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Original Research Article

A STUDY ON CONGENITAL MALFORMATIONS IN FETUSES IN RESPECT TO MATERNAL AGE

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ABSTRACT

Aims and Objectives: Assess the association of the congenital malformations with maternal age & analyse the congenital malformations, involving various systems of the body system wise.

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

Results: The central nervous system involving neural tube defects and musculoskeletal system involvement were the commonest among congenital anomalies affecting elderly maternal age group.

Conclusion: Congenital anomalies are increasing in frequency with increasing maternal age & to prevent them more focus should be laid on maternal education, pre-marital counselling, antenatal care, supplementation with folic acid, prenatal ultrasonography & genetic studies in at-risk individuals.

KEY WORDS: Congenital anomalies, maternal age, maternal education, neural tube defects, prenatal ultrasonography.

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INTRODUCTION

Congenital anomalies are defined as conditions that result in a malformation, deformation or disruption in one or more parts of the body which are present at birth and can have a serious adverse affect on health, development or functional ability of the individual.

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.

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. Congenital malformations not only affects the diseased but also extends to many at risk individuals as well as to their

families and adds to the socio-economic burden of the society.

MATERIALS AND METHODS

The present study was done on congenital malformations occurring in foetuses 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 different maternal ages 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 International Classification of Diseases-10 (ICD-10) and the frequency of various systems involved at different maternal age groups were observed and the findings were tabulated.

Certain congenital anomalies are more frequent in lower maternal age group while others are more common in advanced maternal age group .Thus maternal age is an important factor in the present study.

The pattern of maternal age distribution in all congenital malformed foetuses showed increased occurrence in age group of 41 & above years with a total of 26 cases (23.21%), where as 22 cases (19.64%) belonged to age group between 15-20 years and 10 cases (8.92%) to those between 26-30 years (Table-1).

Table-2 shows system wise distribution of cases of congenital malformations in different maternal age groups. Out of 28 cases of central nervous system, 8 cases were seen in mothers of 15-20 years age group, 7 cases in mothers of 36 to 40 years age group and out of 18 cases of musculoskeletal system, 7 belonged to age group 36 years & above, only 1 case belonged to 15-20 years of maternal age group.

Table 1: Distribution of Cases According To Maternal Age.

Maternal age (Years)	No of cases (Total 112)	%
15-20	22	19.64
21-25	14	12.5
26 -30	10	8.92
31-35	16	14.28
36-40	24	21.42
> 40	26	23.21

Table 2: System wise Distribution of cases based on Maternal Age.

System	Maternal age (Years)					
	15-20	21-25	26-30	31-35	36-40	> 40
CNS(28)	8	2	2	4	7	5
MS(18)	1	4	2	4	3	4
GUS(13)	2	1	1	2	2	5
GIT(13)	1	3	1	2	3	3
Resp Sys(10)	4	2	1	1	0	2
CVS(14)	3	1	1	1	6	2
Multiple Sys(16)	3	1	2	2	3	5
Total(112)	22	14	10	16	24	26

DISCUSSION

Assessment of the incidence of congenital defects is very difficult due to early abortions, stillbirths, prematurity, neonatal deaths, and late manifestation of the defects. Therefore the values depend on the selection criteria of the study group. Even the diagnostic tools used in the study forms important criteria. Therefore the case selection criteria should be stringently defined and followed.

In our study, the malformations of central nervous system predominate with 28 cases (25%) followed by musculoskeletal system 18 cases (16.07%) , 14 cases of cardiovascular systems (12.5%), 13 cases (11.60%) each of genitourinary & gastro intestinal systems & 10 cases of respiratory system (8.92%) . Multiple system involvement is seen in 16 cases (14.28%) (Table 3).

Congenital malformations involving central nervous system were reported to be the commonest in the studies done by Fatema et al [1], Padma et al [2], Swain et al [3] followed by musculoskeletal, gastro intestinal and genito urinary systems in some. Neelu Desai [4] found musculoskeletal system as ranking first and Shamim et al [5] have shown Gastrointestinal

Table 3: Comparative Study Showing System Wise Distribution of Cases.

SL NO	AUTHORS	TOT (n)	CNS	MS %	GUS %	GIT %	RS %	CVS %	MULTIPLE %
1	Stevenson et al. 1966 Bombay [13]	340	41.7	22.9	1.8	11.2	0	2.9	0
2	Stevenson et al,1966 Calcutta [13]	59	18.6	37.3	6.8	5.1	0	3.4	0
3	Swain et al,1994 [3]	48	39.5	14.5	10.4	10.4	6.2	8.3	4.1
4	Kook Lee et al,1998 [14]	200	17	10	16.5	17.5	6.2	10.5	25
5	Neelu desai et al,2006 [4]	79	13.9	18.9	11.3	16.4	1.2	16.4	6.3
6	Nafees et al,2006 [7]	134	58.9	6.7	13.4	8.9	-	0.7	-
7	Munim et al,2006 [6]	170	21.1	14.1	14.1	5.8	8.2	16.4	12.3
8	Somsri et al,2009 [15]	316	17.7	9.4	8.5	7.2	1.2	17.1	2.5
9	Sumit Gupta et al,2010	62	41.9	9.6	6.4	11.2	-	14.5	12.9
10	Shamim et al,2010 [5]	57	14	3.5	24.5	43.8	3.5	7	-
11	Fatema et al,2011 [1]	60	46.6	-	23.3	6.6	-	-	11.6
12	Padma et al,2011 [2]	28	42.8	-	25	28.5	-	3.5	-
13	Taksande et al,2010 [12]	179	8.3	20.1	17.3	12.8	1.1	21.2	4.4
14	Present study,2012	112	25	16.07	11.6	11.6	8.92	12.5	14.28

Table 4: Comparative Study of Maternal age and Birth Defects.

SL NO	STUDY GROUP	TOT (n)	Maternal Age (Years)		
			< 21	21-30	> 30
1	Zilfalil et al, 1997 [16]	44	2.30%	56.80%	40.90%
2	Neelu Desai et al, 2006 [4]	79	0%	73.30%	26.70%
3	Taksande et al, 2010 [12]	179	1.10%	90.50%	8.40%
4	Present study, 2012	112	19.64%	21.42%	58.92%

anomalies topping the list .

The findings in our study were consistent with those in Munim et al [6] (21.1%) and much less than in Nafees et al [7] (58.9%), Fatema et al [1] (46.6%), and Padma et al [2] (42.8%).

The explanation for the association of maternal age and chromosomal abnormalities is usually ascribed to biological aging of ova.

The association of advanced maternal age with Down's syndrome is attributed to various chromosomal abnormalities [8]. It is well established that advancing maternal age is associated with subfertility, chromosomal abnormalities, and multiple gestation.

In patients aged 40 years and older, the higher incidence of antepartum complications such as miscarriage, gestational diabetes, placenta previa, and placental abruption have been documented in the literature. The increased incidence of miscarriage is thought to be secondary to the increased risk of chromosomal abnormalities in these pregnancies. The increased risk of gestational diabetes and placenta previa may be secondary to the relation-ship between aging and progressive vascular endothelial damage resulting in reduced structural and functional health of placenta [9].

It has been suggested that increased biological ageing of the ovaries is a major factor for aneuploidy conditions in females -a 'limited oocytes pool' hypothesis [10]. According to this hypothesis, the ageing of the ovary is associated with availability of limited and less optimal oocytes for fertilization.

Trisomy 21 accounts for 95% of Down syndrome cases, with 88% originating from nondisjunction of the mother's egg cell. Several plausible aetiological factors for Down syndrome include advanced maternal age resulting in altered rate of crossing over between closely linked genes, chromosomal translocations and endocrine imbalances, a strong hereditary component in familial cases with reduced mean maternal age, and maternal-fetal genotype-speciûc susceptibility.

Young mothers especially below 15 yrs age are also at increased risk of anomalous babies probably due to impaired "uteroplacental perfusion" of nutrients and higher prevalence of anemia and other micronutrient deficiencies in young primipara [11].

In our study, an increased rate of malformations (51/112) (45.53%) were seen in age group of 36 years & above and lowest 10/112 (8.92%) in 26-30 years age group. Swain et al [3], Neelu Desai [4], and Taksande et al [12] observed that risk

of malformations increased considerably with advanced maternal age (e"30yrs). Whereas, Fatema et al [1] found increased rate (53.33%) in the age group of 25 – 29 years & only 3.33% of the subjects were beyond 35 yrs.

CONCLUSION

The present study gave us an idea regarding the frequency of distribution of congenital anomalies and also its relation with associated maternal age. Most of the aetiological factors remain obscure, but require detailed history taking and thorough investigations for the early diagnosis and treatment.

There are various confounding factors which affect 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 and 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 counselling is a must.

ABBREVIATIONS

CNS - Central NervousSystem
CVS - Cardiovascular System
GIT - Gastro Intestinal System
GUS - Genito Urinary System
MS - Musculoskeletal system
Sl No - Serial Number
Resp - Respiratory
Wks - Weeks
Yrs - Years
Sys - System
Tot - Total

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Conflicts of Interests: None

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