AN EPIDEMIOLOGICAL STUDY OF CONGENITAL MALFORMATIONS IN RURAL CHILDREN

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ABSTRACT

Children between 0-6 years of age from six villages of Ambala District were screened for congenital malformations. Of 1371 children, malformations were observed in 30 (prevalence 22/1000). Twenty children had major malformations and six had multiple anomalies. Cardiovascular malformations were the commonest (37%) followed by musculoskeletal (30%), gastrointestinal (23%), central nervous system (13%) and genitourinary anomalies (6.6%). An etiological factor (maternal rubella infection or drug exposure during early pregnancy) could be ascertained in only 3 cases. Traditional birth attendents (TBA) and Anganwadi workers (AWW) were helpful in identifying 95% of the cases with externally visible malformations in rural areas. In majority of cases no remedial measures were taken by the parents.

Key words: *Congenital malformations, Epidemiology.*

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Over a period of time with improvement in perinatal care, immunization status(l) and better control of diarrheal and respiratory diseases, it has been possible to bring down the mortality in infants and cbildren(2,3). As a result of these changes the proportion of cases with congenital malformations is likely to increase. Information on such diseases is needed for planning maternal health services and also to monitor the environmental hazards associated with mutation(4). Most of the studies on congenital malformations are hospital based studies and represent mainly an urban or semiurban population. Little information is available on congenital malformations from rural population(5). Due to lack of facilities even for routine medical examination by a qualified doctor in rural areas, a large number of cases with malformations either die undetected or present with mental or physical handicap later in life(6). This study was undertaken to determine the frequency and type of congenital malformation in children, their relation with genetic and environmental factors, remedial measures taken by the parents and suggest ways to detect them at an early age, so that appropriate remedial measure can be undertaken.

Material and Methods

Children between the age of 0-6 years from six villages of Ambala District were screened over a period of two months for congenital malformations. Regular weekly clinics were conducted in all these villages by the Department of Community Medicine, PGIMER, Chandigarh. For the purposes of this study, a pretested proforma was used with details on sociodemographic, heredo-familial, environmental factors, nature of malformations and remedial measures taken by the parents. To collect information regarding children with malfor-

mations, Anganwadi workers (HW) and medical officer (MO) of the respective area were contacted by the chief investigator. In addition, all other children attending the Anganwadi centres (AWC) were examined by the chief investigator (VK). House to house visits were made to cover the younger children and those who were not available at AWCs. However, no repeat visits could be made to cover the children who were not available at the time of initial visits. Each child with a malformation was subjected to a detailed clinical evaluation and the findings were recorded.; Information about, the persons who first noticed the malformation and those who served as informant to the investigators, was also recorded. To confirm a clinical suspicion of some malformations, appropriate investigations were also done.

The malformations which had a considerable effect on normal functioning of all or part of the body were considered as major while those having no serious medical or cosmetic consequences were regarded as minor(7). Some of the minor defects like hydrocele, *cafe-au-lait* spot, or discrete isolated -hemangioma were not recorded. The data was analyzed using simple frequency table and "Chi square" test in order to find out the significance of variables.

Results

The study covered a total population of 19260 from six villages. As per the information from our earlier ICDS survey(8,9) in this area 1900-2100 (15-16%) children in 0-6 years of age were expected in the study population. However, during our AWC/ house visits we could screen a total of 1371 (10.6%) children for congenital malformations. Since no repeat visits were made, about one-third of the children could not be included in the study.

Thirty children had one or more congenital anomalies. Twenty of them had major and 10 had minor malformations. Their ages ranged from 15 days to 6 years and 17 of them were male and 13 females (M : F = 1.3 : 1). *Fig. 1* shows the age and sex distribution of the affected children. Those who were older than 3 years, showed a female dominance while in younger age group, males were affected more frequently ($p < 0.05$).

Malformations involving cardiovascular system were the commonest (37%), followed by musculoskeletal (30%), gastrointestinal (23%) central nervous system (13%) and genitourinary system (6.6%) (*Table I).* Six cases had multiple anomalies and two of them had features consistent with a diagnosis of congenital rubella syndrome and trisomy 13, respectively.

Six cases with malformations were identified at birth and another two (6.6%) within 24 hours. By one year of age it was possible to detect 60% (18) of the cases, while rest 40%(12) of the cases were detected after one year of age. During the present survey, while searching for the cases with malformation, information on 18 cases was

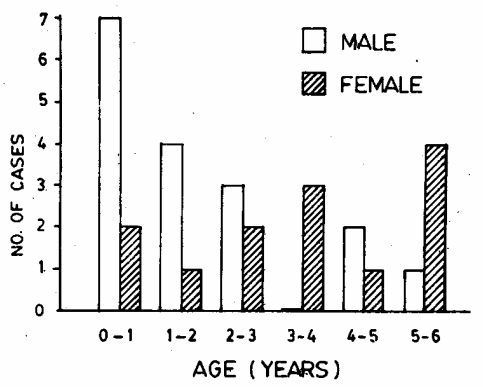

Fig. 1. Age and sex distribution of patients with malformations.

TABLE I-*Congenital Malformations in Children*

Malformations	No. of cases	ICD* Code(17)
Cardiovascular system		745-747
Acyanotic heart disease	9	
Cyanotic heart disease	2	
Musculo-skeletal system		
Polydactaly	2	755.0
Syndactaly	1	755.1
Hypoplasia depressor anguli oris	2	744.8
Talipes equinovarus	1	754.5
Pectus excavatum	1	754.8
Chondrodysplasia	1	756.5
Spondylolysis	1	756.1
Gastrointestinal system		
Cleft lip	2	749.1
Clef palate	1	749.0
Cleft lip and palate	1	749.2
Inguinal hernia	1	550.9
Umbilical hernia	2	553.1
Central nervous system		
Spina bifida occulta	1	756.1
Meningocele	1	741.9
Squint	1	378.9
Microcephaly	1	742.1
Genitourinary system		
Hypospadias	2	752.6
Multiple anomaly syndromes		
Rubella syndrome	1	771.0
Trisomy 13	1	758.1

ICD* - International classification of diseases.

provided by AWW, TBA, HW or parents. The remaining 12 cases were detected on examination by the chief investigator during home/AWC visits. Among all the workers, AWW played an important role of key informant in 47% of the cases. In 5 cases parents were the first one to notice that their child had malformation, while in 7 cases TBAs noticed the defect first and in 11 cases it was the investigator who identified and declared that the child had a malformation.

Maternal Factors

The age of the mothers of the affected children at the time of conception varied from 18-30 years. The maximum number of malformed children belonged to the mothers in 20-25 years of age group. About 37% of the malformations were noted during first pregnancy and showed a gradually decreasing trend for subsequent pregnancies. None of the mothers reported exposure to smoking, tobacco, alcohol, irradiation or trauma. However, 5 mothers had history of fever during early pregnancy and one of them turned out to have rubella infection. One mother was on some herbal medicine to assist conception, while in another an abortion had been tried with some drugs. Two of the mothers had history of previous abortion. A history of unexplained death at day 2 of life of an apparently normal baby was elicited in another. None of the mothers had a history suggestive of antepartum hemorrhage (APH), polyhydramnios or eclampsia.

Only 8 of 20 children with major malformations were taken to the hospitals. Five of them had congenital heart disease, two had cleft palate with feeding problems and one had meningocele. None of the children with minor malformations received any medical attention.

Discussion

The reported prevalence of congenital malformations in different studies shows a marked variability depending upon the nature of the study, inclusion of minor defects and still births and period of follow up(10). Many congenital defects may not be apparent at birth and may manifest later in life(6). In the present study, children between 0 to 6 years of age were taken as denominator (not the total births) and those with malformations as numerator. In most other studies where total number of births were taken as denominator and number of babies with malformations at birth as numerator, the cases who manifest later are likely to be missed. It is however, likely that some of the infants, who died soon after birth or in early part of their life due to incompatible nature of the defect might have been missed in our study. We recorded a prevalence of 22/1000. Which is higher than other hospital based studies(10,ll). This is possibly due to inclusion of older children in whom majority of the malformations would have manifested by that age. This fact is supported by the observation that the proportion of cases with congenital malformations diagnosed at six months of age reached upto 82% as compared to 43% at birth(6). Moreover, in our study an active effort was made to search the cases with malformation in the community and as many as 37% cases were first identified by the investigator himself which could have contributed to a higher prevalence. This also highlights the scope of routine screening of under fives at AWC by the medical officers of the area. Such screening will help in diagnosing cases which are not identifiable by the parents.

In our study, cardiovascular malformations were the commonest compared to NCS and GIT anomalies reported from

other studies(4,10). Though an earlier study(12) from the North Indian population has shown much higher prevalence of neural tube defects, in our study these were least common. This possibly indicates that neural tube defects which are relatively less compatible with life, are more likely to be reported in studies including all live and still born babies, or there may be a decline in the occurrence of neural tube defects(13). In our study which is based on the 0-6 years child population, the defects which are relatively compatible with life are likely to predominate.

In agreement with other observations(4, 10), majority of the cases with malformations were noted in the firstborn baby and showed a decline with increasing birth order. In concordance with earlier observations(ll) we were unable to record any maternal complication or etiological factors in most cases. However, in a few cases there was an association with fever or drug ingestion during early pregnancy. Heredofamilial factors and consanguineous marriages have also been found to have an important role in causation of malformation(14,16), but in our study none of the children with malformations was born of a consanguineous marriage.

TBA and AWW played an important role in identification of cases with malformations from rural area, as almost 95% of the externally visible malformations were identified and reported by these workers to the investigator. Thus these workers can be relied upon as key informants for any programme on identification of such children, as they are involved in care of the newborns and toddlers in the rural areas. However, it is necessary to educate not only the TBA and AWW but the parents also regarding the nature and long term sequelae of malformations and the availability of remedial measures for many of these defects. This will not only help them in detecting anomalies at a younger age **but** also to assist in genetic counselling and in providing intervention at an appropriate time.

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