

THE INCIDENCE OF INHERITED MALFORMATIONS AND ITS ALLIANCE WITH CONSANGUINEOUS MARRIAGES IN A TRIBAL MEDICAL COLLEGE SNCU, RIMS, ADILABAD

Suryakant Sayanna Mundlod¹, Smita Marotirao Thakkarwad²

¹Associate Professor, Department of Paediatrics, Rajiv Gandhi Institute of Medical College, Adilabad.

²Associate Professor, Department of Obstetrics and Gynaecology, Rajiv Gandhi Institute of Medical College, Adilabad.

ABSTRACT

BACKGROUND

Inherited anomalies are a chief cause of stillbirths and neonatal fatality rates. The pattern and occurrence of innate anomalies might vary over time or with geological location.

The aim of this study is to work out the proportion and kind of inherited anomalies in live newborns and to review maternal and perinatal risk factors and its correlation blood kinship.

MATERIALS AND METHODS

In this prospective hospital-based study, all the live born babies admitted in SNCU, throughout a 1-year duration of Jan 2017 to Dec 2017. The newborns were examined for the presence of congenital anomalies and mothers were interviewed for consanguineous marriages.

RESULTS

During the study period, 1486 babies were admitted in SNCU; of those, twenty-two had congenital malformations, creating the prevalence 1.5%. Most of the ladies (62.7%) belonged to the age group between 21 and 30 years. Congenital anomalies were seen more predominately (2.8%) in the multiparas as compared comparison with primiparas (1.6%). The predominant system concern was gastro-intestinal (GI) system (27%). Musculoskeletal system (18.2%) followed by CHD (18.1%). Out of 1486 neonates, 401 (27%) were from kin marriages and 1085 (73%) were from non-familial marriages.

CONCLUSION

Public awareness regarding preventable risk factors is to be created and early prenatal diagnosis and management of common anomalies is powerfully recommended.

KEYWORDS

Birth Anomaly, prematurity, prevalence, risk factors, types.

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BACKGROUND

Congenital malformation (CM) means any abnormality, genetically or otherwise, that is present at birth. To be more precise, it is an abnormality of physical structure that is seen at birth or within few weeks after birth.¹ According to World Health Organisation (WHO) document of 1972, the term congenital malformation should be confined to structural defects present at birth. CM may be minor or major. Minor malformation is defined as structural abnormality present at birth which has minimal effect on clinical function, but may have a cosmetic effect e.g. preauricular tag. Major malformation has a significant effect on function or on social acceptability e.g. ventricular septal defect (VSD) and cleft

lip.² Initially, malformations can be categorised into three groups:² Single malformation; Multiple malformations, recognisable pattern (syndrome); and Multiple malformations, pattern not recognisable. Congenital anomalies are an important cause of neonatal mortality both in developed and developing countries. It accounts for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.^{3,4} It is not only a leading cause of fetal loss, but also contributes significantly to preterm birth, childhood and adult morbidity along with considerable repercussion on the mothers and their families.

Consanguineous marriages have been described as an important factor contributing to increased congenital malformations.⁵ A consanguineous marriage can be characterized by the degree of relatedness between the spouses: first cousins, double first cousins, half first cousins, first cousins once removed, second cousins, second cousins once removed and third cousins. Genetic effects of consanguinity can be traced to the fact that the inbred individual may carry two copies of a gene that was present in a single copy in the common ancestor of his/her consanguineous parents. A recessive gene may thus come

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Corresponding Author:

Dr. Smita Thakkarwad,

Department of Obstetrics and Gynaecology,

Rajiv Gandhi Institute of Medical Sciences,

Adilabad – 504001, Telangana.

E-mail: dr_surya@rediffmail.com

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to light for the first time in an inbred descendant after having remained hidden for generations. For this reason, consanguinity influences the incidence of some inherited diseases.⁶

MATERIALS AND METHODS

This prospective hospital-based study all the live born babies admitted in SNCU during a 1-year period of January 2017 to December 2017. This teaching hospital has a rate of more than 6000 deliveries annually and both high-risk and normal obstetric cases are treated. All neonates from newborn to 28 days of age admitted to the neonatal unit were included. The study was regardless of gender, gestational age, weight, race, ethnicity, geographical distribution and socioeconomic status. (Table 1)

Meticulous neonatal examination for neonatal care and detection of any kind of CM was done by the sncu paediatrician at the time of admission. Mother's history about consanguine marriage were taken by gynaecologist. Necessary radiological, haematological and genetic investigations were done where required. Ultrasonography and radiological investigations were performed by the hospital radiologist to detect and rule out multiple internal anomalies, where it was considered necessary. History of CM in other offspring and members of their family, and parental consanguinity were obtained by interviewing the neonates' mothers by gynaecologist. Data was obtained from SNCU software.

The second part was about neonatal characteristics including sex, existence of congenital malformation and the type of malformation. The type of birth defects was classified by the diagnostic standardization of CM from the international classification of disease (ICD-10) codes. The data was analyzed using SPSS version 13. The rates of malformed newborns and malformations were compared using statistical T-test and the Chi-square tests. The level of significance was determined at $p < 0.05$.

RESULTS

The mean maternal and paternal age was observed to be 22.86 ± 5.4 and 27.07 ± 9.6 respectively. There were a total of 6508 deliveries during the study period, out of which, 2815 had delivered via C-Section.

From 1486, neonates, 401 (27%) were from consanguineous marriages and 1085 (73%) were from non-familial marriages. In this study, the most frequent type of marriage was between first cousins ($n=256$). In 25 (8.3%) cases, the marriage was between double first cousins and in 19 (6.3%) of the cases, the marriages was between half first cousins,

Of the 1486 births, 1465 (98.6%) were single births, 29 (1.5%) were twin births and only 1 (0.1%) were multiple births. The mean number of gravid was 2.08 ± 1.3 and parity was 1.85 ± 1.09 . 35 (2.4%) and 3 (0.2%) of the women had experienced stillbirth once and twice respectively.

During the 1 year period, 1486 newborns were delivered, among whom 867 (58.4%) were males, 615 (41.4%) females. Out of these, 22 newborns were

diagnosed with congenital malformations. The prevalence of CM in this sample was 1.60% (12 males, 10 females).

In this study congenital malformation in consanguine marriage is 68% (14 cases) and Non-consanguine 32% (07) may be because of geographical tribal area with early marriage. The prevalence of CM in this sample was may be because of less exposure (air pollution created by factories or companies in the cities, and carbon monoxide from cars can produce a large percentage of these pollutants), Table 2.

DISCUSSION

Most children who are born with major congenital anomalies and survive infancy are affected physically, mentally or socially and can be at increased risk of morbidity due to various health disorders.⁷ While the prevalence of congenital anomalies at birth in developed countries is reported to be between 3-5%, those reported in Taiwan are said to be approximately 4.3%, 7.92% reported for the United Arab Emirates, 2.46% for Oman, 2.7% for Bahrain, and 3.6% for India.^{8,9}

However, studies that investigated the number of all infants who were born with a congenital anomaly in some cities of Iran such as Tehran and Gorgan found a birth prevalence of 2.3% and 1.01% respectively.¹⁰ Similarly, the results from this study showed that the overall prevalence of congenital malformation among the newborns who were born in Adilabad was 1.6%. which was similar to Tehran study.¹⁰

The frequency of malformations in this study lower compared with other studies which have been conducted in Iran, this may be because of poverty and environmental tribal locality, (air pollution created by factories or companies in the cities, and carbon monoxide from cars can produce a large percentage of these pollutants) and also low economic status of some individuals, some pregnant women cannot get necessary vitamins during their pregnancy.

In this study, the gender of the fetus did not affect the prevalence of CM, and both genders were nearly equally distributed. These findings are consistent with Karbasi et al.¹¹ However, the results from this study were in contrast to those reported by Gorgan, where male newborns were more affected than females.¹²

Increased incidence of genetic malformations in the offspring of consanguineous couples most likely arises from the homozygous expression of recessive genes inherited from their common ancestors.⁸

In this study from 1486, 564 (38%) of the newborns were from consanguineous marriages, while 921 (62%) were from nonconsanguineous marriages. Also, the rate of malformation was 1% and 0.6% in consanguineous and non-consanguineous marriages respectively. Although, the prevalence of anomalies was higher in consanguineous marriages than non-consanguineous marriages. The results are in agreement with results from the study by Movahedian, and are in contrast with the results from the study by Nath et al.¹³

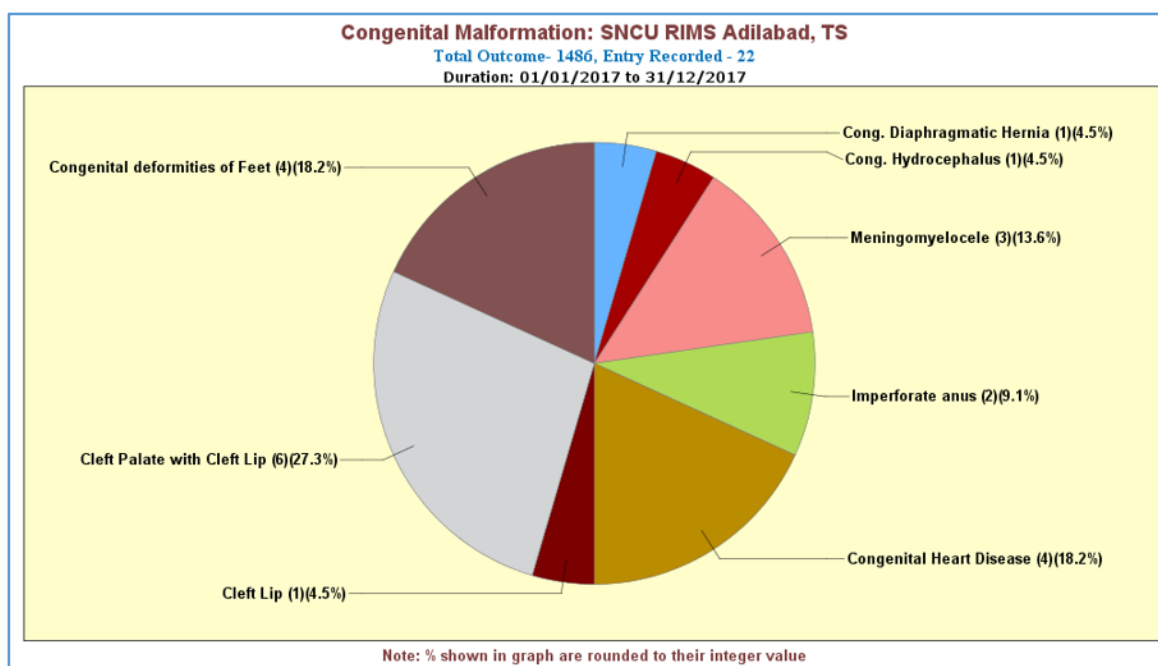
Mehrabi et al. showed that although the consanguinity for malformed patients was high, there was no significant relationship between malformation and the degree of relation of the parents.¹⁴ Also, in a study by Bromiker in Palestine, no statistically significant difference was found in the incidence of congenital malformation with the degrees of parents' relation.¹⁰

Age group	No. of Cases	%
19 years and below (teens)	196	13.2
20-25 Years	927	62.4
26-30 Years	329	22.2
31-35 Years	31	2.1
Above 35 Years	13	0.9
Parity		
Primipara	772	52.3
Multipara (G2-G4)	699	47.1
Grand multipara (G5+)	09	0.6

Table 1. Demographic Analysis of Patients

	Total	Consanguine	Non Consanguine
Cleft lip + cleft palate	7(29%)	4(18.2%)	3(13.6%)
Club foot /foot deformity	4(18%)	3(13.6)	1(4.5%)
Congenital heart disease	4(18%)	2(9.1%)	2(9.1%)
Meningomyelocele	3(13.6%)	2(9.1%)	1(4.5%)
Imperforated anus	2(9.1%)	1(4.5%)	1(4.5%)
Congenital diaphragmatic hernia	1(2.2)	1(4.5%)	00
Congenital hydrocephalus	1(2.2)	1(4.5%)	00

Table 2. The Prevalence of Congenital Malformation in Consanguine and Non Consanguine Marriage



Graph 1

CONCLUSION

In conclusion, consanguinity may play an important role in the high rates of malformations in children, and must be taken into account for genetic counselling in our study, in spite of high rate of consanguineous marriage, the rate of CM is low as compared to other studies.

For possible prevention, genetic counselling before marriage must be applied, not only for consanguineous couples but also for any couples that may have a family history of genetic disorders. Of course, currently suitable

pre-marriage counselling services are provided by the ministry of Health, but there is still room for improvement.

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