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Prevalence of Congenital Anomalous Fetus Born in a Tertiary Care Hospital at Rajasthan

Vineeta Paliwal[1], B. S. Jodha[2]

Abstract: ***Background: Congenital anomaly is commonly expressed as absence/ excess/ malformation of body parts due to faulty development of the embryo which may be inherited genetically/ acquired during gestation/ inflicted during parturition. Aims and Objectives: To know the prevalence of congenital anomalies and also to know the prevalence of individual congenital anomaly in the geographical area. Material and Method: This study is hospital based, short term and cross-sectional descriptive study, carried out in the Department of Obstetrics and Gynecology, Ummaid Hospital, attached to Dr. S. N. Medical College, Jodhpur. Result: Over a period of one year total numbers of deliveries were 21,983 and total 213 malformed babies were delivered having an incidence of congenital anomalous babies 0.97%.Maximum no. of congenital anomaly detected at birth was congenital heart disease (12.68%), followed by cleft lip and palate (11.27%) and hydrocephalus (10.33%). In minor anomalies club foot constituted maximum no. of cases (5.63%) followed by hypospadias (4.69%). Conclusion: Congenital malformations are emerging as a major cause of neonatal mortality and morbidity in developed as well as developing countries. Outcome of infant born with congenital anomalies varies with the area, socioeconomic status, education, type of anomaly, gestational age, birth weight etc.***

Keywords: Congenital anomalies, type of anomaly, musculoskeletal, cardiovascular

1. Introduction

Congenital anomalies are defined as a structural and functional disorder (e.g. metabolic disorders), also known as congenital disorders, birth defects or congenital malformation that happens due to a known or unknown cause, occurs in the intrauterine life, identified and diagnosed during prenatal period or at birth, infancy, or may be detected later in life as per WHO guidelines[(1)] such as hearing defects etc.

Congenital anomaly is commonly expressed as absence/ excess/ malformation of body parts due to faulty development of the embryo which may be inherited genetically/ acquired during gestation/ inflicted during parturition.

Congenital anomaly that occurs physically are those anomaly that shows an abnormality of the structure of a body part which may or may not be look over as a problem in day to day life. If examined carefully many persons have one or more of the minor physical anomalies such as preauricular pits (tiny indentations of the skin near the ears), a third nipple, clinodactyly (curvatures of the 5th finger) and short 4th metacarpal or metatarsal bones.[(2)] Some of these minor anomalies are more significant as they provide a clue to the internal abnormalities associated with syndrome.

National estimates of congenital malformation are important for determining the service planning, monitoring trends and assessing the disease burden so that action can be taken at a definitive time. In 2010, the World Health Assembly adopted a "resolution on birth defects to promote primary prevention" by detecting pregnancies having higher chances of congenital anomalous babies and to improve the health of children with congenital anomalies.

Congenital anomalies are one of the leading cause of childhood death, premature births, need for induction of labor, chronic illness and disability in many countries. Congenital anomalies can lead to long-term disability, which causes significant impacts on life style of individuals, families, health-care systems, and societies.[(3)]Excessive expenditures of hospitalization and treatment protocols for these children impose a large excess burden on health care system and on their families or on society.[(4)]Some congenital anomalies can be preventable by the use of preventive measures such as vaccination, adequate intake of folic acid or iodine through fortification of staple foods or supplementation, adequate intake of vitamin B 12 and adequate antenatal care. Although approximately 50% of all congenital anomalies cannot be linked to a specific cause, there are some known genetic, environmental and other causes or risk factors which are responsible for such type of cases also those cases which does not present at birth but they present later in life such as deafness.

Incidence of congenital anomaly varies from country to country worldwide, which may be credited to racial factors, environmental factors, geographical distribution, consanguineous marriages, differences in survey methods and many others factors.

Table 1: Incidence of congenital anomaly in various countries[(5)]

Country	Incidence of congenital anomaly
Japan	1.07%
Taiwan	4.3%
England	2%
India	1.1%

Incidence of Western Rajasthan is 0.43% [(6)].
Incidence varies with multiple pregnancy and chorionicity, about 1.7 times higher in twin pregnancies than singleton pregnancies and is about 1.8% higher in monochorionic twins than dichorionic twins.[(7)]

There are two types of congenital anomaly
1) Minor anomaly (Incidence is 14%)
2) Major anomaly (Incidence is 2%)[(8)]

There are not identifiable specific causes, while some causes and risk factors are associated. These are –

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1) Socioeconomic and demographic factors-94% of severe congenital anomalies occur in middle to low income countries, because in these countries women often lack access to nutritious food and have increased chances of infection and increase intake of alcohol that induce or increase the chances of mutation so the incidence of abnormal prenatal development increases. Advanced age of either of the parents increases the risk of chromosomal abnormalities, for example Down syndrome in cases of advanced maternal age and Schizophrenia in advanced paternal age. Similarly teenage pregnancies also increase the risk of some congenital anomalies e.g. omphalocoele, gastroschisis etc.

2) Genetic factors- Consanguinity increases the probability ofchances of pregnancy with rare genetic congenital anomalies which are having either autosomal or X linked recessive trait and doubles the risk of neonatal and childhood mortality, intellectual disability due to cerebral involvement and also other anomalies in first-cousin unions so consanguinity may be avoided to decrease the chances of congenital anomalies.

3) Infection-In low and middle income countries, syphilis and rubella are a significant cause of congenital anomaly such as congenital rubella syndrome in fetus of pregnant women suffering from rubella in first trimester of pregnancy.

4) Maternal nutritional status- Lack of access to proper maternal nutrition causes iodine deficiency, folate insufficiency, obesity and diabetes mellitus which are linked to some congenital anomalies and associated with various congenital debilitating anomalies. Risks of neural tube defect are more in cases with folate deficiency. The normal development of an embryo or fetus is also affected by excessive intake of vitamin A.[9]

5) Environmental factors- Certain pesticide, chemicals, tobacco, certain medication, alcohol, radiation, psychoactive drugs etc. during pregnancy increases the risk of congenital malformations. Environmental factors such as mines, smelters or working or living near waste sites may also probably increases the chances of congenital anomalies e. g. pulmonary valve stenosis by organoposphorus exposure.[9]

The detection of congenital anomalies is a tedious task but can be achieved by preconception screening, peri-conception screening, neonatal screening.

2. Material and Methods

This study is hospital based, short term and cross-sectional descriptive study, carried out in the Department of Obstetrics and Gynecology, Ummaid Hospital, attached to Dr. S. N. Medical College, Jodhpur. All cases whether diagnosed or undiagnosed before delivery where included, those who gave informed consent.Data were collected from labour wards of Ummaid Hospital attached to Dr. S N Medical College.All patients admitted to labour room who gave the informed consent were included in this one year study.

Exclusion Criteria

All those cases that delivered congenital anomalous baby outside Ummaid Hospital and referred to this hospital for further work up were excluded.

Investigations included were random blood sugar, hemoglobin, platelet count, coagulation profile, blood grouping and typing, viral markers, thyroid profile, ultrasound for fetal well being to detect congenital anomaly.

Protocol

A detailed epidemiological, socioeconomic status, educational status was taken into account as history of patient. After informed consent detailed physical examination, systemic and per vaginal examination was done. Routine and case specific investigations were done.

After admission patient was under follow up study for outcome to be studied.

3. Observation

Over a period of one year total numbers of deliveries were 21,983 and total 213 malformed babies were delivered.

Table 2: Incidence of congenital anomaly

Total deliveries	Total congenital anomalous babies	Incidence of congenital anomalous babies
21,983	213	0.97%

The incidence of congenital anomalous babies was 0.97%.

Table 3: Systemic distribution of cases of congenital anomaly

S. No.	System involved	No. of cases	No. of minor anomaly	No. of major anomaly
1	Nervous System	55	1	54
2	GIT	42	0	42
3	Skeletal	27	21	6
4	Syndrome	26	0	26
5	CVS	25	1	24
6	Respiratory	18	0	18
7	Genital	10	9	1
8	Urinary	4	0	4
9	Eye	2	2	0
10	Ear	2	2	0
11	Other	2	0	2

The above table shows that out of total 213 delivered malformed babies nervous system accounted for the majority of cases i.e. 55 cases ; followed by digestive system accounting for 42 no. of cases. Nervous system included maximum no. of major anomalies followed by digestive system. While skeletal system accounted for maximum no. of cases having minor anomaly i.e. 21 cases followed by genital system.

Table 4: Number of cases of individual congenital anomaly

S. No.	Anomaly	No. of cases	Percentage
1.	Congenital heart disease	27	12.68
2.	Cleft lip and palate	24	11.27
3.	Hydrocephalus	22	10.33
4.	Meningomyelocele	17	07.98
5.	Syndromes	15	07.04
6.	Annencephaly	14	06.57

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7.	Tracheoesophageal fistula	12	05.63
8.	Club foot	12	05.63
9.	Hypospadias	10	04.69
10.	Others	60	28.18
Total		213	100

Maximum no. of congenital anomaly detected at birth were congenital heart disease (12.68%) followed by cleft lip and palate (11.27%) and hydrocephalus (10.33%). In minor anomalies club foot constituted maximum no. of cases (05.63%) followed by hypospadias (04.69%).

4. Discussion

Congenital malformations, rapidly emerging cause of neonatal mortality, is one of the major worldwide problem associated with pregnancy, having significant impacts on individuals, families, health-care systems, and societies.

In our study the total number of deliveries during the study period was 21,983, out of which 213 were malformed. The incidence of congenital anomalous babies was 0.97% which nearly matches the prevalence of other state. In 2010 in Andhra Pradesh M. Sandhya rani et. al[(10)]have found that 0.9% babies were having any form of congenital anomalies. This study was concordant with our study. Similar incidence was seen in study done by Akruti et al[(11)]in 2009 at Bhavnagar, Gujarat that is 0.88% of the total deliveries. The incidence found in our study is less as compared to the national incidence which is 1.1% and other states have concordant or higher incidence of congenital anomaly as in Table 5.

Table 5: Incidence of congenital anomaly in various states of India

State	Incidence of congenital anomaly
Ahemdabad	1.2%
Jammu	1.5%
Varanasi	1.2%
Central Maharashtra	1.9%

This variability in incidence of malformation in various parts of India is due to geographical, environmental, ethnic factors, use of drugs during pregnancy, diet and undetected anomalies at birth which present later in the life, difference in autopsy rates and different observation period. The present study has excluded aborted malformed fetus of < 20 weeks gestation and malformed babies who delivered elsewhere and referred to our hospital. These might be one of the reason for the low incidence in our study when compared to other states.

In our study majority of cases of anomalies were of involvement of nervous system (25.82%) followed by involvement of digestive system and musculoskeletal system comprising 19.72% and 12.68% cases respectively. Similar results had been seen in studies done by various others in other region like Patel et al[(12)] and Naik et al [(13)]have shown that 33.33% of cases were having malformations of nervous system. The nervous system anomalies occur due to defective closure of neural tube between the 23rd to26th day of gestation or by the use of drugs or deficient diet having deficiency of folic acid and vitamin B12.Most of the other major anomalies were also followed by digestive system, while most of the minor anomalies were of musculoskeletal system followed by genital system.

Basavanthappa et al has reported that the most common congenital malformation that affects the region in South India was musculoskeletal malformations accounted for 27.5% of cases of all malformations, followed by cutaneous 19.16%, genitourinary 15.83%, gastrointestinal 12.5%, neurological 10%, and cardiac malformations 5.83%.[(14)] Vatankhah et al reported that the musculoskeletal anomalies (27.5%) was the most common cause of malformations in the region. [(15)] Other studies have also similar results concordant with us.[(16, 17, 18, 19, 20, 21)]

Pal et al. in West Bengal studied that congenital malformations involving the cardiovascular, musculoskeletal, and genitourinary system were found to be most commonly involved systems that can be affected by various etiologies known till date. [(22)] Some studies have shown that higher incidence of CNS malformations followed by GIT and musculoskeletal system was found which causes more chances of still birth, death after delivery of fetus etc[(23, 24, 25)] whereas Suguna Bai *et al*[(26)]has evaluated that GI malformations are the most common cause of congenital malformation.

On studying the individual congenital anomaly maximum cases detected were of congenital heart disease (12.68%) followed by cleft lip & palate (11.27%) and hydrocephalus (10.33%). On considering minor anomalies maximum cases were of club foot (5.63%) followed by hypospadias(4.69%).

Despite the high prevalence of congenital malformations or anomalies, there are no awareness program, educational programs about maternal care during pregnancyon congenital malformations and the consequences of increased maternal age and parity which also increases the chances of congenital malformations.

5. Result

Over a period of one year total numbers of deliveries were 21,983 and total 213 malformed babies were delivered having an incidence of congenital anomalous babies 0.97%.Maximum no. of congenital anomaly detected at birth was congenital heart disease (12.68%), followed by cleft lip and palate (11.27%) and hydrocephalus (10.33%). In minor anomalies club foot constituted maximum no. of cases (5.63%) followed by hypospadias (4.69%).

6. Conclusion

Congenital malformations are evolving as an important cause of neonatal mortality and morbidity in developed as well as developing countries. Outcome of infant born with congenital anomalies varies with the type of anomaly, gestational age of baby at time of delivery, birth weight etc. This study helps to know the pattern of congenital anomalies in our geographical area and help in calculating the prevalence in our country. Treatment and rehabilitation of these anomalous children is a challenging task as these anomalies required to be corrected for healthy life.

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Thereforeregular antenatal visits and prenatal diagnosis may be very helpful in early trimester so that prevention, timely intervention and even planned termination can be made. Neurological defect as commonest malformation can easily be prevented by pre-conceptional folic acid and vit-B12 treatment.

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