

Cytogenetical and Anthropometric Studies on Clinically Diagnosed Patients with Down Syndrome

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ABSTRACT The blood samples of 9 children and 1 male adult individual (Bengalee parentage) who were clinically diagnosed as Down syndrome (DS) patients were subjected to lymphocyte culture using standard method. Two cultures were unsuccessful. The karyotype analysis of 6 children (2 boys and 4 girls) and 1 adult individual (male) revealed the chromosomal anomaly of trisomy 21. The karyotype of 1 girl was normal. Anthropometric measurements of cytogenetically evidenced 6 DS children with trisomy 21 (2 boys and 4 girls) were compared with the available percentiles of Sarsuna-Barisha study (Das, 1985) of normal Bengalee children and ICMR study (1984) of Indian children for understanding retardation of growth, if any, in these children affected with DS. The results indicated that the boys and girls with DS suffered from growth failure as they do not perpetually succeed to attain the expected body height and biacromial diameter (Sarsuna-Barisha study) and body height, weight and head and chest circumferences (ICMR study) in their respective age groups. In spite of limited sample size considered in this preliminary study, the present findings are consistent with the fact that growth in children with DS is retarded than that of the normal children in the Bengalee population.

INTRODUCTION

One of the best recognized and common congenital chromosomal disorders is Down syndrome (DS) which leads to varying degrees of mental retardation and a spectrum of phenotypic abnormalities in man. Lejeune et al. (1959) discovered the etiology of DS. It is now well established that the chromosomal basis for DS is approximately 95% of affected individuals is trisomy 21 secondary nondisjunction during meiosis. The cytogenetic studies on DS in our population are scanty (Verma et al., 1979; Krishnamurthy et al., 1981). The growth of children with DS

has been noted to be retarded (Hall, 1964; Thelander and Prior, 1966; Rarick et al., 1975; Pueshel et al., 1976). Although it is known that clinical features of DS condition include growth retardation, there exists no work on growth studies of children affected with DS in our population.

In this preliminary note an attempt has been made to prepare karyotype for chromosome analysis in clinically diagnosed DS patients and to compare anthropometric dimensions of cytogenetically evidenced DS patients (growing children) with that of the available percentile values of normal children in Bengalee population for understanding retardation of growth, if any, in these children affected with DS.

MATERIAL AND METHODS

The material for the present study is consisted of peripheral blood samples from 10 individuals. Out of these, blood samples of 9 children were collected from Dr. B.C. Roy Memorial Hospital for Children, Calcutta. These children were clinically diagnosed as DS patients by the pediatricians of the hospital. The blood sample of these 9 children were subjected of lymphocyte culture for chromosome analysis. The blood sample of one individual aged 25 years in the study was collected by personal effort.

The sample description of DS patients are presented in table 1. Anthropometric measurements of the subjects included in the study were taken following the method of Weiner and Lourie (1981). Age determination of all the children measured in this study

was made by verifying the birth certificates issued by a hospital or a nursing home. In this preliminary study the descriptive nature of comparison of anthropometric measurement values of DS children against percentiles of normal children was followed as a method to understand growth failure in DS children. The available percentile values of anthropometric measurements of Sarsuna-Barisha data (Das, 1985), height and biacromial diameter have been compared with the present study. Further, a comparison has also been made with percentile values of ICMR study (1984) of Indian children (height, weight, head circumference and chest circumference).

Table 1 : Sample description

S. No.	Mother's age (in years)	Parity	Age of the patients	Sex
1	22	1	4m.20d.	M
2.	19	2	1yr.2m.	F
3.	34	1	2yr.4m.	F
4.	25	1	3yr.6m	F
5.	25	1	1yr.4m	F
6.	26	1	2yr.	M
7.	24	3	1yr.6m.	F
8.	52	4	25yr.	M

The lymphocyte culture for chromosome analysis was done following Verma and Babu (1989) with some suitable modifications. The prepared slides with chromosome metaphases were stained in buffered giemsa. Giemsa stained metaphases were scanned under binocular microscope (Nikon, Japan) fitted with computerised camera. Five well spread metaphases showing good staining were counted for total number of chromosome under oil immersion and one metaphase for each individual was photographed on slow speed black and white film (Orwo NP 22; 35mm; 125ASA).

RESULTS AND DISCUSSION

Chromosomal analysis was performed on 10 individuals clinically diagnosed as DS pa-

tients. Two cultures were unsuccessful. The karyotype analysis of 6 children (2 boys and 4 girls) and 1 adult individual (male) revealed the chromosomal anomaly of trisomy 21. The karyotype of 1 girl (serial No.4) was normal. The anthropometric measurements carried out on each individual were presented in table 2. The available percentiles in case of Sarsuna-Barisha study (Das, 1985) are height and biacromial diameter and in case of ICMR study (1984) are for height, weight and head and chest circumferences. The comparisons of these anthropometric dimensions

Table 2 : Anthropometric measurements of Down syndrome patients

S. No.	Body height (cm)	Weight (kg)	Chest circumference (cm)	Head circumference (cm)	Biacromial diameter (cm)
1	57.0	5.0	40.0	40.0	15.0
2.	76.0	8.0	42.0	39.0	17.8
3.	75.0	8.0	47.0	43.0	12.4
4.	83.4	12.0	48.0	43.0	19.2
5.	72.3	7.0	44.9	42.3	13.6
6.	70.6	7.5	45.0	41.5	16.9
7.	76.1	8.0	45.7	41.0	15.4
8.	144.5	46.0	86.0	52.5	30.4

of 6 children (2 boys and 4 girls) with the Sarsuna-Barisha study were represented in figure 1a-1d. The body height of boys are below 10th percentile (Fig.1a) of Sarsuna-Barisha children of similar age whereas body heights of girls belonging to serial No. 3, 5, 7 and 2 are below 10th, 50th, just above 50th and 97th percentiles respectively (Fig. 1b). The biacromial diameter of children with DS in both sexes is rarely above 10th percentile of Sarsuna-Barisha children of similar age (Fig. 1b). The biacromial diameter of children with DS in both sexes is rarely above 10th percentile of Sarsuna-Barisha children of similar age (Fig. 1c, 1d) except one girl (serial No. 2, Fig. 1d). Thus the results clearly indicate that the boys and girls with DS suffered from growth failure as they never attained expected height and

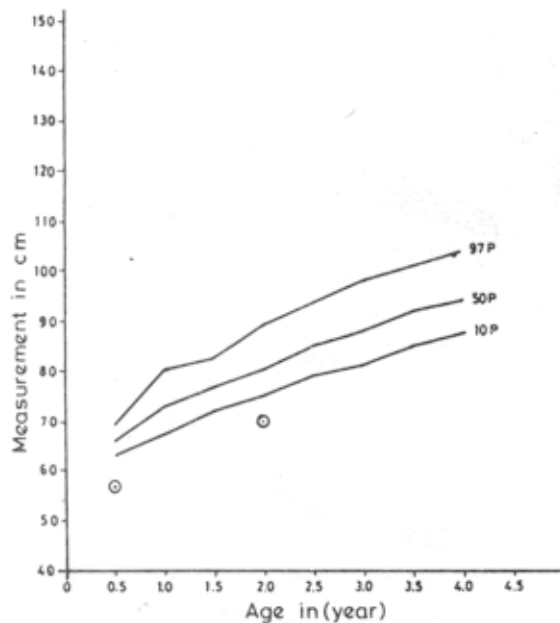


Fig.1a. Body height of Down syndrome patients (boys), Comparison with percentiles of Sarsuna-Barisha growth study

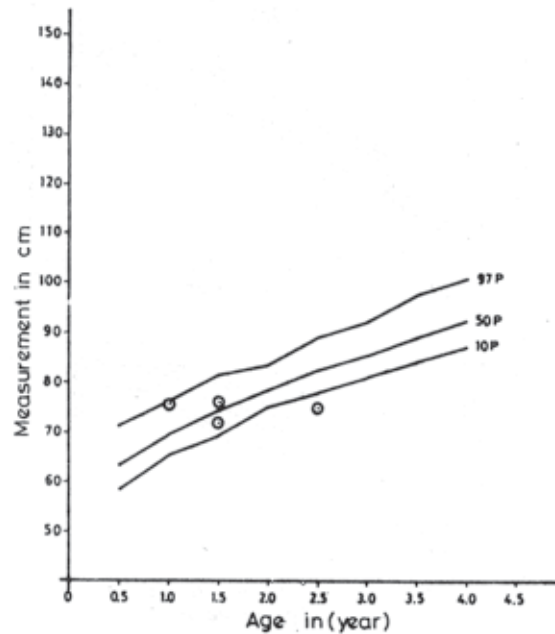


Fig.1b. Body height of Down syndrome patients (girls), Comparison with percentiles of Sarsuna-Barisha growth study

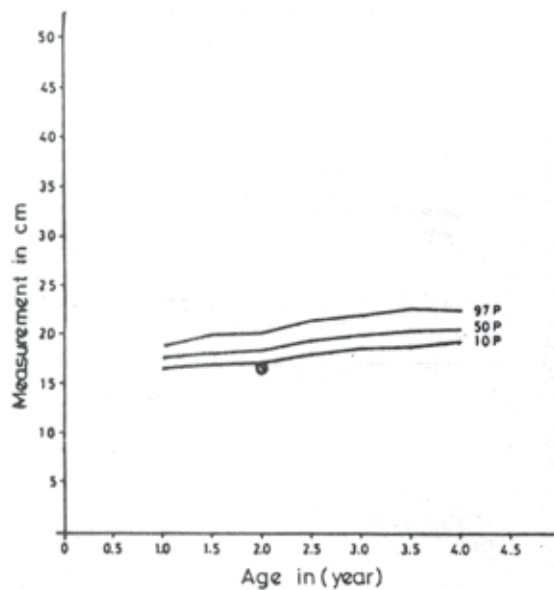


Fig.1c. Biacromial diameter of Down syndrome patient (boy), Comparison with percentiles of Sarsuna-Barisha growth study.

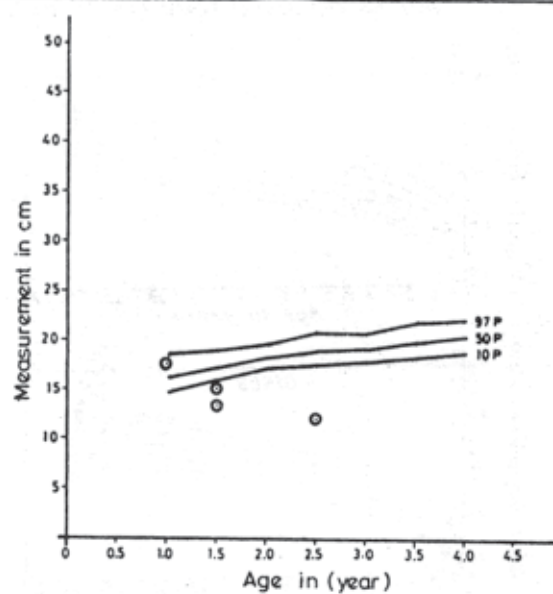


Fig.1d. Biacromial diameter of Down syndrome patients (girls), Comparison with percentiles of Sarsuna-Barisha growth study.

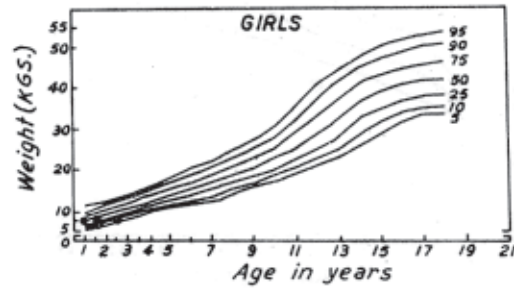
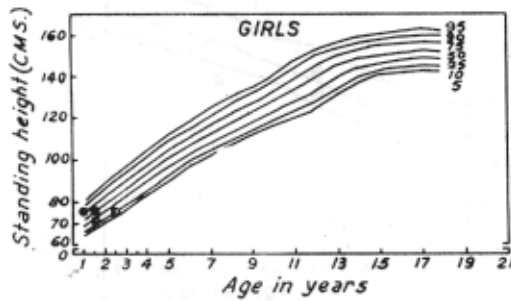
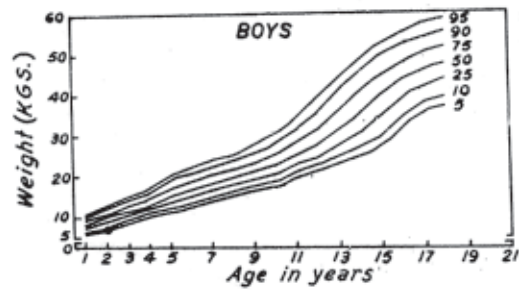
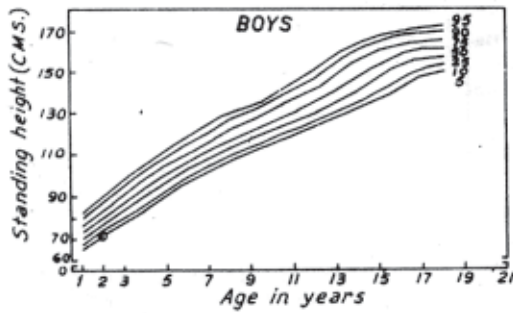


FIG. 2a. Body height of Down syndrome patients (boys and girls). Comparison with percentiles of ICMR growth study.

FIG. 2b. Body weight of Down syndrome patients (boys and girls). Comparison with percentiles of ICMR growth study.

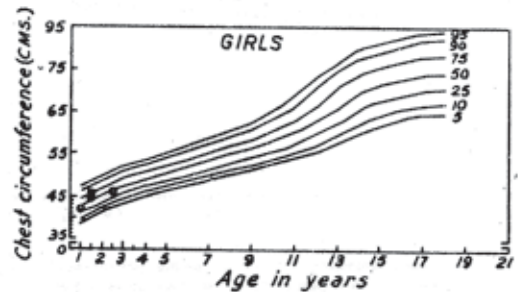
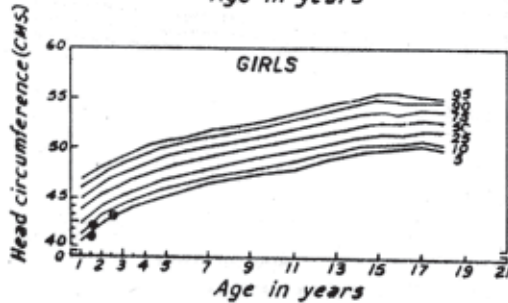
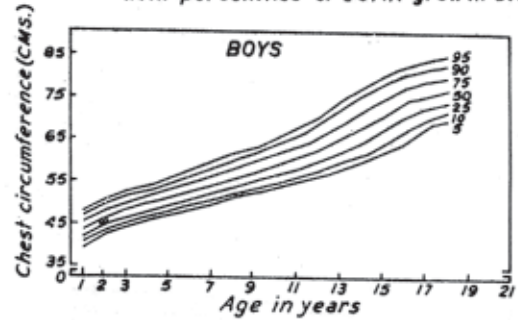
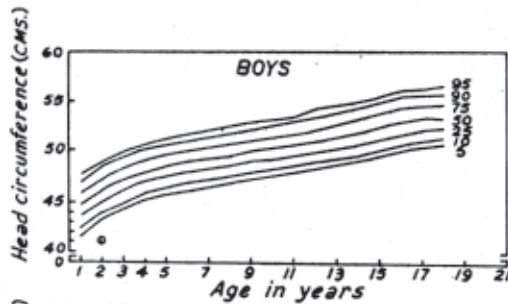


FIG. 2c: Head circumference of Down syndrome patients (boys and girls). Comparison with percentiles of ICMR growth study.

FIG. 2d: Chest circumference of Down syndrome patients (boys and girls). Comparison with percentiles of ICMR growth study.

biacromial diameter in their respective age groups when compared with that of the normal Bengalee children. Our findings are in conformity with the study of Buday (1990) done among the Hungarian children with DS. Growth failure is characteristic of children with DS have been reported in earlier studies (Dutton, 1959; Benda, 1960; Gollesz, 1963; Thelander and Prior, 1966; Rundle, 1970; Rarick et al., 1975).

When anthropometric measurements of each DS child of the present study were compared with available percentiles of Indian children of both sexes with respect to height, weight and head and chest circumferences (Figs. 2a-2d), they are in general below 50th percentile. The 50th percentiles (medians) which approximate with mean values. In other words, it may be said that DS children in the present study are always below the mean values of height, weight and head and chest circumferences of Indian children. Thus the growth of DS children is delineated with respect to those anthropometric dimensions than the Indian children.

The limitation in the present study is that the sample size of DS children is not adequate. A sizeable number of DS children in same age group and comparison of their averages with that of percentile would have more valid and meaningful. But this preliminary study indicates that present findings are consistent with the fact that growth in children with DS is retarded than that of normal children in Bengalee population.

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REFERENCES

- Benda, C. E. : *Mongolism and Cretinism*. Grune and Stratton, New York (1960).
- Buday, J. : Growth and physique in Down Syndrome children and adults. *Humanbiol. Budapest*, 20 : 1-126 (1990).
- Das, S. R. : *Mixed Longitudinal Growth Data for 22 Measures : The Sarsuna Barisha Series, West Bengal, India. Vol. 1, Boys ; Vol. 2, Girls*. Anthropological Survey of India, Calcutta (1985).
- Dutton, G. : The physical development of mongols. *Arch. Dis. Child. Hlth.*, 34 : 46-50 (1959).
- Gollesz, V. : Retardation of somatic development in Down's disease (Mongolism). *Acta Morph.*, 12 : 85-102 (1963).
- Hall, B. : Mongolism in newborns : A clinical and cytogenetic study. *Acta Paediat. Scand. Suppl.*, 154 : 1-94 (1964).
- ICMR : Growth and Physical Development of Indian Infants and Children. *Technical Report Series No. 18*, ICMR, New Delhi (1984).
- Krishnamurthy, D.S., Patel, Z. M., Khanna, S.A., Danthi, V. and Ambani, L. M. : Frequency of trisomy 21, translocation, mosaicism and familial cases of Down's syndrome in Bombay. *Ind. J. Med. Res.*, 73 : 218-222 (1981).
- Lejeune, J., Gautier, M. and Turpin, R. : Le mongolisme, premier exemple d'aberration autosomique humaine. *Ann. Genet. (Paris)*, 1 : 41-49 (1959).
- Pueshel, S.M. and Rothman, K. O. I. : Birth weight of children with Down's syndrome. *Am. J. Ment. Defic.*, 80 : 442-445 (1976).
- Rarick, G. L., Wainer, H., Thissen, D. and Seefeldt, V. : A double logistic comparison of growth patterns of normal children and children with Down's syndrome. *Ann. Hum. Biol.*, 4 : 339-346 (1975).
- Rundle, A. T. : Anthropometry : A ten year survey of growth and sexual maturation in mental subnormality : In : *Modern Trends in Reseach*. B. W. Richards (Ed.) Pitman Medical and Scientific Publishing Co. Ltd., London (1970).
- Thelander, H. F. and Prior, H. B. : Abnormal patterns of growth and development in mongolism. *Clin. Paediat.*, 5 : 493-501 (1966).
- Verma, R. S. and Babu, A. : *Human Chromosomes*. Pergamon Press, New York (1989).
- Verma, I. C., Malhotra, A. K., Malik, G. R. and Ghai, O. P. : Cytogenetic profile of Down's syndrome in India. *Ind. J. Med. Res.*, 69 : 147-151 (1979).
- Weiner, J. S. and Lourie, J. A. : *Practical Human Biology*. Academic Press, New York (1981).