CONGENITAL HEART DISEASE IN DOWN SYNDORME: AN ECHOCARDIOGRAPHIC STUDY

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ABSTRACT

We evaluated the utility of echocardiography in assessing the frequency and nature of cardiac malformations in children with Down syndrome. Fifty cases of chromosomally proven Down syndrome were studied. A physical examination, electro cardiogram, radiograph of chest and two-dimensional echocardiography was performed on all patients. Twenty-two (44%) children had heart diseases. Endocardial-cushion-defect was the commonest anomaly, followed by ventricular septal defect. Three children with heart disease were asymptomatic and had normal X-ray films of chest and ECGs. The prevalence and specific type of congenital heart disease in this study is comparable to the studies using invasive means for diagnosis. The study further suggests that clinical examination of the cardiovascular system alone may not be sufficient in detecting heart disease. Two-dimensional echocardiography offers an excellent non-invasive tool for diagnosing cardiac malformations in Down syndrome.

Key words: *Down syndrome, Cardiac malformations, Echocardiography.*

Down syndrome is the commonest chromosomal anomaly in live born infants, occurring with a frequency of about 1 : 800 newborns in the West(1) and about 1 : 920 births in India, based on an analysis of 75,103 births from eight hospitals(2). Cardiac malformations are a common cause of death in these children(3). Recent advances in cardiac surgery have led to prolongation of life span in patients of Down syndrome with heart disease. It is, therefore, important to detect and accurately evaluate heart disease at an early stage in this high risk population. The frequency of congenital cardiac defects varies with the investigative procedures used. Echocardiography is likely to yield more information than routine clinical examination and tests like electrocardiography and plain radiograph of the chest, while avoiding the disadvantages of invasive investigations.

The present study was conducted to evaluate the utility of echocardiography in assessing the frequency and nature of congenital cardiac malformations in children with Down syndrome in North India.

Material and Methods

Fifty consecutive patients of Down syndrome attending the Genetics Clinic of All India Institute of Medical Sciences from August, 1985 through December, 1986, were enrolled in this study. A clinical diagnosis of Down syndrome was made after a detailed history and clinical examination

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Received for publication: December 12, 1991; Accepted: May 6, 1992

using criterion described by Oster(4). The diagnosis was confirmed by chromosomal analysis of peripheral blood leukocytes, using the microblood culture technique of Arakaki and Sparkes(5). Trypsin banding was carried out by the Seabright method(6). Forty-five patients had regular trisomy, four had translocation and one had a mosaic pattern.

Subsequently a detailed history and physical examination pertaining to the cardiovascular system was conducted. A posteroanterior roentgenographic study of the chest and a 12-lead electrocardiogram was obtained in every case. Irrespective of the clinical findings and the results of the initial investigations, all 50 patients were screened by two-dimensional echocardiography (performed on the ATL UM8 machine). Information on age, sex, cardiac physical examination, ECG and *X*-ray films and echocardiograms was recorded and tabulated.

Results

The patients for the present study were selected on a prospective basis from amongst those referred to the Genetics Clinic for confirmation of diagnosis by chromosomal analysis. The subjects were not sick children and their selection did not therefore bias the study.

The age of the children ranged from six days to 12 years (mean = 2.16 years). Twenty-nine patients were less than one year of age. Twenty-two out of 50 (44%) children had heart disease. A thorough clinical examination, an electrocardiogram and a radiograph of the chest enabled us to detect a cardiac anomaly in 19 of these 22 patients (86.4%). In the remaining three patients the diagnosis was made by echocardiography. These patients had a closing VSD (5 months), cleft mitral valve leadflet (4 years) and a bicuspid aortic valve (4 months).

The pattern of cardiac involvement is depicted in *Table I*. Endocardial cushion defect was the commonest anomaly (31.7%), closely followed by ventricular septal defect (27.2%). Pulmonary arterial hypertension was detected in five patients (age range 2.5 months-12 years). Four of these patients had ventricular septal defect and one had a patent ductus arteriosus.

Discussion

The life expectancy of patients with trisomy 21 remains less than that of the general population, but there seems little doubt that it is rising steadily(7). This is the result of better medical care and advances in cardiac surgery, as cardiac lesions have

TABLE I—*Pattern of Cardiac Involvement in Down Syndrome (n = 50)*

Cardiac lesion		Number	Per cent
Endocardial cushion defect		7	31.7
Complete	2		
VSD of ECD type	4		
ASD (ostium primum)	1		
Ventricular septal defect		6	27.2
Atrial septal defect (secundum)		3	13.6
Patent ductus arteriosus		2	9.1
Tetralogy of Fallot		1	4.6
Biscuspid aortic valve		1	4.6
Cleft mitral valve leaflet (isolated)		1	4.6
Dextrocardia with situs inversus and normal heart		1	4.6
Total		22	100.0

ECD = Endocardial cushion defect.
VSD = Ventricular septal defect.
ASD = Atrial septal defect.

been responsible for a large proportion of the early mortality. Murdoch(8) compared the morbidity of children with congenital heart disease and Down syndrome and those with congenital heart disease alone, and demonstrated that infants with congenital heart disease and Down syndrome were not at higher risk of dying than infants with congenital heart disease alone. This underscores the necessity of an early diagnosis and management of congenital cardiac anomalies in patients of trisomy 21.

Forty-four per cent of the subjects with Down syndrome had congenital cardiac malformations in the present study. This prevalence is comparable to the prospective study conducted by Rowe and Uchida(9), who found an incidence of 40.2%, using invasive investigations. Endocardial cushion defect was the commonest lesion found in the present study, followed by ventricular septal defect. These findings are consistent with those of several other authors(9-11).

The pattern of cardiac involvement in Down syndrome in the present study is compared with a composite of three studies using various methodologies in *Table II*.

TABLE II—*Pattern of Cardiac Malformations: A Comparison of the Present Study with a Composite of Three Studies (9-11)*

Defects	Composite of three studies	Present study
No. of cases	332	22
Endocardial cushion defect	40.4%	31.7%
Ventricular septal defect	32.2%	27.2%
Atrial septal defect	10.2%	13.6%
Patent ductus arteriosus	5.7%	9.1%
Tetralogy of fallot	5.1%	4.6%
Others	6.1%	13.8%

Patients of Down syndrome with congenital heart disease have been reported to have an unusually high incidence of pulmonary artery hypertension and pulmonary vascular obstructive disease(12). Pulmonary artery hypertension was detected in five patients in the present study (22.8%). The youngest patient was 2½-month-old and had a ventricular septal defect of the endocardial cushion defect type. It is, therefore, advisable to carry out early cardiac evaluation of all patients of Down syndrome using 2D-echocardiography, and to institute proper treatment before the onset of pulmonary vascular disease. A recent study by Tubman and colleagues made similar recommendations(13).

In conclusion, this study indicates that clinical examination of the cardiovascular system alone may not be sufficient in deciding the presence or absence of heart disease in patients of Down syndrome. Two-dimensional echocardiography provides an excellent non-invasive tool for determining the presence of heart disease and for delineating the exact anatomic details of the cardiac malformations. 2D-echocardiography should be carried out in infancy in every case of Down syndrome.

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NOTES AND NEWS

XIII ANNUAL CONFERENCE OF A.P. STATE CHAPTER OF INDIAN ACADEMY OF PEDIATRICS

The XIII Annual Conference of A.P. State Chapter of IAP is to be held on *7th and 8th November, 1992* at Kalabharati, Pitapuram Colony, Visakhapatnam. The Scientific Programme includes eminent pediatricians from all over the country. The programme also includes award paper sessions: (*i*) Dr. Y.R. Reddy Gold Medal for Pediatricians below the age of 40 years, and (*ii*) Dr. G.V.R. Gold Medal for Post-graduate students for the work done in neonatology in Andhra Pradesh. Please send 3 copies of full text for award papers and 3 copies of abstracts for the free paper session before *15th September, 1992* to: Dr. K.V. Raghava Rao, Secretary, School Health Clinic, Niloufer Hospital, Hyderabad 500004.

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