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Does any Correlation exist between I.Q levels, Diagnosis Age & Morphological Parameters in Down Syndrome Patient?

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ABSTRACT: The intellectual or mental development of children and adults with Down syndrome has often been described in terms of IQ (Intelligence Quotient) which is calculated on the basis of the various IQ tests. In the present study of Down syndrome patients from Haryana State, all the Down syndrome patients were categorized into four groups on the basis of degree of mental retardation following WHO system of classification (ICD-10, WHO 1993). Maximum number (52%) of Down syndrome cases was with moderate mental retardation. More number of cases of severe Down syndrome group could be diagnosed before six months as compared to mild and moderate group. Two ways ANOVA test was significant at 5% level, showed association between mental level and age of diagnosis of patients. Chi-square (χ^2) test indicated a significant association between chronological age & I.Q. levels in these patients. Delivery complications like mother had Rh incompatibility & bleeding during late pregnancy were more common in moderate & severe Down syndrome group as compared as compared to mild group. This study will be helping in evaluating the brain development & difference to the progress of patients with Down syndrome. The standardization data allows analyst to compare the scores for a particular child with those of a large sample of children of the same age, so that they can see if the child is doing better or worse than the standard of this large, given sample.

Key Words: Chromosomal abnormalities, Down syndrome, Intelligence Quotient, Diagnosis.

I. INTRODUCTION

Present study has been conducted on 200 Down syndrome patients from 30 centers of 12 districts of Haryana. I.Q. levels of Down syndrome patients were investigated with respect to diagnosis age, sex ratio, prenatal, postnatal history, parental age & morphogenetic variation. All the Down syndrome patients showed mental retardation of different levels. These patients were categorized into four groups (from mild to profound) on the basis of degree of mental retardation following WHO system of classification. The different classes were as follows: mild mental retardation (IQ. 51-70), moderate mental retardation (I.Q. 36-50), severe mental retardation (I.Q. 21-35) and profound mental retardation (I.Q. <20). In earlier studies, intellectual or mental development of Down syndrome patients has often been described in terms of IQ (Intelligence Quotient) & it is calculated on the basis of the individual's performance on an IQ test. Most of studies of mental development of individuals with Down syndrome have used this type of methods.

II. MATERIAL &METHODS

In the present study Intelligence Quotient (I.Q.) levels of 200 Down syndrome cases were studied. I.Q. of all the patients was calculated using Seguin Form Board Test. Depending upon level of mental retardation, these Down syndrome patients were divided into four different classes. The classification was done as per WHO system of classification (ICD-10, WHO 1993). Ethical approval for the study was taken by the Institutional Human Ethical Committee, MD University, Rohtak, Haryana.

Calculation of Intelligence Quotient

Trials	**Time Taken**
1st Trial	a
2nd Trial	b
3rd Trial	c

Calculation of mental age (x).

Average time taken = a+b+c/3 = x

x - Correspond to the mental age.

I.Q. = M.A./C.A.×100

M.A= Mental age (average mental ability displayed at given age)

C.A. = chronological age

III. RESULTS

Intelligence quotient (I.Q.): Patients having I.Q. between 51-70 were kept in mild mental retardation group, I.Q. between 36-50 in moderate mental retardation group, I.Q. between 21-35 into severe mental retardation group and last group in the profound mental retardation having I.Q. less than 20.

In the present study no case of profound mental retardation was found, whereas there were 52% cases of moderate mental retardation, 29% cases of severe mental retardation and 19% cases of mild mental retardation. Maximum frequency of Down syndrome with moderate mental retardation was recorded in the present study (Table1).

Table 1: Degree of mental retardation in Down syndrome Patients.

I.Q	% of patients
Mild(51-70)	19
Moderate(36-50)	52
Severe(21-35)	29
Profound<20	Nil

Diagnosis age & I.Q. Levels: Diagnosis age & I.Q. Levels were studied by retrospective analysis. Out of total Down syndrome cases which were diagnosed as mongoloid in neonatal period (<1 month), 21.05% were of mild mental retardation, 25% were of moderate mental retardation and 27.58% were of severe mental retardation. The cases which were diagnosed after two years of age had 21.06%, 14.43% and 15.52% frequency in mild, moderate and severe group respectively (Table 2). Down syndrome cases were diagnosed before one month & after twenty four months were in almost equal percentage in mild group, whereas more number of cases of moderate & severe Down syndrome cases could be diagnosed before one month as compare to after twenty four months (Fig. 1). Two way ANOVA test showed high F value for I. Q. levels which showed significant effect of mental levels on age of diagnosis of Down syndrome patients. The value of F was significant at 5% level for degree of freedom 2 (Table 3).

Table 2: Percentage frequency of Down syndrome cases with respect to diagnosis age and I.Q. (Intelligence Quotient) levels.

I.Q	Diagnosis age.				
	<1 month	1-6 months	7-12 months	13-24 months	>24 months
Mild	21.05%	18.42%	15.79%	23.69%	21.06%
Moderate	25.0%	19.23%	21.15%	20.19%	14.43%
Severe	27.58%	29.32%	13.79%	13.79%	15.52%

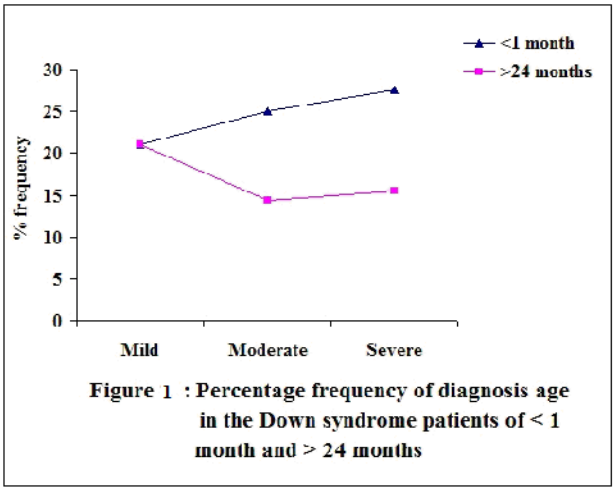

Figure 1 : Percentage frequency of diagnosis age in the Down syndrome patients of < 1 month and > 24 months

Table 3: ANOVA between diagnosis age and I.Q. (Intelligence Quotient) levels of Down syndrome patients.

Square of variation	Sum of square	Degree of freedom	Mean square	F-value
Between columns (diagnosis age)	66.65	4	16.66	1.61
Between rows (I.Q.)	458.13	2	229.06*	22.20
Residual	82.55	8	10.31	--
Total	607.33	14		

*Significant at 5% level.

Table 4: Percentage frequency of Down syndrome patients showing morphogenetic variation in different I.Q. levels.

I.Q → Features	Mild (51-70)	Moderate (36-50)	Severe (21-35)	Profound (<20)
Head & Face				
Brachycephaly	16.47	55.29	28.24	-
Flat occipital region	16.66	55.55	27.79	-
Slanting palpebral fissure	17.77	56.11	26.12	-
Small square ear	16.04	54.93	29.03	-
Protruded tongue	14.47	56.57	28.96	-
Epicanthal fold	15.88	55.88	28.24	-
Skin				
Mottled skin with reddish rim	18.10	60.35	21.55	-
Hand & Feet				
Single transverse palm crease	22.72	40.92	36.36	-
Short stubby hand	16.12	55.00	28.88	-
Wide gap b/w 1^{st} &2^{nd} toes	25.00	48.34	26.66	-
General				
Short stature	All	All	All	-
Mentally retarded	All	All	All	-
Harmless mischief	All	All	All	-
Streak of stubbornness	All	All	All	-

I.Q. Levels &Age group distribution: I.Q. levels of all the Down syndrome patients in different age groups were calculated. Below 10 years of age 36.84% of mild category, 23.07% of moderate category and 15.52% cases of severe category were found. In the present study maximum cases were of moderate mental retardation. Between 11-15 years and 16-20 years of age the percentage of moderate category Down syndrome cases was 29.81% and 27.88% respectively. In severe mental retardation group maximum cases (44.83%) were present between 16-20 years of age.

I.Q. Levels & Sex Ratio: When I.Q. Levels & sex ratio were analyzed in these patients, it was found that male prevalence was much higher in mild and severe mental retardation group as compared to that of moderate mental retardation group.

There were 31.7% females in moderate mental retardation and 18.5% and 15.5% females in mild mental retardation and severe mental retardation group respectively. Male Down syndrome patients of mild mental retardation were 81.5% and males of severe group were 84.5%, whereas males of moderate mental retardation were 68.3%.

I.Q. Levels & Prenatal and postnatal history: Prenatal, postnatal history & Clinical information of Down syndrome cases revealed the complications or deviation from normal history in all the three groups of mental retardation. Delivery complications like prolonged labour duration was found in 2% cases. These patients were of mild & severe categories. Caesarian delivery was found in 5.5% of cases & there I. Q. levels varied from mild to severe.

There was history of abortion in 5.5% cases & all these patients were of moderate category. Bleeding during late pregnancy was found in 7% of mothers.
Foetal movements were abnormal i.e. sluggish/ excessive in 5% of mothers.
Analysis of dysmorphic features in different I.Q. groups revealed more frequency in the Down syndrome cases of moderate group as compared to mild and severe group. Mental retardation, short stature, and streak of stubbornness were reported in all groups. Percentage frequency of Down syndrome patients showing dysmorphic features of head, face, skin, hand and feet was maximum in moderate group of mental retardation (I.Q. 36-50), then in severe group (I.Q. 21-35) and it was least in mild group.(I.Q. 51-70). Percentage frequency of mild and severe mental retardation groups with dysmorphic features did not exceed 40%, where as it was more than 40% in the moderate group (40.92% to 60.35%) (Table 4).

IV. DISCUSSION

The intellectual or mental development of children and adults with Down syndrome has often been described in terms of IQ (Intelligence Quotient) & various intelligence tests have been used for many years to assess mental abilities and different aspects of knowledge & memory of Down syndrome patients. With Down syndrome, general standard of intelligence scores (IQ scores) have been kept between 0 and 85 [1]. In the present study I.Q. scores were between 25 to 70 & on the basis of I.Q. scores these patients were categorized into three groups (from mild to severe) because no case of profound mental retardation was found.

In the study maximum number of cases of Down syndrome had moderate mental retardation. In the literature more than 80% cases of moderate or severe mental retardation has been reported for Down syndrome cases [2]. One study indicated that patients with trisomy 21 mosaicism have better cognitive function as compared to other cases of Down syndrome [3]. It has been suggested by Roelveld *et al.* [4], that more number of moderate and severe mental retardation cases are likely to be due to single pathological causes than the mild mental retardation which are thought to be multifactorial in origin.

Out of 250 Down syndrome patients, only 25% could be diagnosed as Mongoloid before 1 month & most of the cases were of severe mental retardation whereas 16% were diagnosed after two years & maximum number among these patients were of mild mental retardation.

In earlier studies 47.3% were diagnosed at birth or during neonatal period, 14% were diagnosed in the 2nd half of the 1st years and 18% were diagnosed after one year [5]. Diagnoses of maximum number of severe cases in neonatal period (< 1 month) may be due to the presence of more prominent features in severe cases as compared to mild & moderate cases. In another study it was reported that in 70.8% cases, Down syndrome was suspected on the day of birth. And in 1.7% of cases the diagnosis was made after 1 year. When child was delivered at hospital, 96.4% of Down syndrome cases had been diagnosed with in 1 month compared to 81.3% following home deliveries [6]. The place of birth and the expertise of health worker assisting at the delivery, clinical awareness of obstetricians and pediatricians to the diagnosis are some of the important factors should be kept in mind while diagnosing Down syndrome cases. However, this may be due to reflection of variability in the dysmorphic features of Down syndrome, which are not as apparent at birth as is usually thought.

It was reported that up to the early 1900s, people with Down's syndrome were typically viewed as profoundly mentally retarded. Surveys of kids and adults during the first half of this century classified most Down's syndrome people in the severely mentally retarded category. According to earlier reports in the 1960s, up to 10% of cases were mildly retarded. It was suggested in the mid 1970s, approximate, 30-50% of older kids and adults with Down's syndrome were in the mild range [7]. One another study suggested that the majority of Down's syndrome kids fell in the moderately to severely retarded range, with only a very small minority (2-3%) achieving at the mildly retarded level [1]. As in present study, in many earlier studies of children with Down syndrome, IQ results showed that the children's IQ scores decline as they grow older. One of the reasons for the decline in IQ scores is the very slow development of speech and language for most of the children and some early intervention programmes have reported success in slowing or stopping the IQ decline [8]. Such Decline in I.Q. scores & great variability in levels of I.Q. among Down syndrome cases may be due to the interactions of various genes on chromosome 21 or with genes on another chromosome, or due to allelic heterogeneity for chromosome 21or prevailing chromatin environment on gene expression.
Frequency of dysmorphic features are more frequent in the Down syndrome cases of moderate group as compared to mild and severe groups. Percentage frequency of mild and severe mental retardation groups with dysmorphic features was below 40%, where as it was upto 60% in the moderate group.

Present study depicted that in spite of great similarity among mongoloid, there is a wide range of variability in dysmorphic characteristics & I.Q. levels of Down syndrome. The nature and degree of psychosocial problems & dysmorphic features may differ significantly among Down syndrome patients. One earlier study also recorded a considerable range in the degree of mental retardation from profound to mild [2]. This variability in dysmorphic features & I.Q. levels strongly indicates degree of severity of Down syndrome and suggests involvement of complex genotypic interaction. The ultimate goal of study is to construct a phenotypic map of mental levels with various aspects like diagnosis age, sex ratio & clinical history of Down syndrome patients. This will be helpful in selecting the individuals for analysis at molecular level and ultimately the genes that are responsible for the particular phenotypes. To do this, both the phenotypes and molecular data have to be well defined.

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