Volume : 2 | Issue : 9 | September 2013 • ISSN No 2277 - 8179

Research Paper

Folic Acid & Vitamin B12 Deficiency: Commonest Cause of Congenital Anomalies amongst Sub-Urban Population in Pune

Medical science

KEYWORDS : Folate & B12 Deficiency , Congenital anomalies

Dr. Mithil Patil | Assistant Professor, Bharati Vidyapeeth University Medical College , Pune.

ABSTRACT

We have studied the incidence & type of congenital anomalies detectable at birth in Bharati Hospital & research centre over a period of 7 years by retrospective analysis. Bharati Hospital & Research centre is a tertiary hospital having a annual birth rate 1800 . Bharati hospital caters to approximately 5 million population in the vicinity along with innumerous patients referred for specialized care from villages, primary health centers & Subcentres in a perimeter of 100 kms . Most of the population served is lower middle class. After studying their socioeconomic factors we have found that most of the pregnant patients with congenital anomalies are pure vegans & have co-existent anaemia during pregnancy . NTDs are the commonest congenital anomaly in 75 % of patients suggesting deficiency of folate & Vitamin B12.

Introduction

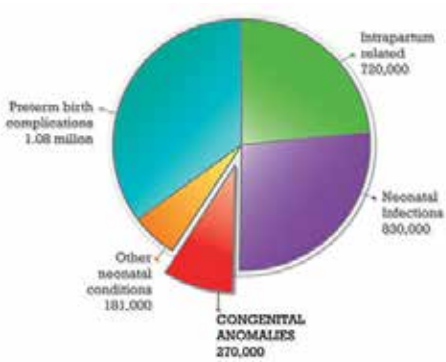
Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth.

Congenital anomalies and preterm birth are important causes of childhood death, chronic illness, and disability in many countries. In 2010 the World Health Assembly adopted a resolution calling all Member States to promote primary prevention and the health of children with congenital anomalies by:

- developing and strengthening registration and surveillance systems;
- developing expertise and building capacity;
- strengthening research and studies on aetiology, diagnosis and prevention;
- promoting international cooperation.

Congenital malformation represents defects in morphogenesis during early fetal life. According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth**.[1]** The leading causes of infant morbidity and mortality in poorer countries are malnutrition and infections, whereas in developed countries they are cancer, accidents and congenital malformations **(figure1).**

Figure 1 : Causes of Early Neonatal death. (WHO Data) (Source: Adapted from WHO. Born too soon. The global action report on preterm birth. Geneva, World Health Organization, 2012



Congenital anomalies account for 8–15% of perinatal deaths and 13–16% of neonatal deaths in India**.[2,3]** Patients with multiple congenital anomalies present a relatively infrequent but tremendously difficult challenge to the pediatrician. The proportion of perinatal deaths due to congenital malformations is increasing as a result of reduction of mortality due to other causes owing to the improvement in perinatal and neonatal care. In the coming decades, this is going to be a leading cause of morbidity and mortality in centers providing good neonatal care.

We have studied incidence of congenital anomalies detectable at birth in Bharati Hospital & research centre over a period of 7 years by retrospective analysis. Bharati Hospital & Research centre is a tertiary hospital having a annual birth rate 1800 . Bharati hospital caters to approximately 5 million population in the vicinity along with innumerous patients referred for specialized care from villages, primary health centers & Subcentres in a perimeter of 100 kms . Most of the population served is lower middle class. .

Aims & Objectives :

To study the incidence & type of congenital anomalies in the fetus & to seek causes of it.

Material & Methods : We have maintained labour register along with Birth Defect Registry Form at the labour room. BDR form has to be filled by the attending obstetrician & pediatrician after thorough neonatal examination at birth. (Doc 1)

BDR form has got all the relevant information such as detailed obstetric history of the patient along with history of exposure of teratogens . Type of anomaly in detail has to be written on the form along with any further investigations if required. Every baby delivered & found to have anomalies further followed up till 1 year of age. Most of the anomalies are diagnosed during 2nd trimester of pregnancy by ultrasonography . Those anomalies which are lethal ies. anencephaly are terminated in the second trimester of pregnancy after due consent of the patient . Minor anomalies such as Cleft lip/ palate , babies after delivery are thoroughly examined by pediatrician to rule out other anomalies.

Observations ;

Neural tube defects has been found to be the commonest anomaly in fetus & newborn delivered at Bharati Hospital & Research Centre .We also have studied all the data pertaining to second trimester termination of pregnancies i/v/o structural anomalies found on routine sonography. We have studied all birth from March 2007 till march 2013 by carefully auditing the BDR Forms along with the labour notes of the attending pediatrician. Over 6 years of duration we have 5490 deliveries at our hospital out of which 149 babies were found to have congenital anomalies.

Type Of Anomaly

Hydrocephalus , anencephaly & spina bifida were found to be the commonest anomalies **(Table 1)** . 29 of the total anomalies were having anencephaly & spina bifida.

Table 1 . Types Of Congenital Anomaly Observed

Sr.No	Anomaly	Number Of Patients
1	Anencephaly	17
2	Spina Bifida	12
3	Hydrpcephalus	27

4	Renal Anomalies	21
5	CVS defects	16
6	Hydrops Fetalis	10
7	Diphragmatic Hernia	11
8	Cleft lip / Cleft palate	06
9	CTEV	03

Hydrocephalus was found in 27 fetus on ultrasonograhy in the second trimester of pregnancy . In majority of cases hydrocephalus is found to be associated with Arnold Chiari malformation & neural tube defect Renal anomalies were found in 21 babies & most of them were polycystic kidney disorder . CVS anomalies were seen in 16 babies **(Figure 2).**

Percentage of lethal anomalies which are incompatible to life was found to be much more than certain minor anomalies egs. CTEV , Cleft lip/palate etc.

Socio-economic factors
We have studied detailed dietary history & socio-economic factors of every patient found to have congenital anomalies in the fetus. Almost 75 % of patients were pure vegans which predisposes them to vitamin B12 deficiency , a common cause of NTDs. Most of the patient belong to the poor socioeconomic group where malnourishment & lack of proper diet is quite common. We have also found coexisting anemia in 80 % of patients having congenital anomalies in the fetus . Anemia has dimorphic picture suggesting deficiency of both folic acid & iron. Consanguinity was rarely found in the study group while there was no history of exposure to known teratogens .

Average age of diagnosis of anomalies in the fetus is 17 weeks of pregnancy which is usually found on Ultrasonography. Such patients were further counseled regarding the type of anomaly & its outcome.

Discussion :
Congenital anomalies (also referred as birth defects) affect approximately 1 in 33 infants and result in approximately 3.2 million birth defect-related disabilities every year.

An estimated 2, 70 000 newborns die during the first 28 days of life every year from congenital anomalies. Congenital anomalies may result in long-term disability, which may have significant impacts on individuals, families, health-care systems and societies.

Congenital anomalies may have a genetic, infectious or environmental origin; although in most of the cases it is difficult identify their cause. About 110 000 cases of babies born with congenital rubella syndrome can be prevented through timely vaccination of the mothers during childhood and the reproductive years. Many birth anomalies can be prevented and treated. An adequate intake of folic acid, iodine, vaccination, and adequate antenatal care is the key. Approximately 50% of all congenital anomalies, however, cannot be assigned to a specific cause. However some causes or risk factors have been associated to congenital anomalies. Although it may be an indirect determinant, congenital anomalies are more frequent among resource constrained families and countries.

It is estimated that about 94% of serious birth defects occur in middle- and low-income countries, where mothers are more susceptible to macronutrient and micronutrient malnutrition and may have increased exposure to any agent or factor that induces or increases the incidence of abnormal prenatal development, particularly infection and alcohol. Advanced maternal age also increases the risk of some chromosomal abnormalities including Down syndrome. Consanguinity (relationship by blood) increases the prevalence of rare genetic congenital anomalies and nearly doubles the risk for neonatal and childhood death, intellectual disability and serious birth anomalies in first cousin unions. Some ethnic communities, e.g. Ashkenazi Jews or Finns, have comparatively high prevalence of rare genetic mutations, leading to a higher risk of congenital anomalies. Infections such as TORCHES are rarely found to be a cause of congenital anomalies in our study . Maternal exposure to pesticides, medicinal and recreational drugs, alcohol, tobacco, certain chemicals, high doses of vitamin A during the early pregnancy, and high doses of radiation increase the risk of having a baby with congenital anomalies. Working or living near or in waste sites, smelters, or mines may also be a risk factor. The annual report of Indian Council of Medical Research[4] says that the commonest congenital malformations are cardiac in nature (0.57%). Mathur *et al*.[5] reported that the musculoskeletal abnormalities were the commonest. Kalra *et al*.[7] reported that the CNS defects have the highest incidence, whereas Sugunabi *et al*.[6] reported gastrointestinal malformations to rank the highest.

Conclusion:
More than 80 % of pregnant patients having fetal anomalies have coexistent anemia. Neural tube defect is the commonest structural defect observed in the study population. Hydrocephalus , anencephaly & spinal bifida were the commonest types of structural defects observed in the study population. Dietary deficiency of vitamin B12 might be the cause of rise of NTDs as 75 % of the population studied was pure vegan. Consanguinity was rarely observed in the couples having structural defects in their babies. There was no history of exposure to known teratogen during the pregnancy leading to congenital anomalies in the fetus.

Patient awareness programs regarding dietary modification & pre conceptional supplementation of folic acid along with vitamin B12 are needed. Eradication of anemia by all means is important especially in pre-marital women as early child bearing, frequent child births , faulty & deficient diet predispose these women to B12 & folic acid deficiency leading to congenital anomalies in the fetus.

So our study points to the dietary deficiency of Vitamin B12 & Folic acid as a single most important cause of most of the congenital anomalies in the fetus in Pune sub-urban population.

Ethical approval : Not required as the study is epidemiological observation study.

Acknowledgement: None
Conflict of interest : None
Funding : None

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