Abstract

Mental retardation is significantly sub average intellectual functioning present from birth or early infancy, causing limitations in the ability to conduct normal activities of daily living. Level of mental retardation, maternal age, family history of mental retardation and consanguinity associated disorder was recorded from data given by parents or center of rehabilitation. Forty seven percent mentally retarded patients were born to the mother’s of age 21-25 years. There were 4% mentally retarded patients in 41-45 age groups. Order of birth varied from 1-3 in family history of mental retardation. Consanguinity was recorded in 16 cases of mental retardation. The information on possible risk factors can be used for genetic counseling and pregnancy supervision. Proper monitoring of risk factors could prevent our human health system from a serious life long disability and burden of the society could be reduced.

Keywords: Risk Factors, Cognitive Disorders, Age of Mother, IQ.

1. Introduction

Mental retardation is one of the most frequent handicaps among children and can be a serious and life long disability placing heavy demands on the society and the Health System (Pollak, 1993 and Swaiman, 1994). It is a common disorder, which imposes a large medical, psychological and social burden. It affects about 3% of the population, yet the pathogenesis is poorly understood (Birch et al, 1970 and Curry et al, 1997). It is a frequently occurring disorder with a major impact on the life of the affected person, their family, and society. Establishing an etiologic diagnosis of patient is usually a challenge for every specialist, as the spectrum of possible underlying disorders is enormous and the range of available additional investigations is extensive. The costs of a complete diagnostic work-up in a child with mental retardation are considerable and can be a major burden to many health care systems. Therefore usefulness of every diagnostic investigation is very important. Aetiologic diagnosis for a disorder is a specific diagnosis that can be translated into useful clinical information for the family, including providing information about prognosis, recurrence risk, and preferred modes of available therapy.

The ability to determine a cause of mental retardation is based largely on the use of specific diagnostic tools. In a given diagnostic setting, the physician or clinician depends on their availability and guidelines for application. There are no such clear cut guidelines available for diagnostic testing of mentally retarded patients. Such guidelines can be established on information from empirical studies. Several investigators have reported the diagnostic outcomes from systematic evaluation of large numbers of institutionalized (Opitz et al, 1982 and Moser et al, 1990) or community based mentally retarded patients (Stevenson, et al, 1996). A small number have either looked at fewer patients with carefully defined types of problems (Chaudley et al, 1998) or at the results from specialized diagnostic centers (Battaglia et al, 1999). However
Diagnostic outcomes of these studies provided little information for the evaluation of patients. Accurate diagnosis of etiology has specific implication regarding treatment. Management of possible associated condition, prognosis, and estimation of recurrence risk and the design of prevention program (Shevell, 1998).

2. Material and Methods

Present study has been done on 500 mentally retarded patients from 30 centers of 12 districts of Haryana. A Questionnaire has been developed for the general assessment of patients including, history of prenatal, neonatal, postnatal disorders, congenital disorder, details of severity of developmental delay, motor behaviour, associated disorders, parental age, consanguinity, family history of mental retardation etc. Level of cognitive functioning (IQ) was determined using Seguin form board test. Depending upon level of mental retardation, these patients were divided into four different classes. The classification was done as per WHO system of classification (WHO, 1993). A three generation pedigree with special attention to the presence of mental retardation, congenital disorders and consanguinity has been prepared for each mentally retarded patient.

3. Results

Maternal age at the time of birth of mental retardation cases was recorded and analyzed. Maternal age of mother was divided into six groups ranging from 15 to 45 years of age. 46.6% mentally retarded patients were born to the mother’s of age 21-25 years. There were 4% mentally retarded patients in 41-45 age group and 23.3% in 15-20 age groups. Frequency of 8.6 % and 5.3 % of mentally retarded patients were found in the age group of 31-35 and 36-40 years respectively (Table 1).

<table>
<thead>
<tr>
<th>Sr. No.</th>
<th>Age of Mother</th>
<th>% Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.</td>
<td>15-20</td>
<td>23.3</td>
</tr>
<tr>
<td>2.</td>
<td>21-25</td>
<td>46.6</td>
</tr>
<tr>
<td>3.</td>
<td>26-30</td>
<td>12</td>
</tr>
<tr>
<td>4.</td>
<td>31-35</td>
<td>8.6</td>
</tr>
<tr>
<td>5.</td>
<td>36-40</td>
<td>5.3</td>
</tr>
<tr>
<td>6.</td>
<td>41-45</td>
<td>4.0</td>
</tr>
</tbody>
</table>

Maternal age > 30 and < 30 of patients of different IQ level was studied in detail and it was found that mentally retarded patients born to mother’s of age less than 30 years were 81.9 % and to mother’s aged more than 30 years were 18.3%. (Table 2).

<table>
<thead>
<tr>
<th>Sr. No.</th>
<th>IQ Level</th>
<th>&lt;30</th>
<th>&gt;30</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.</td>
<td>Mild</td>
<td>31.33</td>
<td>0.33</td>
</tr>
<tr>
<td>2.</td>
<td>Moderate</td>
<td>44.33</td>
<td>11.33</td>
</tr>
<tr>
<td>3.</td>
<td>Severe</td>
<td>6.33</td>
<td>6.66</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>81.99</td>
<td>18.32</td>
</tr>
</tbody>
</table>

Order of birth in mild and moderate group varied from 1-3 and in severe it was first or second. Age of mother of these patients varied from 17-39 years.

Consanguinity was recorded in 16 case of mental retardation. Out of these 16 cases, two patients had mild mental retardation, seven patients had moderate and seven patients had severe mental retardation. Parental age of these patients varied from 22 to 37 years (Table 4).

<table>
<thead>
<tr>
<th>IQ</th>
<th>No. of Cases</th>
<th>Age (Years)</th>
<th>Birth Rank</th>
<th>Mother’s Age (Years)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mild</td>
<td>4</td>
<td>12-27</td>
<td>1-3</td>
<td>24-32</td>
</tr>
<tr>
<td>Moderate</td>
<td>12</td>
<td>7-25</td>
<td>1-3</td>
<td>18-39</td>
</tr>
<tr>
<td>Severe</td>
<td>6</td>
<td>12-27</td>
<td>1-2</td>
<td>17-35</td>
</tr>
<tr>
<td>Total</td>
<td>22</td>
<td>7-27</td>
<td>1-3</td>
<td>17-39</td>
</tr>
</tbody>
</table>

4. Discussion

The association between increasing maternal age and chromosomal anomalies is an arguably the most important...
etiological factor in human genetic disease. Although advanced maternal age is a well-established risk factor for trisomy 21 (Down syndrome), much remains to be learned about the effect of maternal age on the other chromosomally as anomalies well as on mental retardation. Several studies have noted that the age related risk of trisomy 21 does not continue to increase exponentially with increasing age for women of 45 years of age or over (Ferguson-smith and Yates, 1984). The highest advanced maternal age (AMA) fractions occurred among Asian/pacific Islanders, followed by non Hispanic Caucasians Blacks and Hispanics and Native Americans.

Advanced maternal age effect was observed among the patients with developmental disabilities (Drews et al, 1995). Patients of mild mental retardation cases were found to be associated with increased maternal age (Lisa et al, 2001). No maternal age effect was seen among isolated cases. However when cases were categorized by stage of origin and maternal age (>30 years or <30 years), significant association was found with the effect being confined to meiosis II cases (no disjunction) leading to numerical chromosomal abnormalities (Hassold and Serman, 2000). Presence of 81.7% cases born to mother’s age of less than 30 was recorded in the present study. Cytogenetically analyzed cases revealed 98 cases of abnormal karyotype. Maternal age was more than 30 in 32.3% cases and less than 30 in 67.7% of mentally retarded cases. Outcome of the present study were not in favour of positive effect of maternal age on the occurrence of chromosomal anomaly of the patients as well as on the mental retardation.

Deviation from earlier reports could be due to the fact that Indian marriage is mostly performed at an earlier age of 20-25. The data for older mother was not available. It is rare for the women of 40 years of age and over to become pregnant. However as the number of mentally retarded patients was too high irrespective of maternal age, the presence of other intrinsic and extrinsic factors can not be ignored. These factors must be affecting the normal growth and development of fetus inside the uterus.

The population based study of Hou et al (1998) reported that 15% of the families in a large population have a positive history of mental retardation; the degree of familial relationship was not indicated. In the present study 22 cases of mental retardation had family history of mental retardation and 13 cases were analyzed cytogenetically, only 2 cases were having trisomy 21, rest of the cases were of normal karyotype. Parents of 13 cases were cytogenetically normal. Therefore, no correlation could be established between familial mental retardation and mentally retarded patients. More or less the same holds for the parameter, consanguinity. This was reported in many studies (Loxova et al, 1967; Costeff et al, 1972; Bundey et al, 1985; Farag et al, 1993 and Roberts, 1996). The median percentage of families with consanguinity was 9.1% (range 0.7–85.5%). As, again, it was usually not stated to which extent consanguinity was surveyed or reported, and also because of the different ethnic backgrounds of the study populations, it was not possible to draw meaningful general conclusions. Epidemiological study of mental retardation in Pakistan could not find any association between consanguinity and the prevalence of mental retardation (Hasan, 1988). In Bangladesh 60%, mentally retarded children were offspring of consanguineous union. The overall prevalence of serious mental retardation was extremely high (Islam et al, 1993). In the present study there were 16 mentally retarded cases were the offspring of consanguineous marriage. The parents of these children were cytogenetically normal and did not reveal any history of mental retardation. No association could be established between consanguinity and mental retardation. However serious cognitive disabilities along with infection indicate a correlation between the two.

5. Conclusions

These factors will play an important role in the etiologic diagnosis which is important for providing information about pathogenesis, prognosis, recurrence risk and special medical intervention. Present study has identified several variables that are associated with whether a syndrome genetic diagnosis will be made in a patient referred to genetics clinics for the evaluation of mental retardation. These results suggest that selective use can be made of the cytogenetic laboratories, thus increasing the chances of positive test results and decreasing costs at a minimal risk of missing a significant finding. One can also foresee that a genetic clinic could develop a triage protocol where, at initial contacts, an assessment is made as to whether the patient should have an immediate complete clinical morphological assessment, specific laboratory testing, or should proceed directly to a genetic counseling session, based on history and physical findings.

References


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