# A DISSERTATION

## ON

## INCIDENCE OF CONGENITAL ANOMALIES IN NEWBORN IN TERTIARY CARE HOSPITAL

**Submitted to**
**THE TAMILNADU DR. M. G. R. MEDICAL UNIVERSITY**
**CHENNAI**

**in fulfilment of the regulations**
**for the award of**

**M.D DEGREE IN PAEDIATRIC MEDICINE**
**BRANCH VII**


**GOVERNMENT MOHAN KUMARAMANGALAM**
**MEDICAL COLLEGE, SALEM.**

**MARCH 2010**

# CERTIFICATE

Certified that this dissertation entitled **"INCIDENCE OF ONGENITAL ANOMALIES IN NEWBORN IN TERTIARY CARE HOSPITAL"** is a bonafide work done by **Dr.N.MANIVANNAN** post graduate student of Paediatric Medicine, Government Mohan Kumaramangalam Medical College, Salem-636030, during the Academic year 2007-2010.

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# DECLARATION

I declare that this dissertation entitled **"INCIDENCE OF ONGENITAL ANOMALIES IN NEWBORN IN TERTIARY CARE HOSPITAL"** was done by me at Government Mohan Kumaramangalam Medical College Hospital, Salem under the guidance and supervision of my department chiefs **Prof.Dr.R.SIVAGAMASUNDARI** and **Prof.Dr.M.RATHINASAMY.** It is submitted for the fulfillment of the award of the degree of MD (Paediatrics) for the March 2010 examination to be held under the TamilNadu Dr. MGR Medical University, Chennai. This has not been submitted previously by me for the award of any degree or diploma from any other university.

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# ACKOWLEDGEMENT

I am very grateful to our beloved Former Dean, Government Mohan Kumaramangalam Medical College and Hospital **Prof.Dr.P.SHANMUGAM, M.S., M.ch.,** for permitting me to do this study at GMKMC Hospital.

I am also thankful to **Dr. K.V.KANAGASABAI, M.D.,** Present Dean, Govt.Mohan Kumaramangalam Medical College and Hospital, for his whole hearted co-operation and support for the completion of this dissertation.

At the outset I express my sincere gratitude to my most beloved and revered teacher **Prof.Dr.R.SIVAGAMASUNDARI, M.D., DCH.,** HOD, Professor of Paediatrics, GMKMC Hospital, Salem for her constant interest, timely guidance, inspiring analysis and enthusiastic supervision in the preparation of this dissertation.

I am extremely thankful to Associate Professor of paediatrics **Prof.Dr.M.RATHINASAMY, M.D.,D.C.H.,** for his guidance, invaluable help, encouragement and support throughout the study.

I thank **Dr.T.S.SUNDARARAJAN, M.D.D.C.H.,** Registrar in Paediatrics, GMKMC Hospital for showing special interest in my work and for his valuable advice.

I am grateful and thankful to all my Assistant Professors in our department who had offered constructive criticism and valuable suggestion during the preparation and

presentation of this work.

I express my sincere thanks to my fellow postgraduates for their help and co-operation throughout the work.

I remain thankful to the host of persons who have helped me in all respects to bring out dissertation work a success.

Last but not the least I am grateful to all my newborn babies and their parents who co-operated very well during the study.

# CONTENTS

# INTRODUCTION

Just about three decade ago (1976) congenital malformations comprised 8 % of perinatal deaths, from available data and ranked fifth as a cause of perinatal mortality[1]. But the trend is rapidly changing over years. In a recent study from AIIMS, Delhi[2] congenital malformations contributed to 15.7 % of perinatal deaths and ranked fourth as cause of perinatal deaths. In another study from Hyderabad[3] 17.8 % of perinatal death were due to congenital malformation, being the second commonest cause. This changing trend over years warns us that with the control of nutritional and infections diseases, congenital malformations will come to the forefront as it is in the West now. Therefore a systematic study of congenital malformations in live and still births is of paramount importance to evaluate etiological agents, to undertake preventive steps, to intervene early in the treatment of life threatening malformation at birth and thus reducing perinatal mortality due to them.

Malformations and dysplasia both affect intrinsic structures. A malformation is a primary structural defect arising from a localized

error in morphogenesis, resulting in the abnormal formation of a tissue or organ.

**DYSPLASIA**

Refers to an abnormal organization of cells into tissue.

**DEFORMATION**

Deformation is an alteration in shape (or) structure of a structure (or) organ that has differentiated normally.

**DISRUPTION**

Disruption is a structural defect resulting from the destruction of a structure that had formed normally before the insult. A major malformation has serious medical, surgical or cosmetic consequences. A minor malformation is defined as an unusual morphologic feature that are of no serious medical or cosmetic consequences to the patient.

The common pattern of multiple structural anomalies as follows.

**ASSOCIATION**

Non Random combination of anomalies wherein the individual components occur together more frequently than would be expected by chance, Ex.

- VACTERL Association
- CHARGE Association

**SEQUENCE ANOMALIES**

Occur as a result of a cascade of seemingly unrelated consequences, which cannot be explained on developmental and embryologic groups. Ex.

- Potter's sequence

**FIELD DEFECTS**

Anomalies of different body organs which differentiate together during Embryogenesis due to anatomical proximity. Ex.

- Poland anomaly

**SYNDROME**

Defined as unique constellation of multiple anomalies that repeatedly occur in a consistent pattern.

Incidence varies in varies surveys. The present consensus emerged that approximately 3 % of newborn children are affected by significant congenital malformations[4]. Hospital based prospective studies of live and still births from different parts of India, the

incidence of congenital malformations has varied from 2.5 – 40 / 1000 births.

Etiological agents could be broadly divided into 4 categories specific teratogenic agents (8–10%), monogenic (15–25%), chromosomal (15.28%) and unknown (including multifactorial) (40-65%). A teratogen is an agent, applied during prenatal life produces a permanent postnatal damage, change in morphology or function[6]. Such agents can be chemicals, drugs, virus, or physical or deficiency states. Majority of the malformations are due to complex involvement of genetic and environmental factors. A study in Karnataka showed a significant contribution of consanguinity – 8.01 % as compared to non consanguineous group 2.42 %. The information that nutritional deficiency may be responsible for neural tube defects[7] is exciting and highly relevant to a country like ours, where nutritional deficiencies are common. As regards prenatal infections, as a cause of malformations, very little information is available in our country.[8,9]

As most of the Indian studies are hospital based and hence do not represent the problem in the community as hospital records are based by inclusion of high risk mothers, further many of births in India specially in rural areas occur at home and infants with abnormalities are not brought to the hospital for various reasons, and there are many

difficulties in the evaluation of still births in the community. There is no standard protocol in the study of malformation in India. Excellent normal standards for physical features are available for Western populations and there is an urgent need to develop our own standards for Indian population by multicentric collaborative study.

Centres with good laboratory facilities like chromosomal study, biochemical screening for inborn errors of metabolism, ultrasonography and radiological services should develop standard protocols for collection of specimens like blood and skin biopsy, their preservation and transportation to regional centres from the periphery. These centres should take part in training the pathologist in autopsy studies of foetuses and newborns. Primary prevention is the ultimate aim .This is possible through the discovery, elimination, modification and control of teratogens in our environment. Malformations due to single gene mutation and chromosomal disorders and the only way these could be limited is by genetic counselling and induced abortions in areas where the genetic factors operate as a cause of malformations through the practice of consanguineous marriages, education of people against such practice could reduce the incidence of malformations. Gene therapy through exciting is still in experimental stage.

# REVIEW OF LITERATURE

In the past many eminent people have studied about the incidence of congenital malformations across the country and had come out with various results.

**P.C.Mishra et al**[20] and **S.Chandra et al**[33] in their study reported incidence of congenital malformation 8 per 1000 and 36 per 1000 respectively. **Aiyar.R.R et al**[18] and **Tibrewala et al**[19] in their study, prevalence rate of malformation, reported 17 per 1000 and 18.7 per 1000 respectively.

**Nair et al, Saifullah et al**[17] and **Garavalingappa et al** in their study observed central nervous system anomaly to be the commonest. In contrast **Kulshereshtha et al**[28] in the community based study found gastrointestinal anomalies to be the commonest . Among the central nervous system anomalies, **Verma et al**[24] and colleagues reported a high incidence of neural tube defects in Punjab and Rajasthan.

**Anand et al** in his study has reported minor anomalies 4 per 1000 births. **Verma et al, Garvalingppa et al**[36] has shown 4 times

higher rate of malformations in still births when compared to live births. **Thirumalisunder Subramani[42] and Mathur et al** reported a male : female ratio of 1.7:1 and 2:1 respectively. **Guha et al** reported female preponderance for anencephaly though **Choudhery et al[43]** found more male babies with the defect.

In a study carried out at AIIMS the incidence of malformations in preterm babies was 5.3 % and in term babies 2.6 %. As per the studies concluded by **Khanna et al[34] , Dutta et al[32]** and **Anand et al[25]** , the rate of malformations in preterm babies were 3.8 % , 2.4 %, and 2.54 % respectively.

**Kesavan et al[47]** in a detailed study of 23,000 births in Chennai showed a definite increase in the rate of malformation in the offspring of consanguineous marriage 1.64 % are non consanguineous 2.36 % among first cousins, 2.18 % among maternal uncle niece marriage and 1.86 % among distant relations. **Sugunabai et al[35]** reported consanguineous parents had 3.59 % malformed babies . But two survey of the general population in South India failed to reveal any significant effect of consanguinity in the ratio of malformations . One was a planed prospective survey of a population about 10000 near Vellore

which aimed to study the effect of consanguinity of human reproduction.

With regard to maternal age **Sugunabai et al, Kulshuesltra et al** and **Verma et al** found higher incidence of anomalies in elderly mothers.

With the above information , the present study was conducted to know the incidence and distribution of congenital anomalies.

# AIMS OF THE STUDY

1. To find out the incidence of congenital anomalies in a 1 year period of 5000, consecutive births delivered at GMKMCH, Salem.

2. To find out the prevalent types of congenital anomalies.

3. To find out the contribution of consanguinity in the occurrence of congenital malformations.

4. To find out prevalence of malformations in relation to the gestation and sex of the babies.

5. To find out the incidence of congenital anomalies in still birth.

6. To find out the probable aetiology of congenital anomalies.

# MATERIALS AND METHODS

The study was conducted at Government Mohan Kumaramangalam Medical College Hospital, Salem. Five Thousand babies born of consecutive deliveries were taken for the study, over the period of one year . All mothers were interrogated within 48 Hours of delivery as per the proforma prepared, which contains the following particulars like, maternal and paternal age, consanguinity, detailed antenatal history with reference to exposure to teratogens especially during 1$^{st}$ Trimester and medical disease complicating pregnancy like Diabetes, Rheumatic heart disease, Hypertension, detailed obstetric history with reference to previous abortions and still birth. Routine investigations like haemoglobin, urine analysis, blood grouping and Rh typing, VDRL, and HIV were done for all cases and blood sugar, renal function and liver function tests were done when indicated for mothers.

Every newborn was subjected to detailed examination from head to toe within 48 hours of birth. Assessment of the newborn included birth weight, sex, live born/still born, gestational age and details of congenital malformations. All were recorded in a pre designed

proforma. A gavage tube was used to check choanal and oesophageal atresia, Anorectal anomaly in suspected cases. All the newborns were followed up every day till the time of discharge from the hospital. Necessary investigations were done wherever required. Cardiovascular anomalies were subjected to ECG, X ray chest AP view and ECHO cardiography. The umbilical cord stump was examined to note down the anomalies of arteries and vein. Placenta was examined in Detail.

**Autopsy**

Autopsies were performed in still born and neonatal deaths, only for those cases where consent was given by the father and close relatives. A detailed external examination was carried out prior to autopsy.

The internal examination was carried out in the following manner[11]. An-I-shaped skin incision was made extending from the chin straight of umbilicus. Each body cavities were examined separately.

Abdominal cavity was examined as follow:

The recti muscles of the abdomen were divided about 1 cm above symphysis and separated. The thickness of the abdominal wall muscles noted and condition of the abdominal cavity and organs were observed before anything was disturbed to find out fluid or meconium

in the abdominal cavity.

Stomach with the intestine was removed by applying double ligatures one at the lower end of the oesophagus and other at the rectosigmoid junction. Detailed examination was done and noted visceral positions. Stomach was examined for diverticulosis, duplication and perforations. Duodenum was examined for annular pancreas which surrounds the second part of the duodenum. Small bowels were examined for duplications, atresia and perforations. Rectum and anal canal were examined for anorectal malformations.

Liver and gall bladder were examined for congestion, visceral situs and other anatomical abnormalities. Spleen was examined to note down size, accessory spleen or asplenia.

After removal of the other organs kidneys and ureters were freed from the posterior abdominal wall. Capsule was stripped, the position, size and shape of both the kidneys were noted, Ureters were noted for mega ureter and duplication. Pubis were split and pelvic viscera was removed for detailed examination.

Thorax was opened by cutting the costo-chondral junction. The

position of the cardiac apex, morphology of the atrial appendages, and its positions were noted.

**Veins and Arteries**

Aortic arch and its branches were dissected in situ . The state of ductus arteriosus was noted, particularly its size in relation to the pulmonary trunk and aorta.

Dissection of the heart was carried out as follows[12]: Heart was held at the apex and lifted upward and pulmonary vessels , superior and inferior vene cavae and the ascending aorta were cut as far away as possible from the base of the heart . The pericardium was examined and incised with the tip of the scissors and the heart was exposed . Heart was opened in such way in order to minimize the risk of damage to the internal architecture. Each part of the heart was examined in detailed for congenital heart disease.

Lungs were examined to differentiate from live birth or still birth. Oesophagus and trachea examined for oesophageal atresia and tracheoesophageal fistula.

**Examination of Head**

An incision made in the scalp which starts from the region of the mastoid process just behind one ear and was carried over the vertex of the scalp of the back of the opposite ear. The scalp was reflected forwarded to the superciliary ridges and backwards to the point just below the occipital protuberance. Any bruising and cephalhematoma were noted. Skull was opened as described by Beer with a knife, incision is made into the anterior fontanelle at its posterior margin, about 5 mm, from the mid line.

After reflecting the flaps, the vertex of the brain and the terminations of the pia veins into the superior longitudinal sinus were examined for haemorrhage. Hemispheres were gently pushed sideways and falx cerebri examined. Hematoma may be found between the falx and the medial aspect of the hemisphere, or between the two dural layers. Superior longitudinal sinus was opened and examined for thrombi. The falx was separated at its antero-inferior margin and the brain was removed by inserting the fore fingers of the left hand between the frontal lobes and skull and drawing frontal lobes backwards and cutting the vessels and nerves at the base. The tentorium was cut along the posterior border of the petrous bone. Knife was passed into the occipital foramen and cervical cord, first cervical

nerves and vertebral arteries were cut as far below as possible. The right hand grasps the cerebellum and the brain was removed from the cranial vault.

Brain was placed upside down on a board. The medulla was pulled away from the cerebellum to open up the fourth ventricle. Pons and medulla were moved one by one after detailed exam, the cerebral hemispheres were separated and examined. The lateral and the third ventricles were exposed and the aqueduct was traced. Then the cerebral hemisphere were cut in slices at intervals of 1 cm for detailed exam.

# OBSERVATIONS

Of the five thousand consecutive deliveries 48 deliveries were multiple delivers and number of still births were 108. The incidence of congenital anomalies, was 30.4 per 1000 live birth (152 cases). Major malformations were present in 20.8 per 1000(104 cases) while minor malformations were 9.6 per 1000 (48 cases).

**TABLE-1**
**CONGENITAL MALFORMATION : FREQUENCY AND DISTRIBUTION IN RELATION TO SEX AND LIVE AND STILL BIRTH**

| Particulars | Total Babies | Malformation | | Normal Babies |
|---|---|---|---|---|
| | | Number | Percentage | |
| **LIVE BIRTH** | 4892 | 141 | 2.88% | 4751 |
| Male | 2518 | 81 | 1.65% | 2437 |
| Female | 2374 | 60 | 1.22% | 2314 |
| **STILL BIRTH** | 108 | 11 | 10.18% | 97 |
| Male | 59 | 7 | 6.48% | 52 |
| Female | 49 | 4 | 3.70% | 45 |

Congenital malformations were seen more in still births as compared to the live birth, the frequency being 10.18% and 2.88% respectively and was found to be statistically significant ($p < 0.05$).

## SEX DISTRIBUTION AND CONGENITAL MALFORMATION

88 babies were males and 64 were females. Male to female ratio was 1.35:1. In our study there was increase incidence of congenital malformations in male babies and it was statistically significant ($p < 0.001$).

## TABLE – 2

## DISTRIBUTION OF MALFORMATION ACCORDING TO MATERNAL AGE

| Maternal Age | Total Babies | Malformation | | Normal Babies |
|---|---|---|---|---|
| | | Numbers | Percentage | |
| 15-19 yrs. | 536 | 11 | 2.05% | 525 |
| 20-24 yrs. | 2297 | 66 | 2.87% | 2231 |
| 25-29 yrs. | 1104 | 37 | 3.35% | 1067 |
| 30-34 yrs. | 935 | 31 | 3.31% | 904 |
| >35 yrs. | 128 | 7 | 5.46% | 121 |

Maternal age was classified into 5 groups as showed in table-2. The incidence of congenital malformations increases among mothers with age group 35 yrs and above (5.46%) compared to 20-24 yrs of age group it is only (2.87%). This was found to be statistically significant ($p<0.05$).

## TABLE -3

## DISTRIBUTION OF MALFORMATION ACCORDING TO MATERNAL PARITY

| Parity | Total Babies | Malformations | | Normal Babies |
|---|---|---|---|---|
| | | Numbers | Percentage | |
| 1 | 2625 | 66 | 2.51% | 2559 |
| 2 | 1573 | 42 | 2.67% | 1531 |
| 3 | 620 | 32 | 5.16% | 588 |
| 4 | 156 | 10 | 6.41% | 146 |
| 5 | 26 | 2 | 7.69% | 24 |

According to parity mothers were classified into five groups. It was observed that as the parity increases, there was an increase in the incidence of congenital malformations and it was found to be statistically significant ($p<0.05$).

**TABLE-4**

**CORRELATION OF ANTENATAL FACTORS IN 1ST TRIMESTER WITH CONGENTIAL MALFORMATIONS**

| Maternal Factors | Present | Percentage | Absent |
|---|---|---|---|
| Drug Intake | 9 | 6.29% | 143 |
| Fever | 15 | 10.94% | 137 |
| Vaginal Bleeding | 3 | 2.01% | 149 |
| Radiation | 0 | 0 | 152 |

About 17 percent of mothers with malformed babies had significant factors contributed to the aetiology of malformations. 6.29 percentage of mothers gave history of drug intake during the early pregnancy. 10.94 percentage of mothers had fever in the first trimester. Out of 28 cases of central nervous system malformations 4 mother had fever and deliver. 1 baby with anencephaly, 2 babies with microcephaly and 1 had meningomylocele.

Three mother had history of vaginal bleeding during first trimester. One of them had delivered a baby with CTEV.

**TABLE-5**

**CORRELATION OF COMPLICATION DURING PREGNANCY WITH CONGENITAL MALFORMATIONS**

| Complication | Total Babies | Malformation | | Normal Babies |
|---|---|---|---|---|
| | | Numbers | Percentage | |
| Diabetes | 12 | 4 | 33.33% | 8 |
| Hydramnios | 22 | 7 | 31.81% | 15 |
| PIH | 46 | 3 | 6.52% | 43 |

Of 7 cases of hydramnios 2 mothers delivered babies with oesophageal atresia/ tracheoesophageal fistula, 3 babies with hydrocephalus and 2 babies with anencephaly.

Four mothers gave history of diabetes during antenatal period, 1 baby had congenital cyanotic heart disease and died before doing ECHO. Other 2 baby had bilateral CTEV and 4$^{th}$ baby had ASD.

**TABLE-6**

**CORRELATION OF MALFORMATION WITH GESTATION**

| Gestation | Total Babies | Malformations | | Normal Babies |
|---|---|---|---|---|
| | | Number | Percentage | |
| Pre Term | 190 | 12 | 6.31% | 178 |
| Term | 4804 | 140 | 2.91% | 4664 |
| Post Term | 6 | Nil | Nil | 6 |

There is an increased incidence of malformation was observed among pre term babies. This is statistically significant ($p<0.05$).

**TABLE-7**

**MULTIPLE PREGNANCY AND CONGENITAL MALFORMATION**

| Pregnancy | Total Babies | Malformation | | Normal Babies |
|---|---|---|---|---|
| | | Number | Percentage | |
| Single | 4952 | 148 | 2.98% | 4804 |
| Twins | 48 | 4 | 8.33% | 44 |

Out of 48 twin deliveries 4 babies (8.33%) were malformed. In comparison 148 (2.98%) out of 4952 had malformation. This is found to be statistically significance ($p<0.05$).

**TABLE-8**

**CORRELATION OF CONSANGUINITY AND CONGENITAL MALFORMATION**

| Consanguinity | Total Cases | Percentage |
|---|---|---|
| 2nd Degree | 69 | 45.39% |
| 3rd Degree | 53 | 34.86% |
| No Consanguinity | 30 | 19.73% |

80.25% of babies with congenital malformation were born of consanguineous marriage compared to other group which comprises of 19.75%.

## TABLE 9

## SYSTEMIC DISTRIBUTION OF CONGENITAL MALFORMATION

| Systems | Malformation | | No. of Live Birth | No.of. Still Birth | Neonatal Death | MB Per 1000 Birth |
|---|---|---|---|---|---|---|
| | No. | % | | | | |
| Central nervous system | 28 | 18.42% | 23 | 5 | 6 | 5.6 |
| Gastro intestinal system | 23 | 15.13% | 19 | 4 | 3 | 4.6 |
| Skeletal system | 20 | 13.15% | 20 | Nil | Nil | 4 |
| Oro facial | 12 | 7.89% | 12 | Nil | Nil | 2.4 |
| Cardio Vascular | 11 | 7.23% | 11 | Nil | 2 | 2.2 |
| Genito Urinary | 10 | 6.57% | 8 | 2 | Nil | 2 |
| Minor malformation | 48 | 31.57% | 48 | Nil | Nil | 9.6 |

In the study it was observed that majority of anomalies were found in central nervous system. The other systems where major anomalies recorded were gastro intestinal system and skeletal system.

## TABLE-10

## ANALYSIS OF CENTRAL NERVOUS SYSTEM MALFORMATION

| Malformations | No | % of MB | LB | SB | M | F | SGA | AGA | MB/1000 Births |
|---|---|---|---|---|---|---|---|---|---|
| Meningomyelocele | 7 | 4.60% | 7 | Nil | 5 | 2 | 5 | 2 | 1.4 |
| Anencephaly | 5 | 3.28% | 3 | 2 | 3 | 2 | 5 | Nil | 1 |
| Spina bifida | 4 | 2.63% | 4 | Nil | 2 | 2 | 2 | 2 | 0.8 |
| Microcephaly | 4 | 2.63% | 4 | Nil | 3 | 1 | 4 | Nil | 0.8 |
| Hydrocephalus | 3 | 1.97% | 2 | 1 | 2 | 1 | 2 | 1 | 0.6 |
| Hydrocephalus with meningomyelocele | 3 | 1.97% | 1 | 2 | 2 | 1 | 2 | 1 | 0.6 |
| Craniosynostosis | 1 | 0.65% | 1 | Nil | 1 | Nil | 1 | Nil | 0.2 |
| Agenesis of corpus callosum | 1 | 0.65% | 1 | Nil | 1 | Nil | 1 | Nil | 0.2 |

One baby with agenesis of corpus callosum born of 2nd degree consanguinity, it was the 3rd child. Previous two babies died in the neonatal period and this child presented with status epilepticus and diagnosis was established by cranial ultrasonogram. Baby died on the first postnatal day.

## TABLE-11

## ANALYSIS OF CARDIOVASCULAR SYSTEM

| Malformations | No | % of Mb | Live Birth | Still Birth | M | F | SGA | AGA | MB/1000 Births |
|---|---|---|---|---|---|---|---|---|---|
| VSD | 5 | 3.28% | 5 | Nil | 3 | 2 | 3 | 2 | 1 |
| VSD with PS | 2 | 0.4% | 2 | Nil | 2 | Nil | 1 | 1 | 0.4 |
| PS | 1 | 0.65% | 1 | Nil | 1 | Nil | Nil | 1 | 0.2 |
| ASD | 2 | 0.4% | 2 | Nil | 1 | 1 | 1 | 1 | 0.4 |
| Dextrocardia | 1 | 0.65% | 1 | Nil | 1 | Nil | 1 | Nil | 0.2 |

Most common congenital heart disease observed in the study is VSD (3.28%). Out of the 2 babies with congenital malformations in cardiovascular system born to a diabetic mother, one baby presented with congestive cardiac failure and cyanosis at birth. Chest X-ray showed cardiomegaly and ECG showed biventricular hypertrophy. Echocardiogram was not done due to very poor general condition and baby died after 36 hrs of life. The other baby had ASD.

Antenatally two mothers gave history of drug intake during the first trimester .In one case mother had taken anticonvulsants phenytoin and phenobarbitone and delivered with VSD.

5 babies born of consanguineous marriage, 3 of them were 2$^{nd}$ degree consanguinity.

## TABLE-12

## ANALYSIS OF GASTROINTESTINAL SYSTEM

| Malformation | No | % of MB | Live Birth | Still Birth | M | F | SGA | AGA | MB/1000 Birth |
|---|---|---|---|---|---|---|---|---|---|
| Cleft lip and cleft palate | 13 | 8.55% | 11 | 2 | 6 | 7 | 8 | 5 | 2.6 |
| Cleft lip | 5 | 3.28% | 5 | Nil | 4 | 1 | 3 | 2 | 1 |
| Ano rectal anomaly | 5 | 3.28% | 5 | Nil | 2 | 3 | 1 | 4 | 1 |
| Cleft palate | 3 | 1.97% | 3 | Nil | 2 | 1 | Nil | 3 | 0.6 |
| TEF | 3 | 1.97% | 3 | Nil | 2 | 1 | 2 | 1 | 0.6 |
| Megacolon | 2 | 1.31% | 2 | Nil | 1 | 1 | Nil | 2 | 0.4 |
| Exomphalus | 2 | 1.31% | 1 | 1 | 1 | 1 | Nil | 2 | 0.4 |
| Pierre Robin syndrome | 1 | 0.65% | Nil | 1 | Nil | 1 | Nil | 1 | 0.2 |
| Omphalocele | 1 | 0.65% | 1 | Nil | 1 | Nil | 1 | Nil | 0.2 |

Cleft lip with palate formed the major group. 3 of them had multiple anomalies - CTEV in one case, microcephaly in two. Mothers gave history of drug intake in early pregnancy.

Next major group is anorectal malformation. Total of 5 cases, 2 cases were of low anorectal anomalies. 3 of them were female children. In this group we had two male child with high anomaly. 3 cases of tracheao oesophageal fistula, all the cases were diagnosed and underwent surgery.

## TABLE-13

## ANALYSIS OF GENITOURINARY SYSTEM

| Malformations | No | % of MB | Live Birth | Still Birth | M | F | SGA | AGA | MB/ 1000 Births |
|---|---|---|---|---|---|---|---|---|---|
| Undescended testis | 4 | 2.63% | 4 | Nil | 4 | Nil | 2 | 2 | 0.8 |
| Hypospadias | 3 | 1.97% | 2 | 1 | 3 | Nil | 1 | 2 | 0.6 |
| Epispadias | 1 | 0.65% | 1 | Nil | 1 | Nil | Nil | 1 | 0.2 |
| Hydronephrosis | 1 | 0.65% | 1 | Nil | 1 | Nil | 1 | Nil | 0.2 |
| Micropenis | 1 | 0.65% | Nil | 1 | 1 | Nil | Nil | 1 | 0.2 |

Analysis of genitourinary system showed 4 out of 10 cases were undescended testis and 3 cases showed hypospadias and one among them was a still born with abdominal distension. The autopsy of the still born showed bilateral hydronephrosis with posterior urethral valve.

## TABLE-14

## SKELETAL ANOMALIES

| Malformations | No | % of MB | Live Births | Still Births | M | F | SGA | AGA | MB/1000 Births |
|---|---|---|---|---|---|---|---|---|---|
| CTEV | 10 | 6.57% | 10 | Nil | 3 | 7 | 4 | 6 | 2 |
| Polydactyly | 6 | 3.94% | 6 | Nil | 2 | 4 | 2 | 4 | 1.2 |
| Osteogenesis Imperfect | 1 | 0.65% | 1 | Nil | Nil | 1 | Nil | 1 | 0.2 |
| Polysyndactyly | 1 | 0.65% | 1 | Nil | Nil | 1 | Nil | 1 | 0.2 |
| Arthrogryphosis | 1 | 0.65% | 1 | Nil | Nil | 1 | 1 | Nil | 0.2 |
| Achondroplasia | 1 | 0.65% | 1 | Nil | 1 | Nil | Nil | 1 | 0.2 |

10 children with CTEV in this study 3 of them delivered by twin deliveries, 2 of them were delivered by breech, 2 babies were preterm and 1 had cleft lip.

We had a case of osteogenesis imperfecta diagnosed immediately after birth.

## TABLE-15

## MINOR ANOMALIES

| Malformations | No | % of MB | Live Birth | Still Birth | M | F | SGA | AGA | MB/ 1000 Births |
|---|---|---|---|---|---|---|---|---|---|
| Preauricular sinus | 3 | 1.97% | 3 | Nil | 2 | 1 | 1 | 2 | 0.6 |
| Epicanthal fold | 7 | 4.60% | 7 | Nil | 4 | 3 | 3 | 3 | 1.4 |
| Preauricular skin tag | 6 | 3.94% | 6 | Nil | 4 | 2 | 4 | 3 | 1.2 |
| Overriding of toes | 2 | 1.81% | 2 | Nil | 1 | 1 | 1 | 1 | 0.4 |
| Sacral dimple | 3 | 1.97% | 3 | Nil | 2 | 1 | 2 | 1 | 0.6 |
| Low set ears | 4 | 2.63% | 4 | Nil | 1 | 3 | 2 | 2 | 0.8 |
| Hypertelorism | 3 | 1.97% | 3 | Nil | 1 | 2 | 3 | Nil | 0.6 |
| Clinodactaly | 3 | 1.97% | 3 | Nil | 1 | 2 | 2 | 1 | 0.6 |
| Polydactyly | 4 | 2.63% | 4 | Nil | 4 | Nil | 2 | 2 | 0.8 |
| Simian crease | 4 | 2.63% | 4 | Nil | 1 | 3 | 1 | 3 | 0.8 |
| Accessory Nipple | 5 | 3.25% | 5 | Nil | 4 | 1 | 1 | 4 | 1.0 |
| Malformed ears | 4 | 2.63% | 4 | Nil | 4 | Nil | 2 | 2 | 0.8 |

# DISCUSSION

## HISTORICAL ASPECTS OF BIRTH DEFECTS[12]

Congenital malformations present at birth have been known since earliest times . Primitive man's interest and fascination with these bizarre phenomena have found expression in drawings, carvings and scriptures. At various periods in history, children born with congenital defect were attributed to astrological factors, divine will, witches and demons. The ancient Babylonians predicted future events from the birth of deformed children. As far back as 3000 BC achondroplasia was depicted by the Egyptians in paintings. Hippocrates (460-377BC) the father of medicine, has given us first description of hydrocephalus. Emppedocles of Acorages(483-424BC) and Democritus(460-371BC) speculated that maternal impressions were responsible for abnormal development.

Harvey introduced the word "teratology" together with Meckel (1781-1833) Saint Hilaire advanced the concept of developmental arrest which explained the fact that in many congenital abnormalities such as cleft palate, the development appeared to have stopped at some state and the primitive features of that period were retained.

Mendel's work and his law of inheritance, published in 1865, exerted considerable influence on the speculation regarding the causation of malformations. For the first time, a scientific explanation, that of hereditary mechanisms, provided an apparently satisfactory explanation of the occurrence of development defects . From the end of the 18th century , morphological studies, particularly those of an embryological nature, eventually provided a scientific basis for the investigation of abnormal development.

Numerous publications describing an almost infinite variety of malformations appeared (Pannum 1860 , Chance 1862, Foster 1865, Ahifeld 1880-82, Taruffi 1881-94). Development of the science of genetics also contributed to the concern about etiology. Irradiation experiments of the first few decades ( Bagy 1992 , Muller 1927, Speciannil 1938 together with the experiments of the 1940's including those of Wolff's[36,48], Ancel 1950 and Landaner[54] in the chick and nutrition experiments in mammals Warkany[15] and Giround[60]). The observations on the teratogenicity of the rubella virus by Gregg in 1941 opened paths previously untravelled in the study of teratology.

**THEORIES ON THE ETIOLOGY OF CONGENITAL ANOMALIES**

1. Theory of supernatural causes;(1493), Panacelsus(1493);

2. Early theories of natural causes by Aristotle(1853)

3. Theory of maternal impressions[13](18th century)

4. Hybrid theory;(18th century)

5. Mechanical theories including adnexal disorders- By Pare(1649)

6. Foetal disease theory(1749)

7. Embryological development arrest theory: William Harvey (1655)

8. Genetic theory; By Mendel(1865)

Several achievement of the first decade of the 20th century assured the acceptance of the genetic theory as having a role in malformation. This included Garrod's book on inborn errors of metabolism, Farabee's study of five successive generation of brachydactyly in an American family, defective blastocyte before implantation(Corner '61). Although some environmental teratogens were already known, Murphy 47 still emphasized, in his book "Congenital malformation" that genetic causes probably account for the majority of the malformations at least in human beings. Nevertheless, genetic cause per se are still a very important area of

consideration and investigation today and their modifying influence of environmental, maternal, or local factors cannot be over emphasized. Mckusick has catalogued the known Mendelian conditions in men.

The development of experimental teratology is related to studies on experimental embryology, an effort in order to study normal development. Hale produced anophthalmia and cleft palate in pigs by long term depletion of Vitamin A in 1940 's and 1950 's. Many other experiments included use of physical agents (irradiation , temperature change, hypoxia) hormone, nutritional deficiencies and excesses . During the last 25 years numerous publications of experimental teratology and of many clinical genetic studies the elucidation of the multiple and inter twined causes of human malformation is still in early state . Nevertheless only through such understanding will prevention become a reality.

**FREQUENCY OF CONGENITAL MALFORMATION**

The frequency of congenital malformation in the present study is 30.4 per 1000 birth. This rate in the study is comparable to incidence of congenital malformation in other hospital based studies of live and still births. Analysis of 22,434 births from various part of India in different studies varies from 8.6 – 20.2 per 1000 births. A high frequency of 36 /

1000 was reported from Chandigarh[19], probably due to very high autopsy rates, 37.9 / 1000 in Pondicherry wherein 63 % of neonatal deaths and 80 % of still births were autopsied ,another high frequency of 39.1/1000 in Davangere Karnataka and attributed to high prevalence of consanguinity in that area by Kulkarni and Kurian[41]. In the 2 studies in Mumbai involving live births[20,21] the prevalence rate was 17 and 18.7 per 1000. In a study carried out by the Indian council of Medical Research at various centres the incidence of major malformations were as follows Ajmer 19.1 , Chennai 18.9 , Hyderabad 14 per 1000 as comparable with the present study. A high incidence of 5.5 % has been reported by Singh et al[23]. The relative difference in the incidence of various malformations might be due to geographic and racial difference , in addition to the factors likely to be potentially teratogenic. Study by Verma et al[26] incidence was 3.6% and in the same institution 15 years ago prospective study done showed the frequency of congenital malformations as 2.75%.

**TABLE-1**
**COMPARISON OF INCIDENCE OF MALFORMATIONS WITH OTHER STUDIES**

| Locality | Period | No. of Births | Major Malformation per 1000 | % Among still Birth |
|---|---|---|---|---|
| AJMER | 1969 | 2145 | 19.1 | 20.4 |
| MUMBAI | 1960-63 | 29550 | 14.1 | 9.5 |
| KOLKATTA | 1961-64 | 19191 | 3.1 | 1.6 |
| CALICUT | 1962-64 | 3721 | 13.4 | - |
| CHANDIGARH | 1966 | 1000 | 36 | 9 |
| CHENNAI | 1972-73 | 23000 | 18.9 | - |
| MYSORE | 1967-69 | 5554 | 2.5 | 9 |
| PONDICHERRY | 1963-66 | 11056 | 10.5 | 5.1 |
| VARANASI | 1988 | 3932 | 9 | 6.9 |
| ALIGAR | 1997 | 4375 | 16 | 15.6 |
| NEW DELHI | 1998 | 23367 | 14.6 | 12.29 |
| SHIMLA | 2000 | 10100 | 17.8 | 15.1 |
| JAMMU(J&K) | 2002 | 9308 | 13.9 | 4.96 |
| HYDERABAD | 2004 | 6540 | 14 | 13.3 |
| MUMBAI | 2005 | 17653 | 16 | 15.8 |
| PRESENT STUDY | 2008-09 | 5000 | 20.8 | 10.18 |

Hospital based studies on live birth shows only the mean malformation rate was 18.9 per 1000 which is only marginally lower than studies which included both live and still births.

Population based studies of live births, In the 2, one study by Ghosh et al[26] is of great interest as it involved a prospective survey of a population of 1,00,000 in Delhi. The incidence was 26.2 /1000. A survey of these infants after an interval of years was carried out, wherein 3816 children were re examined. Among these new malformation was detected in 54 children , even though the examination at birth was normal.

On adding the later diagnosed malformations 14.15 per 1000 to those detected at birth, a total frequency 40.37 per 1000 was obtained in the birth cohort. The incidence of major malformations increased from 11.07 to 16.5 per 1000. Most of the increase was accounted for by cardiovascular and central nervous system malformations.

In Brimingham study, in which the children were followed till 5 years of age, the incidence of major malformations increased from 17.3 to 23.1 per 1000 at 5 years.

The study by Kulshrestha et al[28] is also unique , as it involved study of births in rural area of Haryana. Rate of malformation being 34.1 per 1000.

Western studies also shows similar incidence[31]. Shown in table 2.

**TABLE – 2**

| Regional Total | No. of Reports | Births | No. of Malformations | MB/1000 Births |
|---|---|---|---|---|
| BRITAIN & IRELAND | 28 | 8,38,728 | 13,051 | 15.5 |
| EUROPE | 70 | 60,98,585 | 47,447 | 7.7 |
| GERMANY | 45 | 21,54,694 | 21,234 | 9.8 |
| USA & CANADA | 45 | 98,05,792 | 1,22,504 | 12.4 |
| IRAN | 1 | 3529 | 109 | 30.9 |
| TURKEY | 18 | 63,159 | 183 | 2.9 |

## GEOGRAPHIC DISTRIBUTION OF MALFORMATIONS

Even in the same country it is different in each area. It has been suggested that in India there is an East- West difference in the rate of malformations[32]. A close analysis of the table one however reveals that this is unlikely to be true. The study from Kolkotta[33] showing a low incidence of malformations is a gross underestimate, as mentioned earlier. Similarly although one study from Pondicherry showed a malformation rate of 10.5per 1000[40] the other one revealed a rate of 37.9 per 1000. It is therefore possible that the difference in the rate of malformations can be explained by the difference in methodology, accuracy, assiduousness with which the malformations were recorded prospective / retrospective studies , inclusion of minor anomalies, still births, autopsy studies, period of observation and not attributed to any intrinsic differences.

The distribution of malformations in the present study are compared with the other studies in table 3.

**TABLE -3**

**COMPARISION OF DISTRIBUTION OF MALFORMATIONS WITH OTHER WORKERS**

| System Involved | Present Study | Anand | Mathur | Chugh | Ghosh | Kulshreshtra |
|---|---|---|---|---|---|---|
| CNS | 5.6 | 5.5 | 7.5 | 4.2 | 7.0 | 5.2 |
| GIT | 4.6 | 2.0 | 6.5 | 1.6 | 6.0 | 2.5 |
| SKELETAL | 4.0 | 9.0 | 9.0 | 1.2 | 1.4 | 3.6 |
| ORO FACIAL | 2.4 | 8.0 | 3.6 | 1.7 | 2.5 | 1.7 |
| CVS | 2.2 | 1.5 | 1.5 | 2.1 | 1.3 | 2.0 |
| GENITO URINARY | 2.0 | 1.5 | 1.5 | 1.8 | 2.5 | 3.4 |

Central nervous system and Gastro intestinal system including orofacial group were the most commonly involved in the present study. Skeletal system is the third commonest and 4$^{th}$ one is cardiovascular system. Our results are also comparable with other studies showed in table 3. The commonest system involved in other studies were central nervous system. In contrast Mathur et al in a community based study found musculoskeletal anomalies as the commonest. Terry et al found gastro intestinal anomalies to occur more commonly among mothers of Indian origin[37].

In the present study, out of central nervous system malformations, neural tube defects were the commonest 3.2 per /1000. Reports of neural tube defects from various parts of India have given the incidence ranging from 0.5 to 8 / 1000. It has been suggested that Sikha have given a high incidence of neural tube defects and that the high incidence is maintained even after migration to U.K.[51]. Verma and colleagues reported such a high incidence of neural tube defects not only in people of Punjab but also in those of Rajasthan.

It is clear that prevalence of anencephaly and spina bifida is high in Punjab , Delhi, Rajasthan as compared to that in the southern and Eastern states of India . In fact incidence in these states is as high as Ireland, where the highest rate of neural tube defects had been recorded. The exact reason for such difference in rate for neural tube defects in different parts of India are not clear. Some of the likely genetic and environmental factors have been established.

In most parts of the country musculoskeletal disorders are the commonest malformations. It is likely that many of the musculoskeletal disorders are deformations rather than malformations , representing an aberrant form or position of a formed structure ,

occurring due to mechanical forces such as unusual intrauterine pressure, twins, abnormal position. Some of these may be due to abnormalities of the pelvis, secondary to osteomalacia.

**MINOR ANOMALIES**

Minor anomalies are defined as unusual morphologic features that are of no serious medical or cosmetic consequences to the patient. The value of this recognition is that they may serve as indicators, of altered morphogenesis or may constitute valuable clue in the diagnosis of a specific pattern of malformations.

Most of the babies have single minor anomaly, but several minor anomalies in the same individual indicates a more serious problem.

In our study 9.6 per 1000 births (48 cases) had minor anomalies. 5 of these children had birth Asphyxia. Incidence of minor anomalies reported by Anand et al was 4 per 1000 births.

**FETAL FACTORS**

**STILL BORN AND LIVE BORN**

In our study congenital malformations were common in still born (10.18%) as compared to (2.88%) live born. This was found to be statistically significant ($p < 0.05$).

This is in conformity with the other workers observation. Verma et al showed 4 times higher rate of malformations. Almost the same findings had been reported by Goravalingappa et al, Mathur et al[22] and Anand et al. Of the 19 cases of Central Nervous System malformations 2 cases (10.52%) were still born. Higher incidence of congenital anomalies in still born babies 13 % as compared to 3.7 % in live born babies was recorded with study by Khrouf et al.

**SEX OF THE BABY**

In the present study sex distribution of malformation, a male to female ratio was 1.35:1 and found to be statistically significant ($p < 0.001$).

Thirumalikolundu Subrahmanian and Mathur et al reported a male : female ratio of 1.7 : 1 and 2 : 1 respectively[54,29]. The specific distribution of malformations showed the genito urinary anomalies were more common (93.6%) in male while anencephaly was more common among female (80%) babies. Verma et al found same sex distribution in the above 2 systems mentioned, Guha et al have also reported female preponderance for anencephaly though Choudhery et al found more male babies with this defect in their study[45,46]. But

Choudhery et al found equal incidence of NTD in both sexes.

**PREMATURITY AND CONGENITAL MALFORMATIONS**

In the present study incidence of malformations in premature babies was higher (6.31%) than that of term babies (2.91%) and this difference in statistically significant $p < 0.05$.

In a study carried out All India Institute of Medical Sciences, New Delhi the incidence of malformations in preterm babies was 5.3% and in term babies it was 2.6%. Incidence of malformations in preterm babies reported by Khanna et al was 3.8% Dutta et al[34] was 2.4% and Anand et al was 2.54%. Our results are comparable with these studies. In Verma et al series, the rate of malformations in preterm was two fold of that in term similar to that of Goravalingappa and Nashi. Prematurity has less practical significance in countries like India as most of the low birth weight infants are mature by gestation.

**MALFORMATIONS AND CONSANGUNITY**

Present study shows 67.11% malformed babies were born of consanguineous marriages. There is a definite increase in the rate of malformations in the offspring of consanguineous marriages. 32.89 % among non consanguineous, 38.81 % among second degree consanguinity and 28.28 % among 3$^{rd}$ degree consanguinity. This was

corroborated by Kesavan et al[49] in a detailed study of 23,000 births in Chennai showed a definite increase in the rate of malformations in the offspring of consanguineous marriages (1.64%) among non consanguineous, 2.36% among first cousin, 2.18% among maternal uncle niece marriages and 1.86% among distant relatives. Sugunabai et al[44] from Trivandrum who reported that consanguineous parents had 3.59% malformed offspring while non consanguineous parents had only 1.69% offspring with malformations.

In the global study of congenital malformations[40] the rate of neural tube defect was observed to be higher in the offspring of consanguineous marriages in Alexandria and Egypt. However, the data from India on this aspect are equivocal. Two surveys of the general population in South India failed to reveal any significant effect of consanguinity on the rate of malformation[50,51]. One was a planned prospective survey of a populations about 100000 near Vellore which aimed to study the effect of consanguinity on human reproduction.

## MATERNAL FACTORS

### Maternal Age

In the present study, incidence of malformations was marginally higher after 35 years. Sugunabai et al[35] found the incidence to be high in mothers above 35 years as in this study. Tibrewala et al reported an incidence as high as 75% in mothers above 45 years. Kulshreshtha et al on the other hand found a higher incidence between 25-35 years age group. Verma et al found higher incidence of malformations after 30 years.

## PARITY AND CONGENTIAL MALFORMATIONS

2.51% of our cases with congenital malformations were born to primiparous women, as they constitute 66 of total malformed deliveries. It was found that highest incidence of congenital malformations occurred in parity 4 and more (6.41% and 7.69 %) respectively. Kulshrestha et al found the highest incidence in the 5$^{th}$ gravida. Ghosh et al could not establish any correlation with parity. Our results were found to be statistically significant ($p < 0.05$).

## CONGENTIAL MALFORMATIONS AND MULTIPLE PREGNANCIES

8.33 % of Multiple pregnancies resulted in malformed babies compared to normal delivery incidence of 2.98%. 50% the malformations belong to genito urinary systems (such as undescended testes and 2 cases Hypospadias) and 20% cases were CTEV. A statistically increase in incidence ($p < 0.05$) of congenital malformations was observed among twin deliveries.

It has been reported that congenital malformations especially due to uterine compressions, vascular communication with embolization (ileal atresia, porencephaly) etc. are more common in twins[16].

## DRUGS AND CHEMICALS

Any insult during 1st trimester, the period of organogenesis, is likely to result in a malformed baby. The teratogenic effects of certain drugs like thalidomide are well known. In our study 6.29 % of mothers with the malformed babies gave the history of drug intake during the 1st trimester, Verma et al and Nair also observed same incidence of drug intake. Mishra and Baveja reported that 13.3% mothers with malformed babies gave history of drug intake.

Two percent of congenital malformations are due to major environmental factors[50]. Teratology is branch of medicine about teratogenesis. According to the aetiology congenital malformations can be divided into 3 categories[51].

1. Chromosomal origin or single major mutant genes.
2. Environmental effects results in congenital malformations.
3. No identified causes – It is the largest group.

The discoveries were made of teratogenic effects of ionizing irradiation, the rubella virus and drugs aminopterin and thalidomide in 1920, 1941, 1952, 1961 respectively. About other classes of teratogens, maternal disorders and chemicals, much still remains to be learned.

**MAJOR ENVIRONMENTAL CAUSES DURING EARLY PREGNANCY FOR CONGENITAL MALFORMATIONS**

## Maternal Infections

- Rubella
- Treponema palladium
- Cytomegalovirus
- Toxoplasmosis
- Herpes simplex virus type 2
- Varicella
- Coxasakie-B
- Urinary tract infections

## Maternal illness

- Diabetes
- Thyroid disease
- Phenyl ketonuria
- Foetal virilizing disease
- Vaginal bleeding

- Hyperthermia
- Urinary tract infections

## Occupational hazards

- Anaesthesia gases
- Fat solvents
- Hair spray adhesives
- Hydrocarbons

## Drugs of habituation

- Alcohol, Cigarettes
- Coffee, Tea
- Gasoline sniffing
- LSD Marijuana
- Methadone
- Tobacco chewing
- Toluene sniffing

## Pharmaceutical drugs

- Female sex hormones
- Adrenocorticoid, Amphetamine
- Nonquinine antimalarials
- Lithium, oral hypoglycemics
- Diazepam

- Erogonovine
- Meclizine, Warfarin
- Dicoumarol
- Phenytoin
- Trimethadioine
- Primidone

- Aminopterine

- Retinoic acid

## Heavy Metals

- Mercury

- Lead, cadmium

## Food colourings

- Hair dyes

- Nitrates and Nitrites

- Saccharin

- Sodium fluoride

- Trichloro phenoxy acetic acid

## Natural substances

- Blighted potatoes

- Cyanide in cassava

- Goitrogens in Brassica

## Nutrition

- Deficiencies of Vitamin A&E
- Deficiencies of folate, zinc iodine
- Increased levels of Vitamin A&D

**MATERNAL ILLNESS**

**TORCH Infections**

In the years since the discovery that maternal rubella infection can be teratogenic, intensive searches have been conducted and the present evidence indicates that besides rubella only cytomegalovirus and Toxoplasma gondii unquestionably causes congenital abnormalities. Other agents associated with congenital malformations are given in table No.4. The 1964 epidemic of rubella in United States caused malformations in more than 20,000 children. Since 1969, when the Rubella vaccine was 1$^{st}$ licenced, the disease has greatly abated now.

**HYPERTHERMIA**

In our study observed that 10.94 % of mothers with malformed babies had febrile illness during the 1st trimester. 47.0% of mothers with the history of febrile illness during 1st trimester delivered babies with NTD. Verma et al observed 4.7% mothers had febrile illness. Western studies, retrospective analysis it was found that febrile illness during early weeks or months of pregnancy was associated with maldevelopment[53-56]. Increased frequencies of anencephaly, spina bifida, microphthalmia, unusual facies were reported. Prospective analysis failed to reveal any association. Two recent studies cancelled each other out, a positive one from Japan and a negative one from Finland.

**DIABETES**

In our study 33.33 % of the mothers with malformed babies gave history of diabetes during pregnancy. Studies showed[57,59] that only frank diabetes that exist before the beginning of pregnancy (that is white classes B to F) appeared to be associated with an increased risk of maldevelopment in the conceptus. The frequency with which malformed children were born to such women and recognized as being malformed in the neonatal period ranged in recent studies[63,66] from 6.6–

13% with a mean rate of 9.1% or about 3 times the number of malformed newborn infants found in the general population and they include 1.44% of the total number of such infants as observed on our study.

Among the types of malformations found in infants of diabetic mothers include cardiovascular, neural tube, sacral malformation. Confirmation of recent findings indicating that good metabolic control of maternal diabetes before and during the early stages of pregnancy may be associated with decreased frequency of congenital malformations[60].

**VAGINAL BLEEDING**

The present study observed 2.01 % mothers with malformed babies had vaginal bleeding in early pregnancy. In association between vaginal bleeding in early pregnancy and congenital malformation has been noted in some studies. Verma et al showed 7.8% incidence but not in others. Whether there is such an association or not, it is unlikely that the relation is casual as has been claimed because early vaginal bleeding is frequently an indication of threatened abortion and because spontaneously aborted foetuses are frequently malformed[52].

In the present study malformations were higher in cases with complications of pregnancy like hydramnios and toxaemia of pregnancy. 31.81% (7 cases) of mothers with malformed babies and hydramnios - 2 babies had trachea - oesophageal fistula, 2 babies had hydrocephalus and one had anencephaly. Correlation with these conditions are known. Kalra et al[61] also found correlation of hydramnios with anencephaly, meningomyelocele, exomphalos and trachea-oesophageal fistula. 6.52 % of mothers with malformed babies had pregnancy induced hypertension.

**ANTENATAL DIAGNOSIS**

Of the 16 cases of neural tube defects 12 cases were diagnosed by using ultrasonogram. But all the cases diagnosis were made at late. The reason for this late scanning were late bookings and low index of suspicion.

4 babies with neural tube defects delivered by emergency LSCS as these cases came in late labour stages and ultrasonogram could not be performed. Early antenatal diagnosis and timely termination of pregnancy is desirable. Ultra sound is an invaluable tool in the diagnosis of neural tube defects, genitourinary abnormalities cardiovascular abnormalities etc. For the diagnosis of Neural tube defects an examination should be performed by about 16 weeks. A high

index of suspicion combination with efforts diagnosis for early booking and early scanning should lead to early diagnosis and timely termination so as to avoid the anguish of giving birth to a malformed baby.

**ROLE OF AUTOPSY IN STUDY OF CONGENTIAL MALFORMATION**

Just as increasing survival of immature foetuses and the accompanying therapeutic interventions have emphasized the importance of neonatal autopsies, as the growing scope of prenatal diagnosis and screening makes the examination of the abortus and small foetuses essential. Various studies[62] have suggested that over 43.70% human conceptions fails. Comparison of the same anomaly in abortuses and term foetuses confirms that the body is very difficult at getting rid of abnormal conceptuses for example more than 90% of cases Turner's syndrome, NTD were eliminated. Morphology of congenital defects should be examined soon after morphological changes begin i.e., beginning from embryo stage itself.

External examination of the foetus is often valuable and photographs may be important for comparative purposes. X ray gives valuable data as in cases of osteogenesis imperfecta and other skeletal abnormalities.

Histological examination will confirm some suspected defects eg. Renal dysplasia. Examination of the heart needs expert pathologist opinion and histopathological examination as in case of PDA. It was found that studies in which autopsies were done in more cases showed 3-6 times higher prevalence of congenital malformations than one in which there were fewer autopsies.

**PERINATAL MORTALITY AND CONGENITAL MALFORMATIONS**

Congenital malformations were responsible for 12.3% perinatal deaths in the present study and fifth as the cause of perinatal mortality after prematurity, birth asphyxia, septicaemia and respiratory distress. Chopra et al and singh et al[73,75] have attributed to 17.8% and 15.7% of perinatal deaths to congenital malformations.

Congenital malformations compromises 8% of the perinatal mortality in India as estimated from the data available from hospital based studies[76] and fifth as a cause of perinatal mortality after asphyxia, respiratory problems, infections and birth trauma. This pattern is changing rapidly with improvement in health care and living standards and a recent study from Hyderabad[77] congenital malformations contributed to 17.8% of perinatal mortality being the second commonest cause.

# SUMMARY AND CONCLUSION

1) Incidence of congenital malformations in the present study is 30.4 per 1000 live births, of which major malformations were 20.8 per 1000 and minor malformations were 9.6 per 1000.

2) The most common system involved in the study is Central Nervous System (5.6 per 1000).

3) The second commonest system involved in the present study is GIT (4.6 per 1000).

4) The incidence of malformations were marginally higher in babies born to mothers over the age of 35 years and mothers with parity 4 and above 5.46% and 14% respectively.

5) Congenital malformations especially neural tube defects were common in babies born of second degree consanguineous marriage. (Significant contribution of second degree consanguinity was noted in all major malformations 38.81%).

6) The incidence of malformations were higher in cases with complications of pregnancy like diabetes 33.33% and hydramnios31.81% and PIH 6.52%

7) Incidence of malformation were higher in preterm babies 6.31%

8) Incidence of malformations were higher in male babies, especially genitourinary system anomalies.

9) Incidence of malformations were higher in still born babies and in twins 10.18% and 8.33% respectively.

10) Antenatal events in the 1st trimester like fever, drug intake could be implicated in aetiology of malformations especially neural tube defects in our study.

11) Prenatal diagnosis using ultrasound is accurate, but performed late. So, a high index of suspicion and early scanning in high risk mothers are necessary.

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# PROFORMA

## CONGENITAL MALFORMATIONS IN THE NEWBORN

Serial no :

Mother's Name : Age :

Father's Name : Age :

Address : Income :

D.O.A. :

D.O.B. :

D.O.D. :

LMP :

EDD :

Booked / Unbooked :

Immunised / Unimmunised :

IFA : Regular / Irregular

Consanguinity : I / II / III / No

Family H/o Congenital Malformation: Yes / No

Parity : G P L A

**Antenatal History :**

| | I Trimester | II trimester | III trimester |
|---|---|---|---|
| Drugs | | | |
| Fever | | | |
| Radiation | | | |
| Vaginal bleeding | | | |
| Diabetes | | | |
| PIH | | | |

H/o of Oligohydramnios / Polyhydramnios / Heart Diseases / Other

illnesses

**Investigations** :

Blood group & Rh :
VDRL :
HIV :
Hb % :
Urine deposits / sugar :
USG :

**Perinatal period :**

H/o of Foetal distress :
Mode of delivery : Spontaneous / Induced / Caesarean / Forceps / Vacuum / Breech

Foetus : Single / Multiple
Male / Female
Live / Still birth
Pre Term / Term / Post Term
AGA / SGA / LGA

Umbilical Cord :
Placenta :
Anthropometry :
Birth Weight :
Length :
Head circum :
Chest circum :
US : LS ratio :
MAC :

**EXAMINATION :**

HEAD : Microcephaly / Hydrocephalus / Anencephaly / Encephalocele/Craniosynostosis

SPINE : Meningocele / Meningomyelocele / Spina bifida

EYES : Microphthalmia / Anophthalmia / Cataract / Aniridia / Coloboma

EARS : Microtia / Anotia / Preauricular sinus / Skin tag

GIT : Cleft – lip/ Cleft palate / Tracheo-Oesophageal fistula / Duodenal Atresia / Ano – rectal malformations/Exomphalos / Hernia / Megacolon

| | | |
|---|---|---|
| CVS | : | Cyanotic / Acyanotic Heart Disease |
| RS | : | Choanal Atresia / Diaphragmatic Hernia / Others |
| SKELETAL | : | CTEV /CDH/Poly Syndactyly /Achondroplasia/Osteogenesis Imperfecta / Over – riding of toes |
| GENITO URINARY | : | Hypospadias / Epispadias / Hydrocele / Undescended testes / Ambiguous Genitalia / PUV / Hydronephrosis /Megaureters |

Miscellaneous :

Investigations :

Consent for autopsy :