ORIGINAL ARTICLE

Pattern of Gross Congenital Anomalies in The Biggest Maternity Hospital of Pakistan

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ABSTRACT

This study was a descriptive case series on "Pattern of gross congenital anomalies in the biggest maternity hospital of Pakistan." The objective was to identify the pattern of gross congenital anomalies in newborns at the tertiary care hospital and to enlist the preventable predisposing risk factors leading to such anomalies. The study period was three years with effect from 01-07-2007 to 01-07-2010. It was conducted in Lady Willingdon hospital Lahore. A proforma was devised to collect data

There were thirty five thousand six hundred and six (35604) deliveries in our study period and six hundred (600) newborns had various gross congenital anomalies. Mean age of the parturient was 25.21±4.63years. 63 % were multi gravid whereas 37 % were primi gravid. Mean gestational age was 30.20±5.95 weeks. Neural tube defects were the commonest congenital anomalies either alone or in combination with other anomalies; accounting for a total 63.0% (378 newborns). It was found that 1out of 59.39 newborns had gross congenital anomalies making the prevalence of 16.85 per 1000 births. The commonest associated risk factor was cousin marriage.

Congenital anomalies are a distressing condition not only for the parents but also for health care providers. Public awareness about such hazards of cousin marriage can reduce the prevalence. Neural tube defects can be reduced by pre conception folic acid & multivitamin therapy. An early prenatal diagnosis and termination of pregnancy can help in alleviating psychological distress related to late diagnosis and consequently late termination of pregnancy.

It was concluded that the neural tube defects were the most common congenital anomalies and the most common associated risk factor was cousin marriage.

Key Words: Spina bifida anencephaly folic acid neural tube defects.

INTRODUCTION

A congenital anomaly is a medical condition that is present at birth. It can be recognized before birth, at birth, or many years later. These disorders can be the result of genetic abnormalities, the intrauterine environment, or unknown factors. Congenital disorders range from minor physical anomalies (e.g., a birthmark) to severe malformations of a single system (e.g., congenital heart disease or amelia of the legs) or combinations of abnormalities affecting several parts of the body.[1] The incidence of significant congenital malformations is about 2.5% at birth and major fetal abnormalities account for 20% of perinatal deaths and many survivors are physically and mentally handicapped [2]. The problem therefore places a heavy responsibility on the health care providers.

Teratogenic exposure such as drug ingestion, maternal illness or infection may cause craniospinal and/or facial malformations at any time of embryogenesis. Maternal exposure to tobacco smoke and radiation during pregnancy, maternal conditions such as diabetes mellitus and infectious disorders including herpes, syphilis, rubella and cytomegalovirus are major associated risks [3]. Significantly higher prevalence is observed among off springs of first cousin couples compared to unrelated ones.

Purpose of the study was to know the pattern & prevalence of congenital anomalies and the factors associated with their causation. It was anticipated that the results of this study will help in prevention and management of these anomalies.

AIMS AND OBJECTIVES

To know the pattern & prevalence of gross congenital anomalies in newborns at tertiary care hospital and to identify preventable predisposing risk factors.

MATERIAL AND METHODS

STUDY DESIGN: Descriptive case series.

SETTING: Department of Obstetrics & Gynaecology, Lady Willingdon Hospital Lahore.
DURATION OF STUDY: Three years with effect from 01-07-2007 to 01-07-2010.
SAMPLE SIZE: 600 newborns having gross congenital anomalies out of 35604 total deliveries.
SAMPLING TECHNIQUE: Non probability: convenience sampling.
SAMPLE SELECTION: Inclusion Criteria
All pregnant women with a gestation of 16 weeks or more delivered during the study period in Lady Willingdon Hospital Lahore.
EXCLUSION CRITERIA: Mutilated & disfigured babies in which gross features could not be recognized.
DATA COLLECTION: An informed consent was obtained from pregnant mothers to use their data in research. Any risk involved was discussed. Detailed history of each parturient such as age, parity, previous congenital anomalies, family history of congenital anomalies, cousin marriage, drug intake, any medical disorder was taken. Clinical examination and relevant investigations of mother were done. After birth, detailed gross examination of baby was done. All this information regarding predisposing factors and presence of gross congenital anomalies was collected through specially designed proforma.
DATA ANALYSIS: collected data was entered and analyzed on spss version -11. The variables to be analyzed were age, parity, previous congenital anomalies, family history of congenital anomalies, cousin marriage, history of drug intake, and any medical disorder. They were analyzed using simple descriptive statistics, as this is a descriptive study so no test of significance was applied.

RESULTS

Out of 35604 deliveries in the 3 year study period 600 newborns had various gross congenital anomalies. It was found that 1out of 59.39 newborns had gross congenital anomalies making the prevalence of 16.85 per 1000 births.

Mean age of the parturient was 25.21±4.63years (Table-1). 63 % (n=378) were multi gravid whereas 37 % (n=222) were primi gravid. (Table-11) Mean gestational age was 30.20±5.95 weeks (Table-111).

78.0% parturient (n=486) had history of cousin marriage. Congenital anomalies were present in families of 13 % parturient (n=78) while 13% parturient (n=78) had history of congenital anomalies in one or more existing children and 3 % patients (n=18) had history of previous still births (Table-1V).

In this study 79 % patients (n=474) were delivered by spontaneous vaginal delivery (SVD) and 21% patients (n=126) delivered through abdominal route either cesarean section or hystrotomy.

61% patients had polyhydramnios and.16 %patient had oligohydramnios with intrauterine growth restriction. 38% patients had fetal malpresentation. 22.0% had diabetes mellitus. 1.0% patients had history of drug intake

Table-1: Distribution of cases by age n=600

Age (years)	Number	Percentage
<20	54	09.0
20-25	288	48.0
26-30	174	29.0
31-35	72	12.0
≥36	12	02.0
Total	600	100.0
Mean±SD	25.21±4.63	

Table-2: Distribution of cases by parity n=600

Parity	Number	Percentage
Primigravida	222	37.0
Multigravida	378	63.0
Total	600	100.0

Table-3: Distribution of cases by gestational age n=600

Gestational age (weeks)	Number	Percentage
< 20	72	06.0
20-25	96	16.0
26-30	180	30.0
31-35	224	28.0
36-40	120	20.0
Total	600	100.0
Mean±	30.20±5.95	

Table-4: Distribution of cases by history n = 600

History	Number	Percentage
H/o Congenital abnormalities in one or more of the existing child	78	13.0
H/o congenital abnormalities in family of either parent.	102	17.0
H/o previous still birth	18	03.0
H/o cousin marriage	468	78.0

Table-5: Distribution of cases by gross congenital abnormalities n = 600

Gross congenital anomalies	Number	Percentage	Overall percentage
Neural tube defects			63.0 %
Hydrocephalous	114	19.0	
Anencephaly + Acrania	126	21.0	
Spina bifida + Meningocele+ Encephalocele	126	21.0	
Microcephaly, Micrognathia	12	02.0	
Skeleton			21.0%
Acrania	12	02.0	
Cleft lip	30	05.0	
Cleft palate	30	05.0	
Talipes equinovarous	30	05.0	
Polydactyly	18	03.0	
limb hypoplasia	06	01.0	
Thorax			5.0%
,Hydrothorax	24	04.0	
Diaphragmatic hernia	06	01.0	
Abdomen			5.0%
Gastroschisis	12	02.0	
Omphalocele	18	03.0	
Genito-Urinary			6.0%
Hypospadiasis	06	01.0	
Ambiguous Genitalia	12	02.0	
Renal agenesis	12	01.0	
Multi/polycystic disease	12	01.0	
Posterior Urethral Valve Abnormality	12	01.0	

Table-V depicts the pattern of gross congenital anomalies. Neural tube defects were the commonest congenital anomalies either alone or in combination with other anomalies and accounted for 63.0% of total congenital anomalies. Acrania was present in 2 %babies (n=12), cleft lip in 5% babies (n=30), and cleft palate in 5% babies (n=30), microcephaly and micrognathia were present in 2 %cases. Talipes equinovarous was present in 5% babies (n=30), polydactyly in 3% babies (n=18) while limb hypoplasia was present in 1% baby (n=6) Hydrothorax was present in 4% cases while diaphragmatic hernia was present only in 1.0%newborns Omphalocele was present in 3% babies (n=18) and gastroschiasis in 2 % babies (n=12). Abnormalities of genitourinary showed hypospadiasis in 1% baby (n=6), ambiguous genitalia in 2% babies (n=12), renal agenesis in 1% babies (n=6), multi/polycystic disease in 1% baby (n=6) and posterior urethral valve abnormality in 1% babies (n=6)

DISCUSSION

Congenital anomalies are one of the major causes of perinatal death. Many survivors are physically & mentally handicapped and they face a low self esteem. The care takers have to face multiple management challenges and the parents have to undergo a continuous psychological stress. The problem therefore leads to a financial& social burden.

In my study it was found that 1out of 59.39 newborns had gross congenital anomalies making the prevalence of 16.85 per 1000 births. This incidence of congenital anomalies is based only on ultrasound findings & gross examination. However same prevalence was found in a study conducted by Dastgiri et al in Iran [4].

Teenage pregnancy increases the risk of congenital anomalies [5] however it could not be documented in our study as only 9% of the anomalous babies were delivered by patients less than 20 years of age. Increased maternal age is associated with high incidence of congenital malformations especially chromosomal abnormalities. Certain environmental factors and diabetes mellitus increase the risk of congenital anomalies. My study shows that 22.0% of anomalous newborns were delivered by diabetic mothers [6] Congenital abnormalities are also more common (68%) in multigrivida. than in primigravid patients.

Many drugs including alcohol, phenytoin, folic acid antagonists, streptomycin, tetracycline, warfarin, thalidomide are teratogenic [7] however the association of drug intake and radiation exposure was not significant in my study (Table 7). The most important predisposing factor associated with congenital anomalies is history of consanguineous marriage [8]. My study has also documented that 78% of anomalous babies were born by mothers with history of cousin marriage. 61 % of marriages in Pakistan are between first and second cousins. [9]. Family history of congenital abnormalities is other important risk factor. In my study 13% of anomalous babies were delivered by patients with previous history of anomalous babies [10].

So how can we prevent and manage the problem of congenital anomalies. First approach should be prevention to reduce the prevalence. Next step should be early diagnosis and early termination of pregnancy. Early termination of pregnancy will decrease the psychological stress on the parents and will also reduce financial burden on the society. Counseling of couples after the anomalous baby is born is very important to prevent recurrence in the subsequent pregnancy...Neural tube defects are the commonest anomalies which can be simply prevented by folic acid supplementation in preconception period & during pregnancy.[11]

An early prenatal diagnosis (PND) can offer selective termination of pregnancy. PND can be made on ultrasonography, biochemical tests, chorionic villious sampling, amniocentesis and FISH (fluorescent in-situ hybridization) technique. Ultrasound alone has its limitations and leads to late diagnosis. The same has been revealed in my study which shows that 36% of anomalies were diagnosed between 20-30 weeks of gestation.

CONCLUSION

It was concluded that the neural tube defects were the most common congenital anomalies and the most common associated risk factor was cousin marriage. The prevalence of anomalies was16.85 per 1000 births.

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