ORIGINAL ARTICLE

# Delayed recognition of congenital heart disease

**M M Massin, H Dessy**

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See end of article for authors' affiliations

Correspondence to: Professor M Massin, Division of Paediatric Cardiology, CHR Citadelle (University of Liège), Boulevard du 12è de Ligne, 1, B-4000 Liège, Belgium; martial.massin@chrcitadelle.be

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**Objective:** The aim of this prospective study was to evaluate the proportion of children with delayed recognition of congenital heart disease (CHD).
**Methods:** Of the 744 children with CHD primarily diagnosed during a 10 year period in one hospital, the patients were identified where the diagnosis of CHD was established with a significant delay.
**Results:** Sixty six patients (8.9%) had delayed diagnosis of CHD. Among patients with cyanotic CHD, 10.4% (7 of 67 cases) were referred after they had initially been discharged home from the birth clinic. Among patients with acyanotic CHD, 8.7% (59 of 677) of all children and 35.1% (59 of 168) of the children who required surgery or interventional catheterisation were referred at an age where elective repair should have already been performed or needed immediate treatment because of their haemodynamic status. Of the 66 patients with delayed diagnosis, one infant with cor triatriatum died at admission because of delayed referral and 10 children had severe complications: preoperative cardiogenic shock in seven cases of aortic coarctation and one case of endocardial fibroelastosis, pulmonary hypertensive crisis in one child after delayed repair of a ventricular septal defect, and infectious endocarditis after dental care in a teenager with undiagnosed moderate aortic stenosis, who required Ross operation a few months later.
**Discussion and conclusion:** A substantial proportion of CHD was detected with relevant delay. In all cases of late diagnosis, clinical cardiac findings were present that should have alerted the physician on the possible presence of underlying CHD.

Most of the relevant congenital heart defects (CHD) are recognised by the neonatal clinical screening or even in utero by fetal echocardiographic screening. Nevertheless, a substantial percentage of CHD are missed in the early clinical screening, and are diagnosed after discharge from hospital during childhood[1] or even during adulthood.[2] Often, this delay in making the correct diagnosis early in life affects the overall outcome of the children in whom an underlying relevant cardiac defect was not detected until rapid action had to be taken.[3–4] On the other hand, the paediatrician is confronted with a significant number of young patients with murmurs that are innocent, and significant cardiac defects may be masked by the presence of a persistent arterial duct and by multiple compensatory mechanisms, thus making clinical diagnosis of CHD difficult and inaccurate in a significant number of cases.[5–6]

It was the aim of this study, to evaluate the proportion of children with delayed cardiac diagnosis in a large cohort of consecutive paediatric patients seen in a single hospital. Furthermore, it was the purpose of the present evaluation to focus on the complications attributable to a delayed diagnosis and on the physical cardiac findings that were not recognised as such by the referring physicians.

## PATIENTS AND METHODS

A prospective study was performed over a 10 year period ending in June 2005 at CHR Citadelle in Liège, Belgium. The main part of the department of paediatrics of the University of Liège, including neonatal intensive care unit and paediatric outpatient clinic, is located in this hospital. During the study period, the division of paediatric cardiology offered the full range of non-invasive testing, as well as diagnostic and interventional catheterisation and simple cardiac surgery. Cardiac patients requiring neonatal or complex surgery were referred to another cardiac centre, mostly to Queen Fabiola Children's University Hospital, Free University of Brussels, Belgium.

All consecutive paediatric patients in whom a diagnosis of CHD was made postnatally during the study period were prospectively indexed. The defects were classified as cyanotic CHD or acyanotic CHD according to usual standards. Excluded from the series were children with prenatal suspicion or diagnosis of CHD, those with bicuspid aortic valve with no stenosis or regurgitation, and those with patent arterial duct or left to right interatrial shunt not present beyond the age of 3 months.

We evaluated the proportion of indexed children with delayed cardiac diagnosis. As in a previous study,[4] these included patients with cyanotic CHD that was only discovered after they had initially been discharged home from the birth clinic. Patients with acyanotic CHD were included, in case the defect was diagnosed at an age where elective cardiac repair should have already been performed, according to contemporary standards of paediatric cardiology, or in case immediate treatment was indicated at initial evaluation because of the patient's haemodynamic status. For the heart defects most frequently involved, the recommended age for correction was defined as being $<$10 years of age for atrial septal defect,[7] $<$6 months of age for complete atrioventricular canal and large ventricular septal defect, $<$18 months of age for patent arterial duct, and $<$3 years for aortic coarctation.[8] For other defects with increasing severity throughout early life, correction was recommended before the onset of symptoms. We then analysed the morbidity and mortality associated with a late diagnosis in such patients, and the clinical cardiac findings that were not recognised before patient referral by the referring physician but were noted during the paediatric cardiologist's specialised examination.

## RESULTS

During the 10 year study period, 744 cardiac children were prospectively indexed. Of them, 235 required surgery or interventional catheterisation because of their CHD during the same time period. Sixty six patients had delayed

**Table 1** Prevalence (%) of common acyanotic chd according to the time of diagnosis

| Defects | Before discharge n = 352 | <1 month n = 57 | >1 month n = 209 | Relevant delay n = 59 |
|---|---|---|---|---|
| VSD | 59 | 61 | 31 | 7 |
| ASD | 15 | 19 | 29 | 24 |
| PDA | 9 | 4 | 7 | 22 |
| PS | 7 | 11 | 17 | 5 |
| AVSD | 3 | 0 | 0 | 0 |
| AS | 1 | 4 | 11 | 3 |
| COA | 1 | 0 | 0.5 | 25 |

VSD, ventricular septal defect; ASD, atrial septal defect; PDA, patent ductus arteriosus; PS, pulmonary stenosis; AVSD, atrioventricular septal defect; AS, aortic stenosis; COA, aortic coarctation.

diagnosis of CHD according to the criteria outlined above, it means 8.9% of the cardiac population and 28.1% of the patients required treatment. When considering the whole cardiac population, the proportion of late diagnoses was similar in the group of patients with cyanotic CHD, where 7 of 67 patients (10.4%) were referred after they had initially been discharged home from the birth clinic, compared with the group of patients with acyanotic CHD, where 59 of 677 patients (8.7%) were referred at an age where elective repair should have already been performed or needed immediate treatment because of their haemodynamic status. However, the proportion is significantly lower in cyanotic patients when considering only those who required surgery or interventional catheterisation because of their CHD during the study period (10.4% for cyanotic patients compared with 35.1% for acyanotic patients).

In seven infants, cyanotic CHD remained undiscovered after birth. At the cardiologist's initial evaluation, oxygen saturation, as assessed by pulse oxymetry, was 90% in our case of tetralogy of Fallot referred to the outpatient cardiac clinic for assessment of a systolic murmur at one month of age. It ranged between 50% and 80% in the six other cases who were all diagnosed during the second month of life. One infant with double outlet right ventricle of transposition type was referred to the outpatient clinic by a general paediatrician for assessment of cyanosis; two infants with L-transposition of the great arteries, ventricular septal defect, and severe pulmonary stenosis were referred to the outpatient clinic by a general paediatrician for assessment of cyanosis in one case and of a murmur in the other one; two infants with isolated D-transposition of the great arteries presented to the paediatric emergency department: one case had a primary complaint of feeding problems and the other one was referred by a nurse of the Birth and Children Office for cyanosis. The last infant had a total intracardiac anomalous pulmonary venous connection and was resuscitated from near miss at home. All these children were previously seen by at least one paediatrician.

In our population, 677 were diagnosed as having an acyanotic CHD. The prevalence of the most common CHD in patients in whom diagnosis was made before discharge from birth clinic (n = 352), after discharge but before one month of age (n = 57), after one month of age but before the age of elective repair (n = 209), or with significant delay (n = 59) is given in table 1. The group of patients with defects detected later than one month of age had an increased prevalence of atrial septal defect, pulmonary stenosis and aortic stenosis, and a lower prevalence of ventricular septal defect. The most frequent delayed diagnosis was aortic coarctation in 15 cases.[9] Other defects frequently missed were atrial septal defect in 14 children and patent arterial duct in 13 cases. Median age of late detection of these defects was 4 years (range 1 month to 14 years) for aortic coarctation, 5 years (range 7 months to 15 years) for atrial septal defect, and 3 years (range 1 month to 7 years) for patent arterial duct. All 12 complete atrioventricular canals were diagnosed before discharge from the birth clinic, probably thanks to a recommendation for neonatal echocardiography screening of all patients with Down's syndrome. The other cardiac malformations with delayed diagnosis were ventricular septal defect (n = 4), pulmonary valvar stenosis (n = 3), aortic valvar stenosis (n = 2), partial anomalous pulmonary venous connection with sinus venosus atrial septal defect (n = 2), double aortic arch (n = 2), systemic arteriovenous fistula (n = 1), coronary fistula (n = 1), endocardial fibroelastosis (n = 1), and cor triatriatum (n = 1).

Many clinical cardiac findings were not recognised as such by the treating physicians and should have led to earlier referral of most of the patients. In the group of cyanotic CHD, significant cyanosis was not recognised as such in six infants by previous paediatricians and in three cases by the referring physicians. All the children with cyanotic CHD had other physical findings that indicated a potential significant heart disease: increased precordial pulsation (n = 4), bad weight gain (n = 3), sweating (n = 3), hepatomegaly (n = 2), right heart sounds (n = 2), diastolic murmur (n = 1), palpable systolic thrill (n = 1), and frequent respiratory tract infections (n = 1). In all children with acyanotic CHD, clinical findings also indicated a potential significant underlying heart disease: bad weight gain (n = 27), increased precordial pulsation (n = 15), poor exercise tolerance (n = 15), hepatomegaly (n = 15), fixed split second heart sound (n = 15), continuous murmur (n = 14), frequent respiratory tract infections (n = 13), abnormal heart sound such as click or gallop rhythm (n = 9), sweating (n = 6), palpable systolic thrill (n = 3), diastolic murmur (n = 1), voussure (n = 1), and right heart sounds (n = 1). One child had a family history of CHD and another one had a polymalformative syndrome. Seven patients with delayed diagnosis were not born in Belgium and only recently immigrated to this country. The importance of blood pressure measurements and femoral pulse palpation during routine paediatric visits was highlighted by the fact that aortic coarctation remained undiagnosed in 15 instances with arterial hypertension and absent femoral pulse.[9] Finally, the lack of relevant systolic murmur probably played a part in the late diagnosis of four cases of cyanotic CHD and 15 cases of acyanotic CHD.

In this population, the delay in making an early correct diagnosis did not affect the overall outcome of the children with cyanotic CHD. Delayed recognition of cardiac defects had major consequences in 11 patients with acyanotic CHD who required treatment. One infant with cor triatriatum died after admission in the emergency room because of delayed referral. He was previously seen by different paediatricians for lack of weight gain and frequent respiratory tract infections during a six month period. Severe pulmonary hypertensive crisis with prolonged stay on the intensive care unit occurred in a 2 year old child after delayed repair of a large ventricular septal defect with pulmonary hypertension. Cardiogenic shock was seen preoperatively in seven cases of

undiagnosed aortic coarctation and one case of endocardial fibroelastosis. Finally, one 15 year old teenager with an undiagnosed moderate valvar aortic stenosis had an infectious endocarditis after dental care without antibiotic prophylaxis. The valvar function deteriorated quickly and he required a Ross operation a few months later.

## DISCUSSION

Unrecognised CHD carries a serious risk of avoidable mortality, morbidity, and handicap.[10] For years, books and reports have addressed this issue aiming at increasing the neonatologists and paediatricians awareness of the suspicious clinical findings and of the possible pitfalls in the clinical diagnosis of CHD, but the proportion of delayed diagnosis has not really changed over the years. Three recent studies again showed a high proportion of late diagnosis of CHD. Meberg *et al*[6] reported 24% of all children diagnosed as having CHD had initially been sent home as healthy from the birth clinic. Kuehl *et al*[11] found that a substantial proportion of infants with initially undiagnosed CHD had an adverse outcome with 10% of all infants dying because of CHD doing so with only postmortem diagnosis. Finally, Pfammatter *et al*[4] noted that 10% of all pediatric patients requiring intervention for CHD were diagnosed with relevant delay and that 22% had complications because of delayed referral.

The main finding of this study was that a substantial percentage of cardiac children had their CHD diagnosed after discharge from their birth clinic (44%). Moreover a significant proportion of children with a significant CHD had their disease diagnosed with a relevant delay to what should be considered adequate for the proper planning of therapeutic actions to be taken.

A substantial proportion of the affected children suffered more serious consequences that would have been avoidable with a correct diagnosis made early in life. There was even one death because of late diagnosis. To those evident complications, we must add the psychological problems associated with surgery in late childhood and teenage period, and long term medical problems such as right ventricular dysfunction for late atrial septal defect repair or systemic arterial hypertension for late aortic coarctation repair.

Another main finding confirmed the study results of Pfammatter *et al*[4]: in all the children with delayed diagnosis there would have been clinical findings that should have alerted the examining physician to a relevant underlying problem. It seems that the absence of a relevant systolic murmur refrained physicians from referring the children to a paediatric cardiologist. We must therefore insist on the fact that the lack of murmur does not exclude the suspicion of CHD. Finally, many complications because of delayed referral were seen in children with aortic coarctation that should have be seen easily diagnosed by blood pressure measurement and femoral pulse palpation.

In Belgium, routine clinical screening of apparently healthy newborn infants consists of two examinations by a paediatrician, one on the first day of life and a second one before discharge. After discharge, surveillance of the state of health is provided free of charge by paediatricians and general practitioners of the Birth and Children Office. Availability of more time in a calm and peaceful setting may improve the conditions for auscultation and clinical screening in the nursery and in the consultation points of the Birth and Children Office. Moreover, lectures, clinical reasoning sessions, and training courses in clinical services organised for medical students, but also continuing medical education resources for paediatricians and general practitioners should insist on the importance of semiology and clinical examination to improve the quality of the detection of congenital heart diseases.

## CONCLUSION

A substantial proportion of CHD were detected with relevant delay. In all study patients with late diagnosis, clinical cardiac findings were present that should have alerted the physician on the possible presence of underlying CHD. Morbidity and mortality were associated with late diagnosis.

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### Authors' affiliations

**M M Massin, H Dessy,** Division of Paediatric Cardiology, Queen Fabiola Children's University Hospital, Free University of Brussels, Belgium
**M M Massin,** Division of Paediatric Cardiology, University of Liège at CHR Citadelle, Belgium

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