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"A Retrospective Study on Maternal Age and Down Syndrome"

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ABSTARCT : *A retrospective study on Maternal age and Down Syndrome was done The present study was an attempt to examine the occurrence of Down syndrome in relation to the maternal age and also the occurrence of Down syndrome in different maternal age groups with respect to gender and level of intellectual functioning of the child. The purpose of this research study was to find out the occurrence of Down syndrome in relation to maternal conception age. Data were obtained from case records of Down syndrome cases registered in General Services, NIMH (National Institute for the Mentally Handicapped) during January, 2010 to December, 2011. Total number of Down syndrome cases registered during this period was 294 cases. . Data obtained was analyzed by using Statistical Packages for Social Sciences version 17. Statistical analysis used for analyzing the data were: frequencies, percentage, mean and chi-square. Frequencies and percentage were calculated for maternal conception age, gender of the child, level of intellectual functioning of the child, maternal age with respect to gender and maternal age with respect to level of intellectual functioning of the child. Mean maternal age was calculated for the whole cases as well as for the two age groups, i.e. below 35 years and 35 years and above. Chi-square test was used to find out whether there exist a difference in the occurrence of Down syndrome at different maternal age with respect to the child's gender and level of intellectual functioning. The research findings revealed that there is a difference in the maternal age with the occurrence of Down syndrome but no significant difference in the occurrence of Down syndrome in different maternal age groups in relation to gender and intellectual functioning of the child. Since there is a predominance of younger maternal age in the occurrence of Down syndrome, there is a need for concerning the factors that contribute to the occurrence of Down syndrome in further studies.*

I. INTRODUCTION

According to the American Association on Mental Retardation [1]. Mental retardation is defined as" Mental retardation refers to substantial limitations in present functioning. It is characterized by significantly sub average intellectual functioning, existing concurrently with related limitations in two or more of the following applicable adaptive skill areas: communication, self-care, home living, social skills, community use, self-direction, health and safety, functional academics, leisure and work. Mental retardation manifests before age 18". (American Association on Mental Retardatio [1]. Down syndrome is the most common genetic cause of mental retardation, specific birth defects and medical condition. It is the common name for a genetic anomaly, trisomy 21 non- disjunction of the 21st chromosome during meiosis, resulting in a zygote carrying an extra 21st chromosome. Down syndrome occurs in one out of every 600 to 800 live births, making it by far the most common form of chromosomal non-disjunction in the living human population.

Down syndrome is caused by non-disjunction during cell division, resulting in an extra chromosome 21 (trisomy 21). Most cases of Down Syndrome involve a non-disjunction during the first meiotic cell division, with mothers contributing the extra chromosome in 85% of cases. When non-disjunction occurs after fertilization, this leads to mosaic Down syndrome, where one line of cells in the developing fetus contains the extra copy of chromosome 21 and a second line of cells in the developing fetus does not. In a small percentage of cases. The most important risk factor is higher maternal age (more than 35 years), with a risk of 1:50 after the age of 45. According to the American Association on Intellectual and Developmental Disabilities Intellectual functioning involves the ability to reason, plan, solve problems, think abstractly, comprehend complex ideas, learn quickly, and learn from experience .Earlier studies had established that Down syndrome cases are associated with late maternal age [2]. However, recent studies indicate an appreciable shift to younger age groups [3,4,5] . As a woman ages, her risk for having a conception with trisomy 21 significantly increases. This effect of an increased rate of Down syndrome with maternal age was noted by Penrose [6] . There have been a lot of hypotheses by researchers and significant progress has been made toward characterizing the maternal age effect and understanding possible mechanisms.

The age of the mother at the time of the conception of a fetus with DS is, by far, the most significant risk factor for meiotic nondisjunction of chromosome 21 [5] . Interestingly, advanced maternal age is a risk factor for both Meiosis I (MI) and Meiosis II (MII) maternal nondisjunction errors [7] .The present study is an attempt to examine the occurrence of Down syndrome in relation to the maternal age and also the occurrence of Down syndrome in different maternal age groups with respect to gender and level of intellectual functioning of the child.It is well established that the risk of Down syndrome increases with higher maternal age. However some studies suggest that Down syndrome is more prevalent among children of younger maternal age whose age groups are less than 35 years. This is attributed to higher number of childbirth among those mothers who are less than 35 years. This study is designed to look at the relation of Down syndrome to maternal age among cases seen in General services, NIMH. (National Institute for the Mentally Handicapped) .The purpose of this research was to conduct a retrospective study for examining the occurrence of Down syndrome in relation to maternal conception age among the cases seen in General Services, NIMH (National Institute for the Mentally Handicapped) and To find out the maternal conception age of children with Down syndrome. And To find out the occurrence of Down syndrome in different maternal age groups.And also To find out the occurrence of Down syndrome in different maternal age groups with respect to child's gender. And finally To find out the occurrence of Down syndrome in different maternal age groups with respect to child's level of intellectual functioning.

II. METHODOLOGY

The topic selected for the present study is 'A retrospective study on Maternal age and Down syndrome. The sample consists of all the cases of Down syndrome registered in General Services, NIMH (National Institute for the Mentally Handicapped) from January, 2010 to December, 2011 . Data were obtained from case records of Down syndrome cases registered in General Services, NIMH during January, 2010 to December, 2011. Total number of Down syndrome cases registered during this period was 294 cases. Data obtained was analyzed by using Statistical Packages for Social Sciences version 17. Statistical analysis used for analyzing the data were: frequencies, percentage, mean and chi-square. Frequencies and percentage were calculated for maternal conception age, gender of the child, level of intellectual functioning of the child, maternal age with respect to gender and maternal age with respect to level of intellectual functioning of the child. Mean maternal age was calculated for the whole cases as well as for the two age groups, i.e. below 35 years and 35 years and above. Chi-square test was used to find out whether there exist a difference in the occurrence of Down syndrome at different maternal age with respect to the child's gender and level of intellectual functioning.

III. RESULTS

Hypothesis 1.1.3 There exists a difference in the occurrence of Down syndrome with respect to maternal conception age.

Table 1.1.3 difference in the occurrence of Down syndrome with respect maternal conception age

Age	N	Chi-Square	Sig
Below 35	**250**	**1.22**	**0.002**
35and above	**44**		

The objective is to find out the maternal age of children with Down syndrome and the occurrence of Down syndrome in different maternal conception age groups. To find out whether there exists a difference in the occurrence of Down syndrome in different maternal conception age groups, frequency, percentage and mean was calculated. Analysis reveals that there is a difference in the occurrence of Down syndrome in different maternal age groups. In according to Table 1.1.3 the obtained value of p: 0.02 and its below the 0.05 Hence, the hypothesis , 'There exists a difference in the occurrence of Down syndrome with respect to maternal conception age ' is accepted. This reveals that there is a significant difference in the occurrence of Down syndrome with respect to different maternal conception age groups.

Hypothesis 1.2.3 There exists a difference in the occurrence of Down syndrome in different Maternal conception age with respect to gender.

Table 1.2.3 difference in the occurrence of Down syndrome in different Maternal conception age with respect to gender

Gender	N	Chi-Square	Sig
Boys	173	1.80	0.09
Girls	121		

The objective is to find out the occurrence of Down syndrome in different maternal conception age groups with respect to Gender. To find out whether there exists a difference in the occurrence of Down syndrome in different maternal conception age groups, Chi-square was carried out for data analysis. Statistical analysis reveals that the obtained value of p : 0.09 and its upper than 0.05 hence there is no significant difference in the occurrence of Down syndrome in different maternal age groups with respect to gender. Hence, the hypothesis , 'There exists a difference in the occurrence of Down syndrome in different maternal conception age groups with respect to gender' is rejected at p< 0.09 level of significance. This reveals that there is no significant difference in the occurrence of Down syndrome in different maternal conception age groups with respect to gender.

Hypothesis 1.3.3 There exists a difference in the occurrence of Down syndrome in different maternal conception age with respect to level of intellectual functioning of the child.

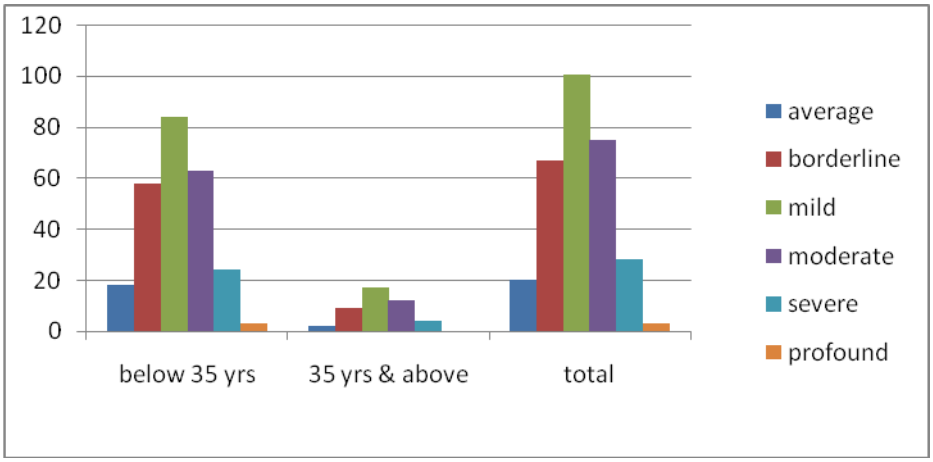
Table 1.3.3 Maternal conception age * Level of intellectual functioning Cross tabulation

Maternal conception Age	Level of intellectual functioning						Total
	average	borderline	mild	moderate	severe	profound	
Below 35 years	18	58	84	63	21	6	250
35 years & above	2	9	17	12	4	0	44
Total	20	67	101	75	25	6	294

Table 1.4.3 Difference in maternal age with respect to level of intellectual functioning of the child

Chi-Square	Value	Df	Asymp. Sig. (2-sided)
Pearson Chi-Square	1.928[a]	5	.859
Likelihood Ratio	2.853	5	.723
Linear-by-Linear Association	.055	1	.815
N of Valid Cases	294		

Fig 1.1.3 Maternal conception age * level of intellectual functioning



The objective is to find out the occurrence of Down syndrome in different maternal conception age groups with respect to level of intellectual functioning of the child. To find out whether there exists a difference in the occurrence of Down syndrome in different maternal conception age groups with respect to level of intellectual functioning of the child, Chi-square was carried out for data analysis. Statistical analysis reveals that there is no significant difference in the occurrence of Down syndrome in different maternal age groups with respect to level of intellectual functioning of the child. Hence, the hypothesis , 'There exists a difference in the occurrence of Down syndrome in different maternal conception age groups with respect to level of intellectual functioning of the child' is rejected at $p< 0.01$level of significance. This reveals that there is no significant difference in the occurrence of Down syndrome in different maternal conception age groups with respect to level of intellectual functioning of the child.

IV. DISCUSSION

The main purpose of the study is to examine the occurrence of Down syndrome in relation to maternal conception age among the cases seen in General Services, NIMH. The results of the study are discussed below. The objective is to find out the maternal conception age of children with Down syndrome and the occurrence of Down syndrome in different maternal age groups. Based on the findings of this study, more number of Down syndrome occur in maternal conception age less than 35 years of age. This finding is in line with many previous studies. In a study done by Hahn and Shaw [8] . they showed a modest increase in the live births of Down syndrome in mothers aged under 35 years. Similar results was found [3,4,5,9,10 and 11].Various studies done in India also indicate an appreciable shift of maternal age distribution to younger ages. In the study done by Parikh and Goyal [12] , 55.9% mothers were below the age of 30 years , 78.2% in Isaac & Reddi's study [3] and 65.9% mothers in the study by Verma et al [4] . In contrast to the finding in this study, some studies found more number of Down syndrome born to women age 35 years or above. Delport et al [13] documented an incidence of 1.33 livebirths of Down syndrome cases in a Pretoria urban academic hospitalwhere 52% of the mothers of Down syndrome infants were 35 years of age or older. In the same year, Venter et al recorded a figure of 2.09 per 1000 livebirths in a rural hospital where 56% of mothers of DS infants are 35 years of age or older and Kromberg et al [14] recorded an incidence of 1.67 per 1000 livebirths at an academic hospital in Johannesburg where 55% of the mothers of DS infants are 35yrs or older. Adeyokunnu in his study in Nigeria [15] found 76% of the mothers of DS infants were 35 years or older. Modi et al [16] in their study found that maternal age specific prevalence of Down syndrome increased from 0.54/ 1000 at 15-19 years to 15.6/ 1000 at > 40 years.

As a woman ages, her risk for having a conceptus with trisomy 21 significantly increases. This effect of an increased rate of Down syndrome with maternal age was noted by Penrose [17]. There have been a lot of hypotheses by researchers and significant progress has been made toward characterizing this effect and understanding possible mechanisms. Sherman, S.L et al [7] conclude that the maternal age effect is restricted to mothers in whom the nondisjunction error occurred. The objective is to find out the occurrence of Down syndrome in different maternal age groups with respect to child's gender. Statistical analysis reveals that there is no significant difference in the measures. Based on the findings, the hypothesis ' there exist a difference in the occurrence of Down syndrome in different maternal conception age group with respect to gender' is rejected. In contrast to this study, other studies showed a difference in gender in the occurrence of Down syndrome where prevalence of male is predominant. Kovaleva et al [16] in their study revealed male predominance and most expressed male predominance was determined in children of mothers aged below 35 years. In general, many studies have reported excess of males among Down syndrome abortuses and livebirths like studies done by

[18,19,20,,12,9,,21 and 22] In a study done by Kaur and Singh [21] 70.2% were males and 29.7% were female Down syndrome children. Meta analysis of publications reporting cytogenic profile of Down syndrome worldwide showed typical male prevalence among both patients with regular trisomy 21 and carriers of translocation trisomy 21 and female prevalence among carriers of mosaicim.The objective is to find out the occurrence of Down syndrome in different maternal age groups with respect to child's level of intellectual functioning. Based on the statistical analysis, there is no significant difference between the measures. Hence, it illustrates that there exist no difference in the occurrence of Down syndrome in different maternal age with respect to level of intellectual functioning. Mental deficiency in Down syndrome varies considerably and as a function of age [18]. Before the age of 5 years, I.Q is difficult to determine. In Down syndrome children, it is around 50 between 2-5 years of age, increasing to 58 around 3-4 en degenerates to a mean value of 38 at around the age of 15 years. There is a definite deterioration after the age of 35 years with dementia occurring in 15 to 30%. DS may be seen on either side of the mean, those with relatively high I.Q's and those condemned to a vegetative life. It is the abstract reasoning faculties that are most afflicted whereas affectivity and social ability are fairly normal, at least in infants. In adults the acquisition of a certain number of automatisms may mask the extent of mental deficiency. Parikh & Goyal [12], in their study demonstrated a majority (54.9%) of cases with I.Q below 40 and 32.9% moderately retarded as compared to the study done by Narayanan et.al [3] where 74.5% of 508 cases were moderately or severely retarded.

V. RESEARCH FINDINGS

[1] There is a difference in the occurrence of Down syndrome with respect to maternal age.
[2] There is no difference in the occurrence of Down syndrome in different maternal age groups with respect to gender of the child.
[3] There is no difference in the occurrence of Down syndrome in different maternal age groups with respect to level of intellectual functioning of the child.

VI. CONCLUSION

The research findings revealed that there is a difference in the maternal age with the occurrence of Down syndrome but no significant difference in the occurrence of Down syndrome in different maternal age groups in relation to gender and intellectual functioning of the child. Since there is a predominance of younger maternal age in the occurrence of Down syndrome, there is a need for concerning the factors that contribute to the occurrence of Down syndrome in further studies.

Implications of the study

[1] The present study is useful for rehabilitation professionals in counseling the parents and the need to create more awareness in prenatal screening of pregnancies.
[2] The present study is helpful to understand the present scenario of the occurrence of Down syndrome
[3] This study will serve as a basis for conducting further research studies.

Limitations

[1] Less sample size
[2] Less variables are taken in account

Recommendations for future research

[1] There can be further studies done on larger sample over a more liberal time frame.
[2] Other variables like cytogenic abnormalities in Down syndrome can be included.

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