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A Study on Congenital Malformations in Fetuses of Mothers with Consanguineous Marriages

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Conflicts of Interest: Nil.

Abstract

Aims and Objectives

Assess the association of the congenital malformations with consanguinity of the parents &correlate the association of consanguinity with the occurance of congenital malformations,

Materials and Methods

Fetuses of pregnant women attending government hospitals in Hyderabad during the a period of one year from October 2011 to September 2012.They were screened through ultrasonographic evaluation in the second trimester (12wks to 28wks) for congenital anomalies

Results

The congenital anomalies more commonly affected the foetuses of mother with a history of consanguineous marriage.

Conclusion

Congenital anomalies are significantly seen among fetuses of consanguineous couples & to prevent them more focus should be laid on maternal education, pre-marital councelling,antenatal care, supplementation with folic acid, prenatal ultrasonography & genetic studies in at-risk individuals.

Keywords: congenital anomalies, consanguinity, prenatal ultrasonography.

Introduction

Though infections and malnutrition are the dominant causes of infant mortality and morbidity in underdeveloped and developing countries, cancer, accidents & congenital malformations are the causes of infant mortality in developed countries.

A congenital malformation is caused by a complex interaction of genetic and environmental factors. So, it is difficult to prevent congenital malformations but the mortality and morbidity caused by them can be prevented by early detection and proper preventive and curative measures.

With the development of science and with advanced screening techniques, in modern era the task of identifying the causative factors, and early detection of congenital malformations has become easier.

Materials and Methods

The present study was done on congenital malformations occurring in fetuses of pregnant women who attended two Government Maternity Hospitals in Hyderabad for antenatal checkup, during a period of one year from October 2011 to September 2012

Foetuses of all pregnant women of both consanguineous & non-consanguineous marriage were screened through ultrasonographic evaluation in the second trimester (12wks to 28wks) for congenital anomalies.

The details regarding the maternal age, antenatal history and other risk factors were taken & recorded as per proforma .Informed consent was obtained from the parents and the data collection was carried out in the vernacular language of the parents.

Results

In our study, a total of 112 cases of congenital malformations were observed. These were further classified according to their family history regarding the

consanguineous status of the mother and the findings were tabulated

Consanguinity of parents plays a major role in causation of congenital malformations. The risk of miscarriages and birth defects are more in consanguineous couples. The risk is even higher in a closer relation.

Table 1:Distribution of cases based on Consanguinity

Consanguinity	No of cases	%
Consanguinous	67	59.82
Non consanguinous	45	40.17
Total	112	100

In our study 67 cases (59.82%) were born to consanguineous couples and 45 cases (40.17%) to non-consanguineous marriages (Table-1).

Table 2: Distribution of cases based on Type of Consanguinity.

Type of Relation	No of cases	%
Uncle – niece	37	33.03
Cousin – I	19	16.96
Cousin – II	9	8.03
Cousin – III	2	1.78
Total	67	100

Out of 67 cases, 37 cases (33.03%) were born to uncle-niece relationship and 19 cases (16.96%) in first-cousin groups (Table-2)

Discussion

Parental consanguinity has deleterious effect on fetal growth and increases the risk of congenital malformations and fetal loss. 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.

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 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.

Inbreeding can occur in a large population as a form of nonrandom mating when the frequency of consanguineous matings is higher than that expected by chance. In this case, the population will show a homozygote excess with respect to a random mating population in which genotypic frequencies are expected to be in Hardy-Weinberg equilibrium.

In ***1976, Gustavson et.al1*** reported many of the congenital malformations in infants dying in the perinatal period have a genetic basis. Some of these represent inherited malformations with a high risk of recurrence in subsequent siblings. ***Sibert et.al*** [2] in ***1979*** found low birth weight babies born to consanguineous than non-consanguineous couples.

A prospective study, in a rural area showed the effects of inbreeding on the incidence of congenital anomalies. The earlier birth orders showed a higher incidence of congenital malformations among the consanguineous compared to non consanguineous marriages ***(Rao et al)***[3]

A survey conducted in seventeen hospitals in Bangalore found that the level of inbreeding in Karnataka population is on a par with that reported for Andhra Pradesh and Tamil Nadu ***(Rama Devi et al)***[4]

A higher incidence of malformations associated such as consanguinity was seen with a marked increase in frequency of cutaneous and neural tube defects among consanguineous marriages ***(Sugunabai et al)***[5]

Malformations of major systems were significantly more frequent among consanguineous couples where as malformations of eyes, ears and skin did not show any significant effect of consanguinity ***(Kulkarni. et.al)***[6]

First cousin marriages may be a significant risk factor for specific type of congenital heart diseases in a consanguineous population (***Becker et al)***[7]**.**Consanguinous couples who already have an affected child are 13 times more likely to have another affected child (***Bagga et al)***[8]

The effect of consanguinity on chromosomal abnormality (structural or numerical) was significant whereas the effect was not significant for the type of chromosomal abnormality (***Amudha et al***)[9]

In a cross sectional study done by ***Tayebi et al***[10] ***in 2010***, the rate of malformations was 2.8% & 0.9% in consanguineous & non consanguineous marriages respectively. There was increased frequency of miscarriages among consanguineous marriages.

Table 3: Comparative Study of Consanguinity In Relation To Congenital Malformations.

S.No.	STUDY GROUP	CONSANGUINOUS	NON CONSANGUINOUS	TOTAL	% OF CONSANGUINITY
1	*Stevenson et al*[11]	52	288	340	25.29
2	*Kulkarni et al*[6]	80	66	146	54.79
3	*Bagga et al*[8]	349	451	800	43.60
4	*Neelu Desai et al*[12]	15	60	75	20
5	*Jahangir et al*[13]	10	8	18	55.55
6	*Tayebi et al*[10]	34	11	45	75.55
7	*Present study*	67	45	112	59.82

A survey of a large series of consecutive births in 24 centres around the world showed that the overall frequency of consanguinity was 3.7% and varied from 30% in Alexandria to less than 0.1% in Zangreb.

In our study out of total 112 cases, malformations are seen majorly in consanguineous couples 59.82% (67/112) than in non-consanguinous couples 40.17% (45/112)

These reports were consistent with the extensive study of **Kulkarni et al**.[6] & **Jahangir et al**[13]**.** (**Table 3**)

WHO studies in Bombay and Alexandria showed the frequency of congenital malformations was significantly higher in offsprings of first cousins and closer relationships than in those related less closely than first cousin.

Thus it seems that closer the family relationships of the parents, the greater the chances of congenital abnormalities.

The term 'Heredofamilial' denotes a condition or disease that may be passed from generation to generation and to several members of on family.For many anomalies, the recurrence risk of a similar malformation in siblings , as well as that in offspring of affected individuals, is relatively high compared with the population frequency, which points toward genetic factors and/or time stable environmental exposures. A history of Down syndrome miscarriage increases the risk of other fetal aneuploidies in subsequent pregnancies **(Bianco et al.** [14]**)**..

Patel and Adhia [15]detected major malformations in 7.92% of 17653 births and were able to attribute chromosomal cause to 4%, polygenic cause to 45.1% and total genetic aetiology to 65.4%.

Conclusions

The present study gave us an idea regarding the association of consanguineous marriage with the occurance of congenital malformations.Most of the aetiological factors remain obscure, but require detailed history taking and thorough investigations for the early diagnosis and treatment.

Parent's awareness about consanguineous marriages and its risk in causing malformations is a preventive factor in congenital malformations and other hazards. (***Mehrabi kushki et al***[16])

There are various confounding factors which effect the results. Some of them are lack of proper history, parents not willing to reveal the health status of siblings, lack of reporting, and unavailability of proper health care facilities. More stress should be laid on prevention by regular antenatal care and avoidance of known

teratogenic agents, maternal education,Premarital counselling, Prenatal ultrasonography at about 8-12 weeks, supplementation of folic acid prior to conception should be given to every pregnant women especially in the embryonic period.

Genetic studies should be made mandatory for all the pregnancies presenting with family history of suspected chromosomal anomalies and in pregnancies of repeated abortions/still births which are highly suggestive of chromosomal aberrations and in such cases prenatal genetic counseling is a must.

References:

[1] Gustavson et al (1976), *Recurrence risks in a consecutive series of congenitally malformed children dying in the perinatal period*. *Clinical Genetics*; 9 :307-314.

[2] Sibert et al (1979), *Fetal growth and parental Consanguinity*. *Archieves of Disease in Childhood*; 54 : 317-319.

[3] Rao et al (1980), *Inbreeding effects on fetal growth and development*. *Journal of Medical Genetics* ; 17 : 27-33.

[4] Radha Rama Devi et al (1982), *Inbreeding in the state of Karnataka, South India*. *Journal of Human Heridity*; 32 : 8-10.

[5] Sugunabai NS., Mary Mascarene, Syamalan et.al (1982), *An Etiological Study of Congenital Malformation In the New Born*. *Indian Journal of Pediatrics;* 19 : 1003-1007.

[6] Kulkarni ML. and Mathew Kurian (1990). *Consanguinity and the effect on fetal growth and development : A South Indian Study*. *Journal of Medical Genetics*; 27 : 348-352.

[7] Susan M. Becker, Zohair et.al (2001), *Consanguinity and Congenital Heart Disease in Saudi Arabia*. *American Journal of Medical Genetics;* 99:8-13 .

[8] Roya Mokhtari, Amrita Bagga (2003), *'Consanguinity, genetic disorders and malformations in the Iranian population'*. *Acta Biologica Szegediensis* Volume 47(1-4):47-50.

[9] S. Amudha, N. Aruna, S. Rajangam (2005), *Consanguinity and chromosomal abnormality*. *Indian Journal of Human Genetics*. May-August; Volume 11 Issue 2:108-110.

[10] Naeimeh Tayebi, et al (2010), *The Prevalence of Congenital Malformations and its Correlation with Consanguineous Marriages*. *OMJ*; 25: 37-40.

[11] Alan C. Stevenson, Harold A. Johnston, patricastewart MI (1966) Congenital Malformations: A report of a study of series of consecutive Births in 24 centres. Bull. WHO; 34: 1-127.

[12] Neelu A Desai, AvinashDesai (2006), Congenital Anomalies : A Prospective Study. Bombay Hospital Journal. July;442-445.

[13] Waqas Jehangir et.al (2009), *Prevalence of Gross Congenital Malformations at Birth in the Neonates in a Tertiary Care Hospital*. *A.P.M.C.* January-June; Vol: 3 No.1:47-50.

[14] Bianco et al (2006), *History of miscarriage and increased incidence of fetal aneuploidy in subsequent pregnancy*. *Obstet. Gynecol*. 107,1098–1102

[15] Z.M. Patel and R.A. Adhia (2005), *Birth Defects Surveillance Study*. *Indian J Pediatr ;* 72 (6) : 489-491.

[16] A. Mehrabi Kushki Msc, B. Zeyghami PhD (2005),*The effect of Consanguineous Marriages on Congenital Malformation.* *Journal of Research in Medical Sciences*; 10(5): 298-301.