding**

Iodine deficiency, adipose deficiency, obesity and diabetes are all associated with some congenital abnormalities. For example, dysphoric insufficiency increases the risk of having a baby with a neural tube defect. Also, excessive intake of vitamin A can affect the normal development of an embryo or fetus.

• **Environmental factors**

Maternal exposure to certain pesticides and chemicals, as well as certain medications, alcohol, tobacco, psychoactive drugs and radiation during pregnancy can increase the risk of having a baby affected by congenital abnormalities. Work or livelihoods around waste collection or smelting of minerals can also be a risk factor, especially if the mother is exposed to other environmental risk factors or nutritional deficiencies.

❖ **Diagnosis of congenital malformations**

A series of tests and tests should be performed before, during pregnancy and after birth to identify these defects.

• **Pre-pregnancy screening** can be helpful in identifying people at risk for specific illnesses, or at risk of transmitting a disorder to their children.

In this case the control includes:

- family history assessment
- monitoring for fault carriers.

• **Screening for pregnancy**

1. Ultrasound at end of first trimester between 11-13 weeks gestational age. During this examination, a parameter called NT is observed and measured, which relates to the amount of fluid or fluid accumulated behind the baby's neck and is one of the parameters that, if above the norm, indicates an increased risk for fetal defects.
2. Bi-test is a non-invasive test performed at the age of pregnancy between 11-13 weeks. Maternal blood is taken for examination and the level of two proteins is measured which, if above or below the norm, indicate an increased probability of congenital defects.
3. Chorionic villus analysis is performed at the age of 10–12 weeks of gestation, and usually when NT measurements on ultrasound or Bi-test result in high risk for abnormalities. The doctor absorbs a small portion of the placenta and examines it for chromosomal abnormalities or genetic defects.
4. Triple and Quad-tests are performed at the age of 15-20 weeks of gestation. Maternal blood is taken again for examination and 3-4 protein levels are measured. This too is an orientation test and not a diagnostic one.
5. Morphological ultrasound examination at 18-22 weeks gestation age. The fetus is already fully developed and the doctor through a careful examination examines and makes the appropriate measurements, noting any possible abnormalities. Again, with this test, despite experience, it is not possible to diagnose all abnormalities.
6. Amniocentesis is a test performed at the age of 15-18 weeks pregnant. Under the direction of the ultrasound the doctor absorbs the fluid or fluid that surrounds the baby. This fluid is taken for examination for the level of protein that may indicate congenital defects. Also, fetal cells taken in fluid are examined for chromosomal abnormalities such as Down syndrome or genetic disorders such as cystic fibrosis. Amniocentesis is usually executed when there are indications or the above tests have indicated an increased risk for defects.
7. NIPT - is a test that can be done at any time after 10 weeks of gestation age. The mother's blood is taken for analysis, and it detects fetal DNA from

the breakdown of fetal placental cells. This DNA is analyzed for chromosomal abnormalities. The good of this method is that it is non-invasive and has no risk of miscarriage.

8. Tests for Toxoplasmosis, Rubella, Cytomegalovirus infections that are organisms that adversely affect fetal development, often causing defects.

- **Neonatal / postnatal screening** includes clinical examination and screening for blood, metabolism and hormonal diseases. Screening for deafness and heart defects, as well as early detection of congenital abnormalities, can facilitate life-saving treatments and prevent the progression of some of the physical, intellectual, visual or hearing impairments. In some places, babies are routinely screened for abnormalities of the thyroid or adrenal glands before leaving the home.

❖ **Prevention of congenital abnormalities**

A key role in preventing them plays:

- folic acid intake
- iodine vaccination
- Proper care during pregnancy.
- Performing due diligence on time

❖ **Avoiding congenital abnormalities**

1. There are many steps a woman can take to lower her chances of having a child with a birth defect, including staying healthy before deciding to become pregnant. That's because a woman often does not know she is pregnant in the first few weeks, which can be crucial for the health and development of the baby.
2. Other steps you can take throughout your pregnancy include:

3. Stop smoking – Babies born to mothers who smoke tend to be lower birthweight; in addition exposure to secondhand smoke can harm the fetus
4. Eat a healthy diet – Eating a balanced diet before and during pregnancy is not only good for the mother's overall health, but essential for providing the developing fetus with essential nutrients for proper growth and development.
5. Maintain a healthy weight – Women who are overweight may experience medical problems such as high blood pressure and diabetes, and women who are underweight may have babies with low birthweight.
6. Medical management of preexisting conditions – Take control of any current or preexisting medical problems, such as diabetes or high blood pressure.
7. Folic acid – Taking 400 micrograms of folic acid each day can help lower the risk of neural tube defects, or birth defects of the brain and spinal cord. The vitamin is also found in some green leafy vegetables, nuts, beans, citrus fruits and fortified breakfast cereals.
8. Avoid exposure to alcohol and drugs during pregnancy – Be sure to inform your physician of any medications and herbal supplements you are taking, since they can all have adverse effects on the developing fetus.
9. Avoid exposure to harmful substances – These include lead, pesticides and radiation (i.e., X-rays), which may harm the developing fetus.
10. Lower your risk for infection. Pregnant women should avoid eating undercooked meat and raw eggs and avoid all contact and exposure to cat feces and cat litter, which may contain a parasite, *Toxoplasma gondii*, that causes toxoplasmosis. Other sources of infection include insects that have been in contact with cat feces.
11. Take a daily vitamin – Begin taking a prenatal vitamin daily, prescribed by your doctor, to make sure your body gets all the necessary nutrients and vitamins needed to nourish a healthy baby.

12. Dealing with domestic violence – Women who are abused before pregnancy may be at risk for increased abuse during pregnancy. Your doctor can help you find community, social and legal resources to help you deal with domestic violence.

❖ Treatment

Some of the congenital abnormalities can be corrected with surgery. Early treatment can be done for children with functional problems such as thalassemia, congenital hypothyroidism, etc.

❖ Conclusions

The nurse is able to understand the risk factors and the anomaly displayed. Provides proper care to these patients and provides their advice. The nurse advises on consultation and diagnosis through tests and analysis to prevent them.

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