Research Paper

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Clinical Profile of Down Syndrome Cases

Medical Science

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ABSTRACT

Introduction: Down syndrome is the commonest chromosomal anomaly seen in all the live births and the most frequent genetic cause of mild to moderate mental retardation, associated with medical problems. Aims & Objectives: The present study was undertaken to analyze various chromosomal anomalies associated with Down syndrome, to correlate them clinically and to counsel the parents / relatives as necessary. Method: 100 clinically suspected Down syndrome cases were selected for the study. A detailed history with pedigree chart and the examination findings were recorded for every patient. Chromosomal analysis (Karyotyping) was also done. Result & Conclusion: The results were statistically analyzed. The most striking clinical feature observed in the present study was mental retardation; others being delayed milestones, mongoloid slanting eyes, etc. Chromosomal abnormality was confirmed in 92 cases. A positive outcome of the present study was that, in maximum patients the diagnosis of Down syndrome had been made very early in their life. Counseling of the parents helped them in the better management of these subjects.

INTRODUCTION

Down syndrome is the commonest chromosomal anomaly seen in all the live births. It is a problem of global proportions and the most frequent genetic cause of mild to moderate mental retardation, associated with medical problems.

Dr. John Langdon Down 1 (1866) - a physician, published an essay in England in which he described a set of mentally retarded children with common features who were distinct from other children with mental retardation. He referred the condition as 'Mongolism', the name that was later changed to 'Down syndrome'.

The overall worldwide incidence of Down syndrome is 1 in every 650 to 1000 live births 2. It appears to occur with approximately equal frequency amongst various ethnic groups. The sex ratio i.e. male: female ratio is approximately 1.2 3. Frequency of Down syndrome in India is almost 1 in 916 live births4.

Down syndrome presents with chromosomal aberrations that lead to the characteristic clinical features and developmental problems. Only a small portion of the chromosome 21 is actually being needed to be triplicated to get the features. It is called the Down syndrome Critical Region

The present study was undertaken to assess the clinical profile of suspected and chromosomally confirmed cases of Down syndrome. Emphasis was also given on genetic counseling of the parents / relatives as it made them aware of the complications that the child may face in his / her future life and the possible management also.

Material and Methods

This was a cross-sectional study conducted in a tertiary institute over a period of 24 months. 100 clinically suspected Down syndrome cases were selected for the study. Approval of the institutional ethical committee was obtained. A written informed consent of the parents was taken in each case.

All the subjects had positive history and / or the clinical features, which were suggestive of Down syndrome. A detailed history was recorded for every patient. History of mental retardation, delayed milestones were given the utmost priority. Relevant past history and family history like consanguinity, similar illness and spontaneous abortion were also noted. Findings of general and systemic examination were also recorded in detail. A detailed pedigree Chart was recorded. Available information regarding other investigations such as IQ, ultrasonography, etc was recorded.

Standard culture technique was implied for obtaining the chromosomes from the patient's peripheral (venous) blood for staining (banding).

Analysis of banded chromosomes was done under a microscope. Minimum 20 to 30 cells were analyzed which was enough to rule out mosacism to an acceptable level. The results were summarized and interpreted taking into account the patient's previous history and other clinical findings. The results were reported as per the norms of International System for Human Cytogenetic Nomenclature 2005 (ISCN2005) 5.

OBSERVATIONS AND RESULTS

In the present study of 100 clinically diagnosed Down syndrome patients, following things were observed.

Age incidence: Maximum number of patients was from the 0 to 5 yrs of age group while only three patients were from the age group of 21 yrs and above. The oldest patient was 42 yrs of age. (Table No. 1)

Range and Percentile of I. Q.: Range of I. Q. was minimum 30 and maximum 62. I. Q. was not recordable in 52 patients. (Table No. 2)

Sex Ratio: In 92 chromosomally confirmed Down syndrome cases of the present study, the sex ratio (Male:Female ratio) was 1.24 with a male predominance.

Clinical Profile: Various clinical features of Down syndrome were observed in the present study. Apart from mental retardation, delayed developmental milestones are noted in maximum patients. Other features listed are also the commonly observed one in Down syndrome. (Table No. 3)

Table No. 1
Age-wise distribution of all subjects

Age Groups	Number of Cases	Percent
0 to 5	56	56 %
6 to 10	20	20 %
11 to 15	16	16 %
16 to 20	5	5 %
21 & >	3	3 %
Total	100	100 %

Table No. 2
Percentiles of I. Q. in all subjects

Percentiles	I. Q.
5	38
10	40
25	43.25
50	48

75	55.75
90	60
95	62

Table No. 3 - Various clinical features observed in all subjects

Feature	No. Present	Percent (n = 100)
Delayed Milestones (n = 86)	81	94.19%
Short stature	47	47%
Hypotonia	60	60%
Flat Occiput	68	68%
Mongoloid Slanting Eyes	75	75%
Epicanthic Folds	71	71%
Malformed, Low set Ears	67	67%
Flat Nasal Bridge	77	77%
Constantly Open Mouth	60	60%
High Arched Palate	62	62%
Protruding Furrowed Tongue	59	59%
Irregular Teeth (n = 74)	43	58.10%
Short neck	46	46%
Short broad hands	42	42%
Clinoductyly	33	33%
Simian Crease	39	39%
Gap Between First Two Toes	66	66%

DISCUSSION

The present study had been undertaken to evaluate the information obtained during the clinical examination carried out on the patients referred as probable Down syndrome cases.

All the subjects were divided into various age groups. Maximum number of patients i.e. 56 was from the 0 to 5 years of age group which was the positive point coming out from the present study.

Hines Stefani, Bennett Forrest 6 (1996) reviewed the effectiveness of early intervention programs for children with Down syndrome. They concluded that children with Down syndrome and their families are likely to be benefited by early intervention.

The most striking clinical feature in the present study was mental retardation; however, I. Q. was recordable only in 48 subjects. The average range of I. Q. was 30 to 62 (table no. 2).This finding correlated well with the standard textbooks 7, 8.

The sex (male:female) ratio of the patients with Down syndrome in the present study is 1.24 (table No. 8). In the present study, overall male predominance can be observed. This finding correlated well with the findings of the other researchers like Hook E B et al 3 (1999) and Kovaleva N V et al 9 (2001).

The typical clinical features of Down syndrome were the basis on which the subjects were selected for this study.

Mental retardation was the universal feature in all the cases selected for the study. History of delayed developmental milestones was evident in 94.19 % of the cases, which was an obvious positive finding for Down syndrome cases. Amongst the associated clinical conditions, ventricular septal defects were noted in two cases while Alopecia aerate in one. All the clinical features with their observed percentages are summarized in table No. 3.

These findings of the present study correlated well with those noted by other researchers like Lee L and Jackson J 10 (1972) and Kava M P et al 11 (2004).

In the present study, counseling of parents of the subjects was done according to the need of the situation. The parents were explained prior to the cytogenetic test (karyotyping) about its nature and outcome. In those cases where cytogenetic studies confirmed the diagnosis of Down syndrome, the counseling was done depending upon the age of the baby as well as the age of the parents and their wish to have a next baby.

A positive point outcome from the present study was that, in maximum patients the diagnosis had been made very early in their life. Therefore exposure to early intervention programs in these cases can lead to overall positive changes in their development and will enable them to live longer and more normal lives.

SUMMARY AND CONCLUSION

Down syndrome is the commonest chromosomal anomaly seen in all the live births. In the present study, various chromosomal anomalies associated with 100 Down syndrome cases were analyzed and correlated with the clinical profile. Chromosomal abnormality was confirmed in 92 cases. The most striking clinical feature observed in the present study was mental retardation. In maximum patients the diagnosis of Down syndrome had been made between 0-5 years of age. This was important for their better management. Counseling of parents of the subjects was done accordingly.

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