EPILEPSY, HYPOTONIA, MICROCEPHALY AND STRABISMUS IN 185 MENTALLY CHALLENGED CHILDREN IN INDIA-A REPORT

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ABSTRACT

Mental retardation is a complex clinical condition and the affected individuals display a wide array of associated disabilities like epilepsy, hypotonia, oro-motor disorders, gross motor abilities, abnormal reflexes, craniofacial anomalies, the presence of major congenital malformations, unclear speech and hyperactivity. The severity and frequency of these comorbid symptoms is often directly related to the IQ of the suffering child. We randomly selected a population of 185 mentally challenged children from the state of Haryana (India) and studied it in relation to epilepsy, hypotonia, microcephaly, strabismus and other physical and behavioural symptoms and observed a strong correlation between epilepsy and strabismus as well as epilepsy and microcephaly. Data generated from such studies can be immensely helpful for the community health workers operating in the rural areas of developing countries like India and also for the poor parents who do not have the access to proper clinical diagnosis and genetic counselling.

Key words: epilepsy, hypotonia, IQ, mental retardation

INTRODUCTION

Mental retardation originates as a consequence of abnormal brain development and impaired cognition. The onset of the condition occurs during the developmental period, i.e., gestation through age 18 years. The condition is present in 2 to 3 percent of the population worldwide, either as an isolated finding or as part of another syndrome. In India, mental retardation was observed to have a prevalence rate of 4.2 per 1000.¹ Causes of mental retardation are numerous and can be grouped from most to least common as alterations in embryonic development, like those caused by chromosomal abnormalities or fetal exposure to drugs or toxins; environmental deprivation and other mental disorders, such as autism; problems of pregnancy and the perinatal period, such as fetal
malnutrition, hypoxia, infection, trauma, or prematurity; hereditary abnormalities, such as inborn errors of metabolism or chromosomal aberrations and medical conditions of infancy or childhood, such as central nervous system (CNS) infection or trauma, or lead poisoning.

Although mental retardation is usually observed as a unique clinical condition, affected individuals do not conform to a homogeneous group as mental retardation is a culmination of many different etiologies. Many individuals with mental retardation have associated disabilities such as epilepsy, cerebral palsy, autism, craniofacial anomalies, disturbed vision, oro-motor disorders (drooling), gross motor abilities, abnormal reflex patterns, postural and balance reactions, the presence of major congenital malformations, unclear speech and hyperactivity.

Epilepsy which is a tendency of occurrence of transient recurrent abnormal electrical discharges in the brain, affects one or more of the following brain functions: motor, sensory, cognitive, speech, behavioral, emotional, and psychological. Epilepsy is also common in children with mental retardation. Several authors have reported the comorbidity of epilepsy and mental retardation in as many as 16-44% of the cases. Frequency and severity of chronic epileptic seizures is closely related to severity of intellectual disability. Researchers have reported that 4% to 10% of all children experience a seizure at some time in their lives, however, only 1% of the population has a diagnosis of seizures or epilepsy by the time they turn 20 but prevalence in mentally retarded population is much higher. Similarly, the lifetime prevalence of epilepsy in the general population is between 2 and 3% but the reported prevalence of lifetime epilepsy among people with mental retardation (IQ< 70) varies between 13 and 24%.

There are several reports stating the co-existence of microcephaly and mental retardation. Though not all microcephalic children turn out to be mentally retarded but a significant part of this population are able to develop only a borderline or very low IQ. In addition to the intellectual disability there is a heightened possibility of hypotonia, spasticity, cerebral palsy, growth retardation, epilepsy and strabismus. Observations like frequent association of microcephaly with mental retardation and that abnormal head circumference, when accompanied by other physical symptoms mentioned above, can be used to identify preschool children potentially at risk for learning disabilities. Strabismus or squint is a common visual impairment in mentally retarded children and the prevalence is much higher than in the general population. Similarly, hypotonia or general motor disability is widespread among clinical presentation in people suffering from mental retardation. Motor disability in mentally retarded individuals includes longer motion and reaction times, balance and postural deficits. In several cases of mental retardation facial hypotonia manifested by tented upper lip, open mouth, and drooling persists till adolescence and even in adulthood in some of them. The neuropathological basis for motor dysfunction in mentally retarded is unknown, but cerebellar dysfunction have been suggested as one of the many possible causes.

All these studies have focussed independently on the problems of mental retardation, epilepsy etc. thus generating a scattered array of data. In the small towns and rural areas of developmental countries like India where specialised psychiatric and neurological clinics are a rarity and socio-economic status as well as health awareness of most of the families is not up to date to cater to the special needs of mentally challenged children, there is a growing need for a wholesome set of data which could aid the existing
information of teachers and community health workers so that developmental delays and
dysmorphic features like microcephaly could be related to the presence and severity of
mental retardation early in childhood. Such information can prove to be extremely
beneficial if made available at the root level of social health workers, midwives best of
all expecting mothers.
We designed this study to look at the prevalence, frequency and severity of epilepsy,
dysmorphic features and motor anomalies in mentally retarded child population and to
generate a wholesome set of data that would link the severity of intellectual dysfunction
to the presence of morphological, behavioural and functional characteristics which are
easy to observe and monitor.

MATERIALS AND METHODS

The study was approved by the Institutional Ethics Committee and pre-informed consent
to observe a participant was obtained from parents/guardian in every case. We randomly
selected a population of 185 children and adolescents aged between 3 and 18 y from the
various special schools from the state of Haryana, India. A vast majority of these
children belonged to families of poor socio-economic status who lacked resources for the
proper clinical tests and diagnosis of the cause of their condition, hence the cause for the
mental retardation of almost all the participants in this study was undiagnosed. As all the
participants were selected from special schools, all of them had a valid IQ score certificate
issued by authorized government agencies. We used the International Statistical Classification of Diseases and Related Health Problems (ICD-10), which classify children with intelligence quotients (IQ) below 70 as mentally retarded. Mental retardation was considered as a *Primary disability* (birth onset or acquired disability that is permanent and lifelong) while epilepsy and all the other disabilities and abnormalities studied were considered secondary conditions. As the accurate measurement of the head circumference of a growing child needs to be calculated as an average of many serial readings taken over the years and as the time span of this study was not that long so we depended on the observations and amateur comparisons made by parents and teachers over the years. Epilepsy was identified by the presence of the ICD-9 Code 345. It was classified on the basis of frequency of seizures; grouping severe epilepsy as more frequent than once a month; intermediate epilepsy as more frequent than once a year. The level of cognitive functioning (verbal as well as performance) of all the mentally challenged participants was estimated by a single observer. The sample IQ-deficit distribution was as follows: borderline [IQ 70–79] 6%, mild [IQ 50–69] 32%, moderate [IQ 35–49] 35%, severe [20–34] 20%, and profound [IQ _20] 6%. None of the 185 children were receiving long-term pharmacological treatment. Of the total population 25% were females and the rest were males. For comparison we studied an age-matched population of 185 healthy school children from the age of 3 to 18 years.

RESULTS

The general composition of the population under study was as given in Table 1. Out of
the total subjects studied 25% were females and the rest were males.
Table 1. Age and IQ distribution in mentally challenged population.

<table>
<thead>
<tr>
<th>IQ Group</th>
<th>Mean Age</th>
<th>Mean IQ</th>
</tr>
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<tbody>
<tr>
<td>Below 20</td>
<td>12.83±3.06</td>
<td>16±1.56</td>
</tr>
<tr>
<td>20-34</td>
<td>11.84±3.75</td>
<td>30.26±3.08</td>
</tr>
<tr>
<td>35-49</td>
<td>12.04±3.85</td>
<td>42.04±4.4</td>
</tr>
<tr>
<td>50-69</td>
<td>13.31±4.27</td>
<td>54.66±5.14</td>
</tr>
<tr>
<td>70-79</td>
<td>15.33±3.2</td>
<td>75±2.13</td>
</tr>
</tbody>
</table>

The results thus obtained are represented as follows:

Profound mental retardation: As much as 42% children in this group suffered from multiple disabilities. The occurrence of epilepsy seemed to increase with age in this group and 90% of these epileptic children suffered from severe epilepsy with fit frequency of once a week. 17% subjects had microcephaly, 42% had strabismus and 8% had both small head and squint. Approximately 50% subjects had facial hypotonia which resulted in open mouth and drooling. 42% children in this group had poor posture and gait owing to a hypotonic trunk and stiffened limbs.

Severe mental retardation: The percentage age was same as that of previous category but the frequency of seizures decreased to just once a month in 76% of the epileptic cases. 8% children were microcephalic and one third of these had poorly balanced, floppy head. One microcephalic child who was under 8 years of age had a grossly underdeveloped sense of direction and a very violent nature. 5% had a disproportionately large head size even in adolescence. 30% subjects suffered from squint. Facial hypotonia manifested in 41% children of this group out of which 27% had problem of drooling too. Approximately 35-40% children had poor posture and gait.

Moderate mental retardation: The prevalence of multiple disabilities had decreased to 14%. The pattern of occurrence of epilepsy in different age groups was reversed in this category as compared to the previous two. 25% of the affected cases had intermediate epilepsy while the rest were severely epileptic. The instances of microcephaly were only 2% in this group while the percentage of macrocephalic adolescents had dropped to 3% and only 2% had floppy head. The prevalence of squint had dropped to 14%. 26% subjects had open, slack mouth and more than half of these (18%) had drooling problem too. 9-12% subjects had poor posture and gait.

Mild mental retardation: Multiple disabilities were observed only in 7% of the cases and except a minor proportion (11%) of children all the rest were able to do their daily chores. The occurrence of epilepsy was also very less as compared to the previous groups. Only 3% of adolescents had fits and all of these cases fell under the category of well-controlled epilepsy. Only 2% subjects were microcephalic and 4% had poorly balanced heads. The instances of strabismus had dropped to 7%. 14% subjects had the problem of open mouth and out of these only 5% drooled. 10-15% subjects had poor posture and gait.

Borderline mental retardation: Here also only 7% of the cases had multiple disabilities. None of the subject in this category was epileptic and all of them were normocephalic. Incidence of strabismus was approximately 8% while that of slack mouth was 17% out of which only 8% drooled. Only 8% had poor posture and all of them could walk normally.

Figure 1 presents a summarized account of the results discussed above.
Figure 1: Prevalence of head, eye and oro-motor abnormalities in mentally challenged population.

We found a strong correlation of 0.94 between the occurrence of epilepsy and microcephaly in the mentally challenged participants (Figure 2). Similarly the correlation of epilepsy with strabismus was also noteworthy (0.87). 60% of epileptic subjects had multiple congenital abnormalities.

Figure 2: Prevalence of epilepsy in various IQ groups.
Besides epilepsy, microcephaly, strabismus and hypotonia the pattern of occurrence of certain other disabilities like mutism despite normal hearing and inability to perform the basic self-care tasks like bathing, clothing, feeding etc. was also noteworthy. The prevalence of mutism despite apparently normal hearing was highest in profoundly mentally retarded group (25%) and decreased with increasing IQ through the latter categories. A similar pattern was observed in case of self-care and personal hygiene tasks.

Figure 3. Eye presentation.

A mentally retarded child with strabismus

A normal child with normal eyes

The prevalence of above discussed disabilities and epilepsy in the healthy group of children (that served as control) is shown in Table 2.
Table 2. Result summary of Healthy (control) population.

<table>
<thead>
<tr>
<th>Problem</th>
<th>Prevalence</th>
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<tbody>
<tr>
<td>Squint</td>
<td>2%</td>
</tr>
<tr>
<td>Open mouth and drooling</td>
<td>none</td>
</tr>
<tr>
<td>Delay in speech</td>
<td>0.03%</td>
</tr>
<tr>
<td>Mutism despite normal hearing</td>
<td>none</td>
</tr>
<tr>
<td>Poor posture and gait</td>
<td>0.1%</td>
</tr>
<tr>
<td>Epilepsy</td>
<td>0.4%</td>
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</table>

**DISCUSSION**

All the participants in this study suffered from deficiency in one or all of the five developmental domains i.e motor function, exploration, sociality, activities of daily living and language.

Epilepsy, microcephaly and strabismus- Epilepsy exhibited an inverse proportionality with IQ. The strong correlation (0.94) between epilepsy and microcephaly also verified with the extreme IQ deficit as the incidence of both these disorders was maximum in profoundly mentally challenged group. Forsgren et al (1990)\(^{18}\) observed similar trends. We observed an almost identical relation in prevalence of epilepsy and strabismus (0.87), as strabismus was also most prevalent (67%) in profoundly mentally retarded kids. These observations signify the fact that active epilepsy in mentally retarded children is often associated with additional neuroimpairments, especially severe visual impairment.\(^{19}\)

Hypotonia and motor anomalies- Oro-motor deficiencies like open mouth and drooling occur less frequently in normal children than in mentally challenged ones and in almost all the normal cases the drooling stops by the time the child reaches the age of three while in mentally handicapped kids not only the drooling was more common in lower IQ groups but also it persisted well into adolescent age unlike the normal children. Gross motor disabilities were scantily observed only in cases of polio or paralysis in normal children whereas poor posture and gait due to reduced muscular tone and stiffened or slack limbs were observed repeatedly in mentally challenged children especially those suffering with profound and severe mental retardation. Stiffness in hands and fingers and the resulting inability to grasp and manipulate objects for activities like eating, writing, dressing, building and playing was also present more in profoundly and severely mentally children. This problem decreased considerably in the higher IQ groups. Since the functions of voluntary movement, balance and motor-tone control are primarily cerebellar in nature so a deficit in these along with malfunctional limb movements and speech indicates cerebellar impairment.

Multiple developmental anomalies- Compared with MR children, the number of observed developmental anomalies per child in healthy children was one fourth to half the MR's. Although some of the developmental disabilities studied here like delay in speech, squint, etc. are also observed in healthy children but rare is an intellectually normal child suffering from more than one of these problems simultaneously. Contrarily, we found that most of the MR children had multiple abnormalities and the highest number of such
children was observed in profoundly mentally retarded group and decreased gradually in higher IQ groups till less than 10% in borderline cases.

In addition to all the above disabilities, mentally challenged population also faces the peril of decreased life expectancy as compared to normal population. A number of studies have acknowledged relatively high mortality rates among individuals with mental retardation and severe disabilities. Variables found to be related to life expectancy include basic skills such as ambulation or mobility, method of feeding, bowel and bladder control and level of mental retardation. Retarded persons in institutions are living longer than previously, but their life expectancy does not meet that of the general population.20

CONCLUSION

Keeping in view all the above observations, it becomes significant that the parents of mentally retarded children and community workers in developing countries all over the world should be taught about recognizing the symptoms and problems associated with varying degrees of mental retardation.

Neuro-developmental disorders like epilepsy and microcephaly when observed together with hypotonia, strabismus and malfunctions in speech can provide a rough indication of the extent of intellectual deficit in a child and can thus help teachers and parents to care for their wards accordingly. Early identification and intervention can aid tremendously in increasing the longevity of a mentally retarded child especially in underdeveloped and developing countries where genetic counselling, clinical diagnosis and specialised care are not accessible to a large segment of mentally challenged population.

CONFLICT OF INTEREST

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REFERENCES


