

CARDIOVASCULAR MALFORMATIONS IN MALAYSIAN NEONATES WITH DOWN'S SYNDROME

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ABSTRACT

A prospective study was done to determine the incidence of cardiovascular malformations in neonates with Down's syndrome. 17/34 (50%) of the babies with Down's syndrome born at the Maternity Hospital, Kuala Lumpur, Malaysia had congenital heart defects. These included 7 cases of ventricular septal defect (VSD), 3 cases of patent ductus arteriosus (PDA), 2 cases of atrio-ventricular canal defect, 2 cases of ventricular septal defect with patent ductus arteriosus, 1 case of hypertrophic cardiomyopathy, 1 case of hypertrophic obstructive cardiomyopathy and 1 case of complex cyanotic heart. Only 8/17 (47%) of these babies had any clinical signs suggesting underlying cardiac defects. In view of the common occurrence of cardiac anomalies, it is recommended that echocardiographic screening should be carried out on all neonates with Down's syndrome.

Keywords: Down's syndrome, cardiovascular malformations, Malaysian neonates

SINGAPORE MED J 1990; Vol 31: 474 - 476

INTRODUCTION

The association of cardiovascular disease with Down's syndrome is well recognised and a wide variety of cardiovascular malformations have been identified in these babies. The incidence of cardiovascular malformations in Down's syndrome is reported to be about 40%⁽¹⁻⁵⁾. The true incidence of congenital heart disease in Down's syndrome is unknown and it is estimated to be 0.36 to 0.64 per 1000 livebirths^(6,7). We present here a study of the types of cardiovascular malformations present in neonates with Down's syndrome.

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METHODOLOGY

From 1 July 1986 till 31 December 1987, 34,522 livebirths were born at the Maternity Hospital, Kuala Lumpur. During routine screening examination, all neonates diagnosed as having Down's syndrome were referred to the Paediatric Cardiologist in the first week of life. The method used in the diagnosis of Down's syndrome is described elsewhere⁽⁸⁾. A full clinical examination of the cardiovascular system, chest x-ray, electrocardiogram and an echocardiographic examination with an Aloka PS 188 echomachine were done on these babies.

RESULTS

There were 34 neonates born with Down's syndrome. Seventeen (50%) of these neonates were found to have congenital heart defect on echocardiographic examination. Thus during the study period, the incidence of Down's syndrome with congenital heart defect in the Maternity Hospital, Kuala Lumpur was 0.49 per 1000 livebirths.

The echocardiographic findings were as follows: of the 34 neonates, 11/34 (32.4%) of the babies had a completely normal cardiovascular system, 2/34 (5.9%) had a patent foramen ovale (PFO) and 3/34 (8.8%) had a closing patent ductus arteriosus (PDA) and 1/34 (2.9%) had a combined PFO and closing PDA (on follow up echocardiography, these closing PDAs were noted to have closed). If we consider that the PFOs and closing ductus arteriosus as normal variants of the neonatal cardiovascular system at birth, then these 17 babies (50%) had a normal cardiovascular system at birth. The echocardiographic findings of the remaining 17 neonates are listed in Table I.

Table I
Abnormal echocardiographic findings at birth in babies with Down's syndrome born at the Maternity Hospital, Kuala Lumpur, Malaysia. (1 Jul 1986 – 31 Dec 1987).

Echocardiographic findings	No. of cases	Percentage (%)
Ventricular septal defect (VSD)	7	41.1
Patent ductus arteriosus (PDA)	3	17.6
Atrio-ventricular canal defect	2	11.8
VSD + PDA	2	11.8
Hypertrophic cardiomyopathy	1	5.9
Hypertrophic obstructive cardiomyopathy	1	5.9
Complex cyanotic heart (VSD + PDA + severe PS)	1	5.9
TOTAL	17	100

PS : Pulmonary stenosis

The baby with the hypertrophic cardiomyopathy died on Day 1 of life with cyanosis, respiratory distress and cardiac failure. Unfortunately, no postmortem was performed.

Of the 17 babies with echocardiographically proven congenital cardiac defects, only 8 (47%) had clinically abnormal cardiac findings on clinical examination and 9 (53%) had normal findings on clinical examination (Table II).

DISCUSSION

The incidence of Down's syndrome with congenital heart defect at the Maternity Hospital, Kuala Lumpur of 0.49 per 1000 livebirths is comparable with the results obtained from other studies where the incidence was quoted as between 0.36 - 0.64 per 1000 livebirths⁽⁶⁾.

We find that the Malaysian neonates with Down's syndrome had equally high proportion of congenital cardiac abnormalities as was reported in other series (Table III). In our study, we have considered those babies with PFO, closing PDAs or both as normal.

The commonest cardiac defect in our Malaysian neonates with Down's syndrome was Ventricular Septal Defect (VSD) followed by PDA. This is in contrast to

Table II
Comparison between echocardiographic diagnosis and clinical findings of neonates with Down's syndrome.

Patient	Diagnosis	Clinical Finding
1. S	A-V canal defect	NAD
2. CPY	A-V canal defect	NAD
3. F	VSD	Loud S2, no murmur
4. P	VSD	Systolic murmur
5. NKL	VSD	Systolic murmur
6. NH	VSD	NAD
7. NZ	VSD	Systolic murmur
8. VNN	VSD	Systolic murmur
9. SE	VSD	Systolic murmur
10. J	VSD + PDA	NAD
11. TK	VSD + PDA	NAD
12. HZ	PDA	NAD
13. TI	PDA	NAD
14. NSM	PDA	NAD
15. LFL	HOCM	NAD
16. AZ	Hypertrophic cardiomyopathy	Systolic murmur
17. R	VSD + PDA + PS	Cyanosis, no murmur

NAD : No abnormality detected

HOCM : Hypertrophic obstructive cardiomyopathy

PS : Pulmonary stenosis

AV Canal defect : Atrio ventricular canal defect

Table III
Comparison of percentage of babies with Down's syndrome who had congenital heart defect as was reported by different authors.

Author	Total no. of cases	No. of babies with congenital heart defect	Percentage with congenital heart defect (%)
Evans (1)	63	28	46.0
Berg (2)	141	79	56.0
Liu/Corlette (3)	44	16	36.3
Rowe/Uchida (4)	174	70	40.2
Cullum (5)	80	40	50.0
Total	502	233	46.4
Our study	34	17	50.0

Table IV
Types of Congenital Heart Defects in Babies with Down's Syndrome Reported by Different Authors

Type of heart defect.	Rowe/Uchida ⁽⁴⁾	Greenwood ⁽⁹⁾	Park ⁽⁶⁾	Laursen ⁽¹⁰⁾	Our Study
A-V Canal defect	36%	48.7%	34.5%	24.7%	11.8%
A-V Canal defect + PDA	—	—	6.8%	1.2%	—
A-V Canal defect + pulmonary stenosis	—	—	4.3%	1.2%	—
Secundum atrial septal defect	9%	2.6%	8.4%	—	—
Ventricular septal defect (VSD)	33%	28.7%	21.3%	37.6%	41.1%
VSD + PDA	—	—	7.2%	5.9%	11.8%
VSD + Pulmonary stenosis	—	—	4.7%	—	—
Persistent ductus arteriosus (PDA)	10%	6.9%	4.7%	5.9%	17.6%
Other congenital heart defects	12%	13.1%	8.1%	15.3%	17.7%
TOTAL	100.0%	100.0%	100.0%	100.0%	100.0%

what was reported by others where atrio-ventricular defect followed by VSD were the 2 commonest cardiac malformations in Down's syndrome (Table IV)^(4,6,9,10).

It is also interesting to note that 9 cases (53%) of our 17 babies with Down's syndrome who had echocardiographically proven congenital cardiac lesions had no clinical features to suggest underlying cardiac abnormalities. Thus unless echocardiographic screening examination was carried out on all neonates with Down's

syndrome, congenital cardiac abnormalities might be missed.

ACKNOWLEDGEMENT

We would like to thank Professor Datuk (Dr) M N Mahmud, Dean, Faculty of Medicine, National University of Malaysia for giving us permission to use the data from the Maternity Hospital, Kuala Lumpur.

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