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Patterns and Prevalance of Congenital Malformations

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ABSTRACT

The aim of the study was to study the patterns and prevalence of congenital malformations. A hospital based cross sectional study was conducted on patients for a period of one year June 2015 to June 2016 taking into account all the deliveries that occurred in our institution. Total number of deliveries for the period was 17890.all the cases booked and unbooked were taken into consideration. Some cases have been diagnosed incidentally in our institution. The most common malformation encountered in our hospital were CNS defects because they are picked up more by antenatal USG. Only major lethal cardiac defects were detected antenatally.

KEYWORDS

INTRODUCTION: About 3% of newborns have a major physical anomaly that has cosmetic or functional significance. The cause of congenital anomaly seems to be multifactorial meaning a complex interaction of multiple minor genetic abnormalities and environmental risk factors

CAUSES: It include genetic cause, infections, drugs etc. There are 3 categories of genetic cause include numerical defects, structural defects, mosaicism. Among numerical defects down syndrome is the most common. Other trisomy include edward, patau, cat eye syndrome. Structural abnormalities include deletion, translocation ,inversion, mosaicism. Turner syndrome 45x0and klinefelter's are the sex chromosomal abnormalities. Mendelian disorders caused by a mutation in the single gene or loci. Types are autosomal dominant, autosomal recessive ,x linked disease. Dietary deficiency of folic acid is associated with spina bifida. Ingestion of harmful substances by the mother (eg alcohol, mercury, some drugs)can cause recognisable birth defects. Infections include toxoplasmosis, cytomegalovirus, rubella ,varicella zoster, syphillis. Other causes include diabetes mellitus , drugs such as warfarin, alcohol, thalidomide, retinoic acid, phenytoin and finally idiopathic.

CLASSIFICATION OF MALFORMATIONS: RCOG in 1997 classified malformations into lethal, severe, moderate. LETHAL malformation include anencephaly, bilateral renal agenesis, giant hygroma, osteochondro dysplasia, icthyosis congenita. SEVERE malformation include hydrocephalus, spina bifida,esophageal atresia, tof, absent uterus, ectodermal dysplasia of skin, posterior urethral valve, pda, asd, pulmonary hyperplasia, trachael stenosis, pierre robbin sequence etc. MODERATE include imperforate hymen, septal deviation, choanal atresia,craniostosis,hip dysplasia,microtia,aniridia,cataract,eyelid defects.

PRENATAL DIAGNOSIS : It is recommended for women 35 years or older at the time of delivery. Previous child with a chromosomal abnormality, several miscarriages, parental consanguinity, exposed to some drugs, infections, radiation exposure. Screening is achieved by biochemical, sonological and genetic assessment. Invasive procedures like chorionic villus biopsy, amniocentesis, percutaneous skin biopsy. Molecular genetic procedure include PCR, southern blotting, FISH, linkage analysis.

AIM OF THE STUDY: To study the prevalence and incidence of malformations in out institute. To study the pattern of malformation. To assess the influence of various risk factors such as age, gravidity, previous obstetric history,consanguinity,1st trimester events on the occurrence of malformation.

MATERIALS AND METHODS: A hospital based cross sectional study was conducted on patients taking into account all the deliveries occurred in our institution for a period of one year. Total number of deliveries was 17890. A detailed history regarding the patients and husband age, parity ,occupation, previous obstetric outcomes, family history of malformed babies, degree of consanguinity, any history of exposure to fever, teratogens ,environmental factors. Mediscan systems extended a big hand in helping out in further confirmation of diagnosis. Genetic counselling was extended to the patients by the genetic department at mediscan systems. Prenatal diagnosis were done to find the karyotype of the fetuses where we suspect genetic etiology. Post delivery of the fetus was examined in detail with regard to obvious external anomaly. A live born with anomalies were admitted to the newborn intensive care unit of our hospital. Surgically correctable anomalies were referred to the department of paediatrics surgery at institute of child health. Mother was given genetic counselling before discharge from the patient.

TABLE 1. INCIDENCE OF VARIOUS MALFORMATIONS

SYSTEM	INCIDENCE
CNS	131
SYNDROMES	44
CVS	19
SKELETAL	17
FACE	16
GUT	9
DIAPHRAGMATIC HERNIA	8
GIT	7
SACROCOCCYGEAL TERATOMA	3

TABLE 2. MALFORMATIONS DETECTED BY ULTRASOUND

DETECTED	190	74%
NOT DETECTED	37	14%
USG NOT DONE	27	10%

RESULT OF THE STUDY: CNS defects were the most recognized malformation at birth. CNS malformations are better detected in the antenatal USG than other systems and most of the anomalies are potentially lethal, hence accounting for the higher numbers.

The pick up rate of cvs defects by USG is very low requiring expertise and high resolution recovery. Most of the anomalies are not detected at birth as they show up symptoms later in the first week of life and only lethal anomalies like complex cardiac disease manifest in labour room. Cardiac defects need to be reconfirmed by imaging like echo and doppler. From this it is inferred that still cardiac defects form the core of congenital defects, for which we need to expertise upon their detection rate and prevention.

Target or anomaly scan has halved the burden of birth defects and help out the mother to prepare herself for the termination and plan the future pregnancies.75% of the anomalies were picked up in our study by ultrasound. There were of course 15% missed cases by routine usg. 10% of the mothers had no ultrasound done throughout the pregnancy. Some anomalies were picked up later in the third trimester scans which were unnoticed in previous scans.

CONCLUSION: Congenital malformations though cannot be prevented totally, can be minimized and if detected earlier will reduce the mental agony in the mother and her family. It can be minimised by prenatal counselling , periconceptional folate and prenatal diagnosis. It has become our professional responsibility to identify those couples who are at risk of having abnormal fetus . Early antenatal diagnosis results in earlier MTP which will decrease the maternal morbidity and the mental health in the mother .Conditions amenable to surgical correction in the neonatal period or in utero treatment can be planned if possible. A HAPPY MOTHER IS OBSTERICIAN''S PRIDE.A HEALTHYS BABY IS NATION'S PRIDE.

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