

Original Research :

Congenital Anomalies in Neonates – A Study at Medical College Hospital in Himachal Pradesh

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Abstract:

The Purpose of study was to find out the overall incidence of clinically detectable congenital anomalies in newborns in hospital deliveries.

All the newborns delivered at Kamla Nehru Hospital, Shimla were examined for congenital malformations over a period of one year. Five thousand nine hundred and ninety seven newborn babies of consecutive deliveries were examined at birth for the presence of congenital malformations. The overall incidence of malformations was 1.63%. Musculoskeletal anomalies were commonly found followed by gastrointestinal and cardiovascular anomalies. The present study was carried out with the aim to determine the overall rate of congenital malformations, incidence in live births, as well as incidence affecting various organ systems, at a medical college hospital in Himachal Pradesh.

Keywords: *congenital anomalies, musculoskeletal anomaly, major anomaly.*

Introduction:

In a developing country like India due to high incidence of infectious diseases, nutritional disorders and social stress, the developmental defects are often overshadowed, but the present scenario is changing rapidly. Congenital anomalies represent defective morphogenesis during early

foetal life. A broader definition includes metabolic or microscopic defects at a cellular level. A recent study shows that congenital anomalies contribute to 9% of perinatal deaths as compared to 8% a decade ago. About 2% newborn infants have major anomalies diagnosed at or soon after birth⁽¹⁾. Congenital anomalies account for 8 to 15%^(2, 3) of perinatal deaths and 13 to 16% death in India^(4,5). Congenital anomalies can be due to abnormality being either result of genetic constituent or antenatal environment ,plus all condition, known to be caused by specific genes at whatever age they become manifest and whether or not they are associated with a demonstrable abnormality of form^(6,7).

Major anomaly is defined as an anatomic abnormality severe enough to reduce normal life expectancy or compromise normal function e.g. heart defect, spinal defect, intestinal defect⁽⁸⁾. Major anomalies have serious medical, surgical and cosmetic consequences.

Minor anomaly is a physical feature often familial that is present in only a small proportion (1 to 5%) of normal individuals e.g. simian crease of palm ,epicanthal folds⁽⁸⁾.

Incidence and geographical distribution of congenital anomalies:

The worldwide incidence of congenital disorders is estimated at 3-7% but actual number varies between countries. Population and hospital

based studies from different parts of India show that 2.5% of newborns have birth defects. Even here pattern of malformation varies from region to region e.g. Neural tube defects are common in northern India as compared to musculoskeletal defects are more common in rest of India⁽⁹⁾.

Aims and Objectives:

- 1) To study the overall incidence of clinically detectable congenital anomalies in newborns in hospital deliveries.
- 2) To classify the congenital anomalies in major and minor groups.

Material and Methods:

It's a prospective observational type of study. Study population consisted of five thousand nine hundred ninety seven babies delivered at the Department of Obstetrics and Gynaecology, Kamla Nehru hospital, Shimla who were examined at birth for the presence of congenital malformations. All the newborns were looked for major and/or minor congenital malformations and everyday during routine ward rounds. Relevant information regarding maternal age, gestational age, sex, community, birth weight, birth order and consanguinity was documented. Significant antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications during labour was recorded. Antenatal ultrasonography (USG) findings were noted. Relevant radiological, histo-hematological tests were carried out. Baby's gestational age, birth weight, sex and symptoms in postnatal period were noted. The detailed general and systemic examinations of the babies were carried out. Thorough physical examinations of newborn babies were done. All macroscopically anatomical defects were recorded in a predesigned Performa. A meticulous general and systemic examination was carried out by a consultant at the time of birth to detect any malformations. Ultrasound was employed routinely to detect multiple congenital anomalies and to rule out majority of the internal congenital anomalies. 2D echocardiography was also used for

all congenital heart diseases, along with the routine X-ray chest and electrocardiogram. Those babies who were born with any external malformations were subjected to relevant investigations to rule out internal anomalies. Malformations were categorized in major and minor defects. The major malformations were divided into central nervous system (CNS), musculoskeletal, gastrointestinal, genitourinary, cardiovascular system (CVS), syndromes and miscellaneous disorders.

Observations:

During period of one year, total 5997 deliveries were conducted, 5867 were live births and 130 were stillbirths. The number of babies with congenital malformations diagnosed at birth or within the first week of life was 77, while the total number of malformations was 96 (1.63%).

Table I and II give the sex distribution and incidence of congenital malformations. The sex wise distribution was 39 males and 38 females, giving an M: F ratio of 1.02:1, while *p* value was 0.6062 which was not statistically significant.

TABLE -I

Total number of deliveries	5997
Total number of vaginal deliveries	4873
Total live birth	5867
Total still birth	130
Total live male	3121
Total live female	2746
Total number of congenital malformed babies	77
Total malformations	96
No of malformed male babies	39
No of malformed female babies	38
Incidence of anomalies	1.63%

Table II - Sexual Variation In Incidence Of Anomalies

Sex of baby	No of live births	No of Malformed babies	Percentage
Male	3121	39	1.24
Female	2746	38	1.38
Total	5867	77	1.31

Incidence of malformations in general was found to be apparently more in female (1.38%) than in male (1.24%).

Pattern Of Congenital Anomalies :Musculoskeletal system was the most common system involved accounting for 23.84% of total congenital anomalies followed by decreasing frequency as cited in Table III. Of the 96 anomalies, there were 83 major anomalies and 13 minor. In the major anomalies Musculoskeletal anomalies were commonly found followed by gastrointestinal and cardiovascular anomalies. In the minor anomalies polydactyly was the most common followed by syndactyly and low set ears.

Table III (A)- System wise distribution of congenital anomalies

System	No of anomalies	Percentage	Incidence/1000 live births
Musculoskeletal	23	23.84	3.92
GIT	21	21.85	3.57
C.V.S	16	16.65	2.72
Face	12	12.40	2.04
C.N.S	10	10.40	1.70
Genitourinary	07	7.18	1.09
Eye	03	3.12	0.51
Skin	02	2.08	0.34
Respiratory	02	2.08	0.34
Total	96	100	16.3

TABLE III(B): System wise distribution of individual congenital anomalies

Anomalies	Number	Percentage
1. Musculoskeletal		
CTEV	14	14.58
Polydactyly	4	4.16
Syndactyly	3	3.12
Limb defects	2	2.08
2. Gastrointestinal System		
TEF	7	7.29
Duodenal Atresia	4	4.16
Exomphalos	3	3.12
Oesophageal atresia	3	3.12
Duodenal hernia	2	2.08
Imperforate anus	2	2.08
3. Cardiovascular System		
PDA	6	1.02
ASD	4	0.68
VSD	3	0.51
Pulmonary atresia	1	0.17
Tricuspid atresia	1	0.17
TGA	1	0.17
4. Central Nervous System		
Meningomyelocele	5	0.85
Anencephaly	1	0.17
Hydrocephalus	3	0.51
Encephalocele	1	0.17
5. Face		
Cleft lip	5	5.20
Cleft palate	3	3.12
Low set ears	3	3.12
Hypertelorism	1	1.04

Anomalies	Number	Percentage
6. Genitourinary system		
Hypospadiasis	4	4.16
Epispadiasis	2	2.08
Renal agenesis	1	1.04
7. Respiratory system		
Choanal atresia	1	1.04
Laryngeal atresia	1	1.04
8. Eye		
Congenital bupthalmos	1	1.04
Congenital cataract	1	1.04
Enophthalmos	1	1.04
9. Skin		
Skin tags	1	1.04
Haemangioma	1	1.04
Total	96	100

Higher frequency of congenital anomalies was seen in the babies born with low birth weight however P value not being significant as shown in Table IV.

TABLE IV-Relation of congenital anomalies with birth weight.

Birth weight (grams)	Congenital anomalies present	Congenital anomalies absent	Total	P value
<1500	9	677	686	0.992
1500-2500	40	2999	3039	0.978
>2500	28	2114	2142	0.933
Total	77	5790	5867	

There were higher number of congenital anomalies in babies born to mothers in the age group 20-30 yrs of age (74%) and next in order were > 30 years accounting for (18.2%), least number of congenital anomalies were in the babies born to age less than 20 years. P value was not significant. Incidence of congenital anomalies was higher in multigravida as compared to primigravida. The incidence increased as the parity increased as shown in Table V. P value was significant (<0.0.5) and in multigravida (>Gravida 4)

TABLE V: Relation of congenital anomalies with maternal parity

Parity	Congenital anomalies Present	Congenital anomalies absent	Total	P Value
Primigravida	33	3193	3226	
Multigravida				
G2	21	1351	1372	0.49
G3	10	839	849	0.86
G4	9	352	361	0.09
>G4	4	55	59	0.014*
Total	77	5790	5867	

Discussion:

Congenital anomalies are important causes of still births and infant mortality, and are contributors to childhood morbidity. The number of birth defects in infants is increasing antenatally and during the neonatal period due to advanced diagnostic technology, especially USG. The incidence in the present study is lower than as reported by studies quoted in table below, this difference may be due to the fact because in the present study only live newborns have been taken and still born have been excluded. Other factors can be different culture, geographical conditions, inaccurate detection at birth, period of observation, autopsy rate as in certain centers autopsies have been performed and this leads to higher incidence in some studies. Other factors which can contribute to the difference in the incidence like genetic factors, geographical area of settlement, socioeconomic status, maternal nutrition and habits, prenatal health care services and large number of environmental factors which could not be measured.

Incidence of congenital anomalies in various studies:

Authors	Type of study	Total births	Incidence
P.chaudhary et al 10 (1989)	Prospective	3000	27.2
Swain et al 11 (1994)	Prospective	3932	12
V.Dutta et al 12 (2000)		2869	12.4
Neelu desai et al13 (2006)	Prospective	2188	32.0
Arjun Set al14 (2005)	Prospective	9308	15.0
Present study	Prospective	5867	16.3

Association of low birth weight with increased risk of congenital malformations was very well documented¹⁵, our study was in accordance with this. The incidence of congenital anomalies was significantly higher in term babies as compared to Pre term babies. Male preponderance amongst congenital malformed babies was found in this study which was statistically insignificant.

In present study no consanguinity was recorded. Previous studies have¹⁵ reported that significantly higher incidence of malformation among the mothers of gravida 4 or more and our results are consistent with this finding. This indicates that as the birth order increases, the incidence of congenital anomalies also increases. Certain maternal diseases may occasionally lead to increased risk of birth defects. According to Ordonez et al., 16 diabetes mellitus, arterial hypertension, and hypothyroidism show a positive association with congenital malformation. In our study Antenatal history of mothers were suggestive that 45.5% were anemic, 13% had hypertension and 3% had gestational diabetes mellitus.

With regard to pattern of congenital malformations in present study the system involved in descending frequency were musculoskeletal (23%), followed by gastrointestinal system (21%) and cardiovascular (16%), face(12.45%), CNS (10%) and genitourinary system (7.2%). In the present study congenital anomalies involving musculoskeletal system were found in 23.7% with an incidence of 3.92 per 1000 live births, talipes was the most common anomaly.

As musculoskeletal anomalies anomalies were most common in our study reason being that these anomalies are externally visible hence were easily picked up. Internal organs (gastrointestinal system, cardiovascular system) anomalies were less detected because of invisible nature of the systems and also neonates have been asymptomatic in particular during first 24 hours of life. Other reasons are lack of follow up.

With regard to the cardiovascular system, patent ductus arteriosus was the most common lesion followed by atrial septal defect and last being ventricular septal defect. Among the genitourinary tract anomalies, hypospadias, was the most prevalent lesion. Regarding the central nervous system, the most prevalent anomaly encountered was meningomyelocele seen in 5.2% cases and congenital hydrocephalus found in 3.12% cases. With special reference to the neural tube defect (NTD), the incidence of NTD has markedly reduced in the developed countries following mass promotion and mandatory prescription of folic acid for pregnant mothers.¹⁷⁻²⁰

Incidence of facial anomalies was 2.04 per 1000 live births, percentage being 12.4% among other congenital anomalies. Cleft lip most common followed by cleft palate and low set ears. Incidence of cleft lip and palate in present study was 1.21 per 1000 live births. V. Dutta¹²(2000) recorded cleft palate as the most common anomaly but it has been cited in gastrointestinal tract anomalies.

Conclusion:

Congenital anomalies are a major cause of stillbirths and infant mortality. By thorough clinical examination, the life-threatening congenital malformation must be identified, as early diagnosis and surgical correction of the malformed babies offer the best chance for survival.

Conflict of Interest - None

Source of Funding - Nil

Contribution of authors-Vijay Yadav:

Conducted the study under supervision,

Rakesh Sharma: Chief supervisor and guide,

Jyotsna Sharma: Assisted the study,

Pancham Kumar: Co guide,

Deepak Sharma: Biostatistics and manuscript.

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