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Review Article

A review of prevalence, risk factors, diagnostic methods, and prevention measures of congenital anomalies

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Abstract

Congenital anomalies are functional abnormalities that occur during pregnancy and are present from birth. The prevalence of congenital anomalies was one in every 44 births. There are many factors responsible for the cause of congenital anomalies environmental, genetic and micronutrient deficiencies. The main cause of congenital anomalies is Less folic acid supplementation during early pregnancy. Mostly 2-4% of newborn children have congenital anomalies due to the deficiency of micronutrients, and due to consanguineous marriage. Several screening and diagnostic tests are available for prenatal diagnosis of congenital anomalies, such as maternal blood sampling, maternal serum screen, foetal echocardiogram, high-resolution ultrasound, chorionic villus sampling and amniocentesis, etc. Even if infants look healthy and there are no signs of health issues, neonatal screening is mandated between the ages of 2 to 7 days. To decrease the prevalence of congenital anomalies primary and secondary preventive measures are taken. The present article discusses the total information about congenital anomalies, prevalence, risk factors, types of anomalies, screening and diagnostic tests, and prevention of congenital anomalies.

Keywords: Birth defects, Congenital anomalies, Prevalence, High-resolution ultrasound, Chorionic villus sampling, Amniocentesis, Neonatal screening, Primary and secondary prevention.

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1. Introduction

Birth defects, congenital disease, and congenital malformation are all terms used to describe congenital malformations. Congenital anomalies are anatomical and functional abnormalities that are present from birth, such as metabolic problems of the body that originate during intrauterine life and can be discovered prenatally, at birth, or later in childhood, such as hearing difficulties phrased.[1]A range of factors associated with birth defects should be reported, including genetics, environmental teratogenic factors, micronutrient deficiencies, and multifactorial inheritance. The factors reported for the congenital anomalies are consanguineous marriage, maternal age, medications, smoking, alcohol consumption, and maternal illness.[2]

Congenital anomalies are caused due to genetic, infectious, nutritional, or environmental factors, that are responsible for the disease About 60-70% of the causes of birth defects are unknown. The congenital anomalies occur mostly in 2-4% of all births. The estimated rate of incidence of congenital anomalies in various systems of the body is 10-50/1000 of new live birth, and the rate of incidence is varying from one country to another country.[3] The maximum rate of congenital malformation is higher in children born with low birth weight and in consanguineous marriages. According to Global Report on Birth Defects annually at least 7.9 million people are born with a congenital anomaly approximately 240,000 newborn children die within the first 28 days due to their birth defects. A further 3.3 million children die between the ages of one month to below five years and 3.2 are physically and mentally disabled for life. Nine out of ten of the congenital anomalies occur in low and middle-income developing nations.[4]

Congenital abnormalities are raising child mortality rates globally; in 2012, child mortality rates in the United Kingdom and Belgium were around 60% higher than those in Sweden for children aged 0 to 14 years old, and there was a

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30% rise in 10 Western European nations compared to Sweden.[5]Birth defects can lead to long-term disability, significantly damaging individuals, families, health-care systems, and societies. In low- and middle-income nations, nine out of ten children are born with a significant birth defect. [6]

This review prioritises practical preventative efforts, highlights regional differences in incidence and healthcare access, and incorporates current diagnostic improvements. By tackling both regional needs and global issues, it offers a multifaceted viewpoint that makes a distinctive contribution to the literature.

1.1 Birth-weight

>2.5 kg	**Normal**
<2.5 kg	low birth weight (LBW)
<1.5kg	very low birth weight (VLBW)
<1kg	extremely low birth weight (ELBW)

Congenital disorders are the major cause of newborn deaths within the perinatal period, which can result in long-term disability with a significant impact on individuals, families, societies and healthcare systems 30% of newborn deaths occur during the perinatal period are majorly caused due to congenital disorders.[7]

Thirty percent of newborn deaths occur during the perinatal period, with congenital disorders being the leading cause. Prenatal diagnosis of congenital disease provides information for decisions during pregnancy and appropriate treatment parentally (timed delivery in tertiary care centre), it is assumed to improve perinatal and long-term outcomes.[8]

1.2 Major symptoms of the congenital anomalies

Congenital defects usually are seen after birth or during the first few months of life. Signs and symptoms are

1. Blue or pale grey lips, tongue and fingernails (cyanosis)
2. Rapid breathing
3. Swelling in the legs, belly and areas around the eyes
4. Childs can have poor weight gain because Shortness of breath during feedings.

Some of the congenital defects may not be diagnosed during childhood. Signs and symptoms of congenital defects in older children.[9]

1. Easily becoming short of breath during exercise or activity
2. Easily tiring during exercise or activity
3. Fainting during exercise or activity
4. Swelling of hands, ankles and feet

1.3 Prevalence of major congenital anomalies

The first cohort study from India found that one in every 44 babies in the cohort had a congenital abnormality and that this rate is approximately equivalent to the prevalence of stillbirths.[11] The data of 1.5 million annual births in 22 countries by the European Surveillance of Congenital Anomalies (EUROCAT) recorded a prevalence of 23.9 major congenital anomalies per 1000 births.[12] The results of the review article in Iran obtained that the total prevalence of congenital anomalies in Iran is 2.3% and 2.5% in Egypt Zagazig University Hospital annual study and its rate is higher in boys than in girl infants.[13] Results of a cross-sectional study in India say that congenital anomalies are more common in women who had more than one pregnancy when compare to first-time pregnancy. The incidence % of congenital anomalies affected for different systems was shown in **Figure 1**.[14]

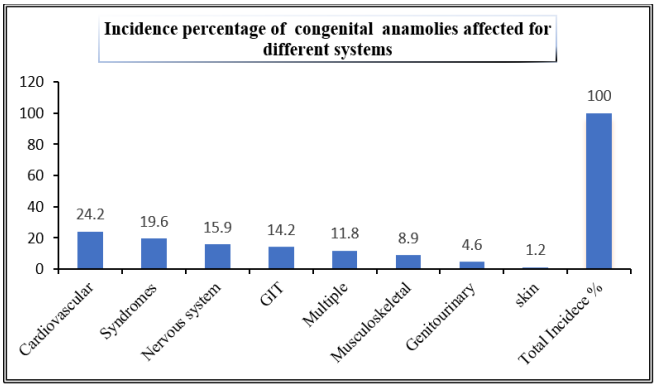

Figure 1: Incidence percentage of congenital anomalies affected for different systems.[14]

1.4 Risk factors

The embryonic membrane in the uterus provides good protection for human embryos, but teratogen exposure during early pregnancy may lead to developmental disturbance. The majority of the risk factors influencing the emergence of congenital abnormalities are unknown.[15]

Genetic mutations or environmental exposure to alcohol or cigarette smoke are the leading causes of congenital anomalies or multifactorial inheritance.[16] Consanguinity is one of the major genetic factors causing congenital anomalies because inbreeding increases the probability of inheriting disease-causing recessive genes from two common ancestors.[17]Environmental factors such as drinking contaminated water (containing heavy metals, nitrates, chlorinated, aromatic solvents, and chlorinated by-products), pesticide exposure, air pollution and industrial pollution sources, living near waste disposal sites, food contamination, and disasters involving large-scale chemical releases that are accidental, negligent, or deliberate.[18] While alcohol and cigarette smoke are teratogenic, herbal preparations and self-medication during pregnancy were linked to a three to fourfold increase in the risk of anomalies. Less folic acid supplementation during the early pregnancy.[19] The majority of birth defects, according to numerous case records, are caused by exogenous factors such as environmental, chemical, or physical factors.[20]

2. Types of Congenital Anomalies

For congenital malformations, deformations, and chromosomal abnormalities, the patterns of congenital anomalies were categorised in accordance with the 10th revision of the International Classification of Diseases and Related Health Problems (ICD-10).[21]

Heart defects, neural tube defects, and Down syndrome have been the most common severe birth defects all these shown in **Table 1**.

Table 1: Type of congenital anomalies and its percentage of occurrence

Type	Percent
Congenital heart defect	24.2
Down's Syndrome	14.2
Multiple congenital anomalies	11.8
Cleft lip/palate	11.1
Spina bifida	7.8
Hydrocephalus	5.3
Club foot	5.1
Hypospadias	2.6
Edward syndrome	2.0
Limb deformity	1.9
Pierre Robin syndrome	1.4
Ichthyosis Vulgaris	1.2
Renal Agenesis	1.2
Anencephaly (abnormal development of skull)	1.0
Polydactyly	1.0
Omphalocele	0.9
Potter syndrome	0.9
Dandy-Walker syndrome	0.7
Microcephaly	0.7
Patau syndrome	0.7
Blader ex-trophy	0.5
Encephalocele	0.3
Undescended Testes	0.3

2.1 External congenital anomalies

2.1.1 Neural tube defects

Anencephaly- Anencephaly is a neural tube defect. Anencephaly is a defect in a baby born without a skull and brain. As the neural tube forms and closes, it helps to form the baby's brain and skull (upper part of the neural tube), spinal cord and back bones (lower part of the neural tube).[22] (**Figure 3**)

2.1.2 Craniorachischisis

Findings keys of craniorachischisis

Head – anencephaly (absence of the brain and cranial vault).

Covering – no skin covering residual brain tissue, spinal cord tissue, or cranial vault (calvarium).

Spine–open (rachischisis) might be limited to the cervical spine, but the open defect can extend to the thoracic spine or even lumbar or sacral spine (craniorachischisis totalis).[23] (**Figure 3**)

2.1.3 Iniencephaly

Iniencephaly is the neural tube malformation from the primary neural anomaly that causes abnormalities with a deficiency in the primary mesoderm. the causes of this anomaly are actually a defect in the occipital bone and rachischisis of the posterior vertebral arches leads to the herbination of neural tissue by the opening of the bone during gestation.[24] (**Figure 3**)

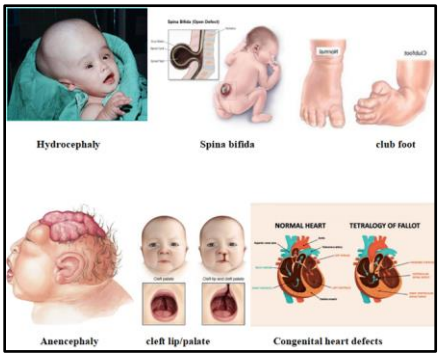

Figure 2: External congenital anomali

2.1.4 Encephalocele

During the third and fourth weeks of pregnancy, the neural tube can be folded and closed can lead to the form of an encephalocele. Which affects the brain and spinal cord. Encephalocele is a sac-like projection of the brain and the membranes that cover it through an opening in the skull. Encephalocele happens during pregnancy when the neural tube does not completely close.[25] (**Figure 3**)

2.1.5 Spina bifida

In womb the baby's spinal cord fails to develop or close properly. Spina bifida can be seen on the skin as symptoms for the spinal defect. They include an abnormal tuft of hair, a protruding spinal cord tissue. Close the defective part of the spinal cord by surgery during the treatment.[26] (**Figure 2**)

2.1.6 Microcephaly

Microcephaly is a birth defect when a baby's head is smaller than compared to normal babies which have similar sex and age. In these babies, the brain is not developed properly. Microcephaly is a without the combination of major birth defects and a combination of other major birth defects.[27] (**Figure 3**)

2.1.7 Microtia/ Anotia

Microtia/ anotia is a congenital birth defect caused by the external ear. The deformity in the external ear or auricle is called microtia. The completely undeveloped pinna is called

an Anotia. In microtia, the right ear is most commonly defective.[28] (**Figure 3**)

2.1.8 Orofacial clefts

Orofacial clefts disorders can result in feeding problems, speech problems, hearing problems and ear infections. Less than half the time the condition is associated with other disorders.

The development of the baby without proper joining of the face during development. It can lead to form a Cleft lip and palate. Cleft lip and cleft palate can diagnose the ultrasound exam during pregnancy.

A cleft lip or palate can be treated successfully with surgery. In 1000 births 1 or 2 members can be affected by this disease.

Three types of Orofacial clefts

1. Cleft lip only
2. Cleft palate only
3. Cleft lip and palate.[29]

2.1.9 Exomphalos (omphalocele)

Exomphalos is a congenital anomaly which can affect abdominal wall of the baby. It can formed when the abdominal wall cannot developed properly in the womb. It is a rare abdominal wall defect. The omphalocele result in the Persistence of intestine or the presence of other abdominal viscera in the umbilical cord.[30]

2.1.10 Gastroschisis

Gastroschisis is a birth defect. The babys intestine extended to outside of abdomen through a hole and next to belly button. The gasroschisis occurs due to immaturity, prematurity and intrauterine growth restrictions. It occurs due to Failure of mesoderm to form in the body wall.[31]

2.1.7 Hypospadias

Hypospadias is an second most abnormality in males, it can occurred in one of 20 members. It is a common in featal stage, the penis and the urethra can not open from the head of the penis. In most cases, the foreskin is less developed and it cannot wrapped completely around the penis. Also, a downward bending of the penis is called as chordee.[32]

2.1.11 Reduction defects of upper and lower limbs

These occurs in the condition of one or more limbs are undersized or missing of some parts during the birth of child. It includes amelia, ectrodactyly and phocomelia among some others.[33,34]

2.1.12 Talipes equinovarus/club foot

It is an congenital anomalies which occurs on foot. But can effect both the leg and foot. Upon 1000 members 1 can effected with these disease. The deformity of foot can persistent and leads to pain and impaired to walk.[35]

2.2 Internal congenital anomalies

2.2.1 Congenital heart defects

Worldwide, congenital heart abnormalities (CHDs) afflict up to 0.6 to 1.9 out of every 100 infants, making them one of the most prevalent birth disorders. CHDs are prevented by taking folic acid supplements throughout pregnancy. Supplementing with folic acid during pregnancy seems to lower the prevalence of CHD by about 20%.[36] (**Figure 2**)

1. **Hypoplastic left heart syndrome:** It is the most challenging form of congenital heart disease to treat the hypoplastic left heart syndrome. It can consume more amount of energy, time and bed capacity for the heart diseases. It has inadequate capacity to perform the function of systemic perfusion to left ventricles.[37]

2. **Common truncus:** It is an effect characterized by the anatomical changes in the heart of the arterial trunk or it can separate the main pulmonary artery and the arota. The arterial trunk can carry blood to different organs and the body.[38] (**Figure 3**)

3. **Interrupted aortic arch:** The interrupted arotic arch is occurs in 3 for million births. It is an very rare disease in this disease the arota of the heart does not completely developed. There is an gap between the descending and ascending thorasic arota.[39]

4. **Transposition of great arteries:** It is a combination of congenital heart diseases involves an abnormal spatial arrangement of superior and inferior venae cavae, pulmonary artery, pulmonary veins and arota. When the defect occurs between the pulmonary artery and arota is called as Transposition of the great vessel.[40]

5. **Pulmonary valve atresia:** Pulmonary artresia is the condition caused due fail of the development of valve orifice of pulmonary valve. In this condition obstruct the blood outflow from heart to lungs. Pulmonary valve is located between the right ventricle and pulmonary artery.[41]

6. **Large intestinal atresia/stenosis:** Bowel obstruction due to structural malformation in the intestine leads to the large intestinal atresia. It occurs in both small and large intestines.[42]

7. **Anorectal atresia/stenosis:** The malformation that occurs between the anus and rectum is called anorectal atresia. It occurs 1 in 1500 members of childbirths.[43]

Time of screening	Methods	Use to check
1) First-trimester screening (11-13 weeks)	A) Maternal blood screen B) Ultrasound	Heart-related birth abnormalities or chromosomal conditions like down syndrome.
2) second-trimester screening (15-20 weeks)	A) Maternal serum screen B) Foetal echocardiogram	To determine whether the mother is more likely than not to give birth to a child who has a particular birth abnormality, neural tube defect, or genetic condition.

3. Screening and Diagnostic Tests for Congenital Anomalies

3.1. Screening tests

A screening test is a method or test used to determine whether a woman or her infant is at risk for certain health issues. These screening tests are generally administered to pregnant women to check for birth abnormalities or other disorders that may affect the woman or her baby. Approximately half of all major structural anomalies, including acrania/anencephaly, abdominal wall defects, holoprosencephaly, and cystic hygromata, can now be detected in the first trimester. Some abnormalities will not become apparent until later in the pregnancy. To this extent, professional societies recommend second-trimester anatomy as the standard investigation for detecting foetal structural anomalies.[44] A screening test does not provide a precise diagnosis that necessitates further investigation. Even when there is nothing wrong with the mother or her infant, a screening test can produce an aberrant result.[45] (**Table 2**

Table 2: Screening methods

3.2. Diagnostic tests

Doctors typically advise extra diagnostic testing to see whether the infant has any abnormalities or other issues if the screening test findings are abnormal. These diagnostic tests might be made available to women who are 35 years of age or older, who have a history of pregnancies that resulted in birth defects, who have chronic illnesses including lupus, high blood pressure, diabetes, or epilepsy, or who use particular medicines.[46]

High-resolution ultrasound.
Chorionic villus sampling.
Amniocentesis.

1. **High-resolution ultrasound:** Ultrasounds produce images of the infant. At around 18 weeks of gestation, all women attending the Antenatal Clinic were screened for fetal well-being via ultrasound examination level I can by a qualified radiologist. Women who had multiple pregnancies were barred from participating in the study. It was possible to use ultrasound to examine internal foetal anatomy and identify abnormalities in the foetal cranium, spine, chest, abdomen, abdominal wall, placenta, cord and limbs. Women who were found to be carrying abnormal fetuses were referred to a senior radiologist for a detailed anomaly scan using high-resolution ultrasound, also known as a level II ultrasound. It is usually completed between weeks 18 and 22 of pregnancy.[47] Maternal obesity, a growing global issue, has been shown to reduce ultrasound examination accuracy and visualization in obese women.[48]

2. **Chorionic villus sampling:** Chorionic villus sampling (CVS) is a procedure performed to biopsy placental tissue between 10 to 13 weeks gestation for prenatal genetic testing. CVS is a safe and efficient method for detecting genetic abnormalities results early in pregnancy, but that it likely has a little higher risk of procedure failure and foetal loss than amniocentesis.[49] The procedure is performed under continuous ultrasound guidance using an aseptic technique. The transcervical or transabdominal route used to perform the procedure is based on provider preference, but the placental location, uterine position, feasibility, operator preference, patient weight, and parity influence the decision.[50] However for the first trimester diagnosis of foetal anomalies, transabdominal and transcervical CVS appear to be equally safe techniques.[51]

3. **Transcervical sampling:** The patient is put in a lithotomy position and a sterile speculum is introduced into the vagina in the transcervical route. An iodine solution is used to clean the cervix. To help the catheter pass through the cervix, a single tooth tenaculum can be put to the anterior lip. A transcervical CVS catheter is introduced into the placenta under continuous ultrasonography supervision. A flexible guidewire with an echogenic tip, which can be seen on ultrasound, is included in the catheter. The stylet is removed once the catheter is in the right position, and a 20cc syringe containing media is inserted into the end of the catheter to create negative pressure. Before the method is completed, the sample is assessed for its suitability. It is also done by using small biopsy forceps.[52]

4. Tissue samples are sent to the lab for culture and additional testing, such as karyotyping fluorescence in situ hybridization, and chromosomal microarray. Rapid evaluation results take 2 to 4 days to arrive, while cultured samples take 1 to 2 weeks. Families should visit with their providers to talk about their treatment options. At 16 weeks, further ultrasounds and MSAFP may be taken to test for open neural tube abnormalities.[53]

5. **Transabdominal approach:** The appropriate spot for exposing the placenta's longest axis is located using the transabdominal technique. Bladder emptying or filling was sometimes required to obtain a more favourable position of the uterus. In a supine position, the patient's abdomen is cleaned with a chlorhexidine or iodine solution. To establish a sterile field, sterile curtains are placed. A local anaesthetic may be given. Under continuous ultrasound monitoring introduces an 18 or 20gauge spinal needle into the placenta. Once the stylet is removed, a 20cc syringe with collection media is affixed to the needle's end. The needle is pushed up and down through the placenta, collecting the tissue under negative pressure. No more than two needle insertions were performed. After the sample has been taken, it is examined to confirm that enough chorionic villi have been aspirated.[54] Transabdominal CVS is more similar to amniocentesis, it is widely suggested that amniocentesis practitioners will find it simpler to learn.[55]

6. **Amniocentesis:** Amniocentesis can be performed for a variety of reasons, including determining whether a baby's lungs are mature enough for birth, down syndrome, draining excess amniotic fluid from the uterus, diagnosing fetal infection, and paternity testing and other conditions. Abdominal amniocentesis can be performed routinely in the 16th week of pregnancy.[56] Women who underwent amniocentesis and CVS had a lower risk of miscarriage earlier to 24 weeks 0.81% and 2.18%, respectively. Than previous studies.[57]

Amniocentesis is usually performed by two operators. There are several options for the procedure, and at least three hands are required: one for the ultrasonography probe, one for the needle, and one to two for the syringe used to draw amniotic fluid. The primary operator typically holds the probe and the needle in each hand. The assistant adjusts the syringe and withdraws the fluid.[58]

First, the health care provider will use ultrasound to pinpoint the baby's precise location within the uterus. The patient should lie back on an exam table, exposing the abdomen. Following that, a gel is applied to the patient's abdomen, and the baby's position is monitored using an ultrasound transducer. Following that, your doctor will use an antiseptic to clean your abdomen. In general, no anesthetic is used. The majority of women report only minor discomfort during the procedure.[59] A thin, 20 or 21gauge disposable hollow spinal needle will be inserted through your abdominal wall and into your uterus, guided by ultrasound. During the 16th week of pregnancy, a large amount of amniotic fluid (up to 20 mL) was aspirated without difficulty. The amount of amniotic fluid withdrawn is determined by how far the pregnancy has progressed. The patient must remain still while the needle is inserted and amniotic fluid is withdrawn. If the first attempt failed to withdraw the amniotic fluid then the new site with new needle were chosen. If it was failed again then rescheduled again in 7-10 days. When the needle enters the skin, the patient may experience a stinging sensation, and when the needle enters the uterus, the patient may experience cramping.[60]

3.3. Neonatal screening

Neonatal screening or new born screening (NBS) began in the 1960s in UK for screening phenylketonuria (PKU). With the development of tandem mass spectrometry (MS/MS), which made it possible to screen for 40–50 conditions with a single blood spot, the panel of screened disorders rapidly increased, receiving a boost in the late 1990s. [61] The ethical considerations and criteria for including a test in a newborn screening program have remained consistent.[62] The term "state newborn screening" refers to a test given to each infant born in each state of the union within the first few days of life in order to identify serious, perhaps fatal disorders. Even if the infant seems healthy and shows no signs of a health issue, state regulations mandate that neonates be tested between the ages of 2 and 7 days. Depending on where you live, different diseases are examined for. PKU, hypothyroidism, galactosemia, sickle cell anaemia (SC), and other haemoglobin abnormalities must all be tested for in the majority of states.[63]

4. Prevention

4.1 Primary prevention

Primary prevention aims to lower the birth prevalence of congenital abnormalities by removing causative causes. Public education regarding pre conceptional and prenatal risks is the foundation of primary preventive efforts. Teratogen information programmes and prenatal screening for foetal malformations are examples of prevention based on reproductive alternatives.[64]

1. **Expansion of rubella immunization:** Rubella is one of the most common causes of vaccine-preventable birth abnormalities. Although rubella virus infection normally results in a moderate febrile rash sickness in children and adults, blindness and deafness, infection during pregnancy, particularly during the first trimester, can result in miscarriage, foetal death, stillbirth, known as congenital rubella syndrome (CRS). WHO has issued recommendations for CRS prevention, and the number of countries implementing rubella vaccination programmes has increased and rubella cases decreased from 670,894 in 2000 to 22,361 cases in 2016.[65]

2. **Advanced maternal age:** From 13-19 to 24-34 may results in the decreased risk of major anomalies. Mothers over the age of 35 who smoked were also linked to congenital anomalies. As a result, it is preferable to reproduce after the age of 19 and before the age of [35.66]

3. **Periconceptional supplementation of folic acid:** A significant public health opportunity is the primary prevention of birth defects with sufficient periconceptional folic acid supplementation, many studies reported that periconceptional usage of folic acid can reduce both mortality and morbidity associated with both birth defects and a number of adult ailments.[67]

4. **Consanguineous marriages:** Recent studies indicate that India's consanguinity rates range from as low as 1% to 4% in the north to as high as 40-50% in the south.[68] Consanguineous couples are more likely than non-consanguineous couples to get married young and have more children at their first pregnancy. They are also more likely to have higher postnatal death rates, congenital abnormalities, and genetic disorders.[69]

   Other preventive measures are using iodized salt, and access to adequate prenatal care, including nutrition, control of maternal infections and avoidance of teratogens.[70]

5. **Secondary prevention:** Secondary prevention was a programme for detecting congenital malformations at birth and treating them as soon as possible. Prenatal diagnosis followed by pregnancy termination is sometimes referred to as prevention, specifically secondary prevention.[71] Only when the aim is to facilitate more effective surgical or other treatment, either in utero or postnatally, can prenatal screening be categorised as secondary prevention. In the developed world, biochemical newborn screening to detect phenylketonuria, congenital hypothyroidism, and other metabolic conditions is a well-established method of secondary prevention of birth defects. However, due to inadequate infrastructure and organisation, a lack of sound cost-effectiveness studies, and competing priorities for scarce funds, its implementation in developing countries has been difficult.[72]

5. Discussion

Congenital anomalies affect 2% to 4% of infants globally, representing a significant public health challenge. These conditions result from a complex interplay of genetic and environmental factors, including consanguinity, exposure to teratogens, and insufficient maternal intake of folic acid. Environmental hazards such as pollution, contaminated water, and poor living conditions further exacerbate their prevalence, particularly in low-resource settings where access to healthcare and preventative measures is limited. Understanding these interconnected factors is essential for developing effective strategies to mitigate their impact.

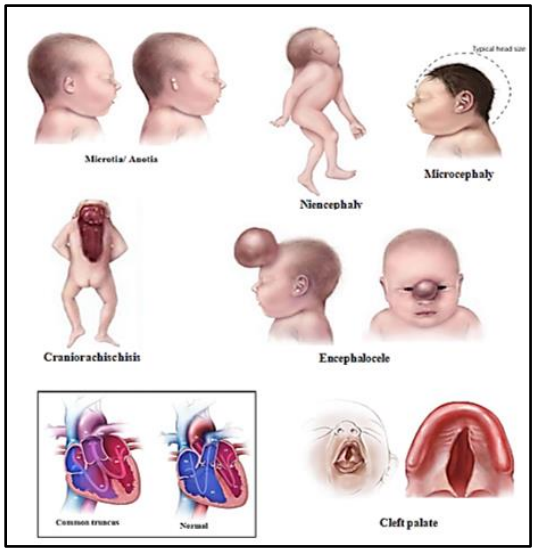

Figure 3: Different types of congenital anomalies[73,74,75]

5.1 The importance of early diagnosis

Early diagnostic methods, including non-invasive prenatal testing (NIPT), amniocentesis, and newborn screening, have transformed the capacity to quickly identify congenital abnormalities. By enabling prompt treatments, these techniques raise survival rates and enhance quality of life. Prenatal imaging, for example, enables medical professionals to schedule any required medical or surgical procedures either before or soon after delivery. However, inequalities in access to these technologies—especially in LMICs—emphasise the pressing need for fair healthcare policy to guarantee that diagnostic tools are more widely available and reasonably priced.

5.2 Preventive actions

In order to lessen the burden of congenital abnormalities, prevention is essential. In high-risk groups, incidence rates can be considerably reduced by addressing risk factors like consanguinity via culturally responsive education and community engagement initiatives. One of the best ways to avoid neural tube abnormalities and other associated disorders is to improve maternal nutrition, especially by making sure the mother consumes enough folic acid both before and throughout pregnancy. It is equally important to address environmental risks, such as lowering exposure to industrial pollutants, infectious agents, and contaminated water. For these preventative measures to be implemented and maintained, cooperation between governments, healthcare professionals, and non-governmental organisations is required.

5.3 Prospects for the future

Significant obstacles still exist in spite of improvements in early identification and prevention. Enhancing surveillance systems to track congenital anomaly prevalence and risk variables might yield vital information for focused therapy. More investigation into the genetic-environmental relationships that underlie these disorders will advance our knowledge and open the door to more individualised treatment and prevention strategies. To close the gap between medical innovations and their real-world applications, efforts should also concentrate on guaranteeing that all socioeconomic groups have fair access to prenatal care and diagnostic technology.

6. Conclusion

Most thirty percent of newborn deaths occur during the prenatal period leading to cause by congenital disorders. During pregnancy take an appropriate treatment parentally to improve perinatal and long-term approvals from congenital anomalies.

Genetic mutations and exposure to alcohol or cigarette smoke and Consanguinity are one of the major genetic factors causing congenital anomalies Environmental factors such as drinking contaminated water and pesticide exposure, air pollution and industrial pollution sources, living near waste disposal sites, food contamination.

Different diagnostic methods are used to determine the presence of any abnormality in children are neonatal screening, transcervical sampling, amniocentesis and transabdominal approach. Mainly approaches to Neonatal screening because it can use to determine blood-related disorders.

Mainly congenital anomalies are mainly due to Consanguineous marriages are responsible for birth defects and diseases. These are prevented by decreasing consanguineous marriages, increasing awareness of pregnancy, and knowing the requirements are mainly useful for baby growth and development.

To provide the medication, improving the infant's prospects for a typical life. It is crucial to identify these disorders to avoid mortality, intellectual incapacity, and other disabilities as soon as possible.

6.1 Data sources

The following indexed databases and grey literature sources were surveyed during the preparation of this manuscript

6.2 Surveyed indexed databases

1. PubMed/Medline, Scopus/Elsevier, Web of Science, Springer Link, Cochrane Library.
2. Regional/National Indexed Journals.
3. MedlinePlus, Google Scholar

6.3 Surveyed grey literature

1. Hospital/Clinical Resources and Online Open-Access Databases.
2. Reputable Institution Websites (e.g., WHO, Boston Children's Hospital).
3. Government/Public Health Portals (e.g., CDC, National Health Portal, India).
4. Specialized Databases (e.g., OMIM for genetic disorders).
5. Medical Textbooks and Literature (e.g., Human Embryology & Teratology, Texas Heart Institute Journal, New England Journal of Medicine).
6. Health Portals (e.g., Stat Pearls, Mayo Clinic, Stanford Children's Health).
7. Archived Resources from CDC, NINDS, and Medical Reports (e.g., Morbidity and Mortality Weekly Report).
8. CDC Health Portals on birth defects (e.g., Neural Tube Defects, Craniorachischisis, Microcephaly)

6.4 Data availability statement

No new data were generated or analysed in this review. All data used are publicly available from the cited sources.

7. Conflict of Interest

No conflict of interest related to this manuscript.

8. Source of Funding

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