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Prevalence of congenital malformations in newborns

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Introduction

Potter defined congenital malformation as an " abnormality of structure detected at birth or during first few weeks of life26 ". It can be expanded to include functional disturbances in the organs.
Congenital conditions can be referred to as diseases, defects, disorders, anomalies, or simply genetic differences. The usage overlaps, but also involves a valued judgement as to the harmfulness of the condition.
Congenital malformations involving the brain are the largest group at 10 per 1000 live births, compared to heart at 8 per 1000, kidneys at 4 per 1000, and limbs at 1 per 1000. All other physical anomalies have a combined incidence of 6 per 1000 live births16.
Congenital malformations of the heart have the highest risk of death in infancy, accounting for 28% of infant deaths due to birth defects, while chromosomal abnormalities and respiratory abnormalities each account for 15%, and brain malformations about 12%42.
The cause of 40-60% of congenital physical anomalies (birth defects) in humans is unknown. These are referred to as sporadic birth defects, a term that implies an unknown cause, random occurrence, and a low recurrence risk for future children25. For 20-25% of anomalies there seems to be a "multifactorial" cause, meaning a complex interaction of multiple minor genetic abnormalities with environmental risk factors. Another 10-13% of anomalies have a purely environmental cause (e.g. infections, illness, or drug abuse in the mother). Only 12-25% of anomalies have a purely genetic cause. Of these, the majority are chromosomal abnormalities41. The rapid development of technology allowing early and accurate prenatal diagnosis of fetal disorder has revolutionised the practice of obstetrics over 20 years. Beginning with simple cytogenetics17 to detect chromosomal abnormality in amniotic fluid cells, there are new methods that permit rapid detection of mutant genes by using minute quantities of fetal DNA37. These techniques coupled with molecular genetics allow detection of a list of inherited conditions that is expanding8 almost daily. Of paramount importance is the ability to provide counselling regarding various screening, diagnostic technique and treatment options.

Objective:
To estimate the prevalence of congenital malformations in newborns

MATERIALS AND METHODS

Study design : Prospective cohort study
Study population : All women delivered vaginally were recruited for the study based on the inclusion and exclusion criteria.
The study was approved by the hospital ethical committee.
All the cases detected to have congenital malformations by an antenatal ultrasound were analysed by detailed questionnaires. A detailed history regarding the patient and husband's age, parity, occupation, previous obstetric outcomes, family history of malformed babies, degree of consanguinity and the events in the present pregnancy were analysed. Any history of maternal exposure to fever, teratogens, drugs and other environmental factors were ascertained. Enquiries were put forward with regard to maternal medical diseases like diabetes mellitus. Patients were enquired regarding the intake of preconceptional folate. Investigations pertaining to the case were done if necessary. Blood sugar was done in indicated cases.

Post delivery, the fetus was examined in detail with regard to obvious external anomalies, weight, sex. A live born baby with anomalies was admitted to the newborn intensive care unit of our hospital and

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followed up thereafter. Surgically correctable anomalies were referred to the department of paediatric surgery.
Mother was given genetic counselling before discharge from the hospital. She was instructed regarding future pregnancy and stressed upon the importance of pre-conceptional folate intake.
Defects which caused serious structural, cosmetic and functional disability requiring surgical or medical management were classified as major anomalies. The rest were categorised as minor anomalies. The major malformations were divided into Central nervous system, Skeletal, Gastro-intestinal, Genitourinary, Cardio vascular system syndromes and miscellaneous disorders. The babies were followed up

Results
The bulk of our deliveries are confined to the age group of 21-25 yrs. Though 50 % of malformations occurred in this age group, there is no actual correlation between age and defects.
There is no significant relationship between parity and congenital malformations.
In our study the malformations occurred mostly in non- consanguineous marriages. No correlation could be made out between the two.
The malformations showed slight predilection towards male sex.
Around 22 % of malformed babies were born to mothers with previous history of pregnancy wastage and 11 % had previous malformed babies. This shows there is a genetic role playing important in the etiology of malformations 24 % of the parents were married consanguineously who gave birth to malformed fetuses.
The outcome of malformations are dependent on the gestational age at detection. Earlier they are picked up, more the number of recommended termination . 7.3 % of the terminations were done for malformations.
Maternal febrile illness in the first trimester had a definite predeliction for malformations as pointed out in previous studies.
Infant of diabetic mothers on evaluation showed a higher rate of cardiac and neural defects.
One baby had fetal Hydantoin syndrome, the mother had been taking phenytoin through out her pregnancy. TORCH screening is not routinely done for all patients, but three of the mothers with recurrent pregnancy wastage showed increased titre.
The most common malformations encountered in our hospital were Central nervous system defects. The reason sought is they are picked up more by antenatal Ultrasound and most of them are lethal anomalies.
Anencephaly was the most reported defect followed closely by spinal defects. Peri conceptional folate were not taken by the patients. Only post conceptional folate were taken by few of them.
Only major lethal cardiac defects were detected antenatally and at birth which accounted for 7.5 % of malformations.
Ultrasound could pick up only 75 % of the anomalies. 15 % of the anomalies were missed by routine Ultrasound which were mostly cardio vascular anomalies.

DISCUSSION
The global incidence of congenital malformations detected at birth is 2-3%. In India, the nation wide prevalence is high. However there is no systematic surveillance for birth defects in India.
In studies from other parts of India, the incidence varied from 0.3% to 3.6%. The rate in the present study is comparable to the studies from Varanasi, Manipal and Allahabad.
A higher incidence of congenital malformations has been reported from centers like PGIMER Chandigarh and JIPMER Pondicherry, which may be because of higher autopsy rates at these centers.
Central nervous system defects were the most recognised malformations at birth and formed the bulk of defects accounting of 76\10000. Nervous system malformations are better detected in the antenatal ultrasound than other systems and most of the anomalies are potentially lethal, hence accounting for the higher numbers. Moreover they are quite obvious to the labour room personnel, requiring no additional confirmation by investigations and imaging.

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3. As against the wide prevalence of cardiovascular defects across the total population, the reported prevalence in the present study is extremely less. This joins hands with rest of the studies done in many centres across the country. The reason being;
a) The pick up rate of cardio vascular defects by ultrasound is very low requiring expertise and high resolution machinery.
b) Most of the anomalies are not detected at birth, as they show up symptoms later in the first week of life and more. Only lethal anomalies like complex cardiac disease manifest in the labouring room.
c) Cardiac defects need to be reconfirmed by imaging like Echo and Doppler which are reported later.
d) Around 94 cases of congenital heart disease were suspected and picked up by postnatal follow up of the baby. (stastistics collected from Institute of child health).
e) 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 its prevention.
4. Targeted or anomaly scan has halved the burden of birth defects and help 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 ultrasound. 10% of the mothers had no scan done through out the pregnancy. Some anomalies were picked up later in the third trimester scans, which were un-noticed in the previous scans.

Conclusion

Congenital malformations though cannot be prevented totally, can be minimised and if detected earlier will reduce the mental agony in the mother and her family. It can be minimised by prenatal counselling, peri conceptional folate and prenatal diagnosis. It has become our professional responsibility to identify those couples who are at risk of having abnormal fetus. Early ante-natal diagnosis results in earlier termination which will decrease the maternal morbitity 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. Broader the diagnosis of birth defects made if Obstetricians, Perinatalogist, Sonologist and Laboratory Personnel work hand in hand.

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