Prenatal evaluation of congenital heart disease in high-risk pregnancies

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Summary: The authors report their experience of echocardiography performed on a group of 736 pregnancies with specific risk-factors for congenital heart disease (CHD).

The aim of the study was to evaluate the influence of specific risk-factors (established through the genetic counselling) in detecting fetal cardiac anomalies and diagnostic accuracy of extended fetal echocardiographic examination in high-risk pregnancies.

Twenty-seven heart defects were observed (3.6%), of these 24 were detected at ultrasound. Specificity and sensitivity were 99% and 90% respectively.

These results suggest the importance of an early detection of pregnancies at increased risk for CHD and confirm the good diagnostic accuracy of a multiple cardiac examination.

Key words: Congenital heart disease; Fetal echocardiography; High risk pregnancies.

INTRODUCTION

Congenital heart defects, with a live birth incidence of 0.8-0.9%, represent the most common of fetal anomalies and account for about 50% of death from lethal malformation in childhood. The importance of early diagnosis is emphasised by the fact that 20-30% of perinatal deaths are due to congenital heart disease ([1]).

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Fetal echocardiography is an accurate method for prenatal diagnostic evaluation of the fetus at risk for congenital heart malformations, and pregnancies with any of a number of risk-factors should be screened to identify affected fetuses. Mothers at increased risk for fetal heart defects include: those with a family history of congenital heart disease, those with diabetes, and those in whom fetal hydrops, arrhythmia or an extracardiac anomaly has been found ([2, 3]). The estimated relative risk percentage of these and other factors is listed in Table 1 ([4]).

An important step in order to select pregnancies at risk for fetal heart defects is represented by genetic counselling, which, through an accurate personal and familial anamnesis of the couple, allows the discovery of table favour conditions and the estimation of the risk that the pathological event could happen.

Table 1. – *Expected risk by genetic counselling.*

Risk factors	Relative risk (%)
Fetal factors	
Arrhythmia	20-25
Extracardiac anomalies	20-50
Nonimmune hydrops	20-80
Chromosomal abnormalities	50-80
Maternal factors	
Diabetes	3
Lithium	8
Collagenopathy	variable
Viral infections	"
X-Ray exposure	"
Familiar factors	
Previous sibling	1-3
Parental cogenital heart defects	4-17

In this study we report our experience of fetal echocardiography performed on a group of pregnant women at increased risk for congenital heart diseases.

MATERIALS AND METHODS

From a group of 2000 patients undergoing genetic counselling, we selected seven hundred and thirthy-six pregnancies with specific risk factors for congenital heart disease.

All patients were studied by an extended echocardiographic examination and followed-up. Table 2 shows the indications for fetal echocardiography in our study group.

The apparatus used was an Ansaldo AU 440, equipped with a 3.5 MHz convex transabdominal probe and an Ansaldo Esacord 80 equipped with a convex and a sectorial transducer. Fetal echocardiography included two dimensional real time assessments of the cardiovascular structures. All examinations were performed by two experienced ultrasonographers.

Table 2. – *Indications for referral.*

Indications	No. of referrals	CHD	%
Genetic factors	196	6	3.1
Teratogen exposure	78	1	1.2
Maternal infections	67	2	3
Maternal diabetes	250	9	3.6
Collagenopathy	19	0	0
Fetal arrhythmias	77	5	6.5
Extracardiac anomalies	66	4	6.0
Total	736	27	3.6

The standard views for fetal echocardiography included the four-chamber view, the left ventricular long-axis view with visualization of the aortic outflow tract, the short-axis view with visualization of the pulmonary outflow tract and ductus arteriosus and longitudinal view of the aortic arch. The inflow tract was also evaluated.

The evaluation of the fetal heart activity was performed in time-motion analysis, in atrial and ventricular districts, above all in cases of arrhythmia.

When a cardiac abnormality was detected a further evaluation was performed in collaboration with the pediatric cardiologist using Doppler colour flow.

The gestational age for scanning was, whenever possible, between 18-22 weeks.

The duration of the examination was about 25 minutes. A detailed biometric and structural evaluation of all fetuses studied was performed in order to establish the exact gestational age and to exclude other of systemic organ malformations.

We obtained autopsy reports (in cases of termination of pregnancy or of fetal or neonatal death) and postnatal echocardiograms to confirm prenatal diagnosis.

RESULTS

Complessively 27 congenital heart defects (3.6%) were observed in 736 examined fetuses at risk. Table 3 summarizes all cardiopathies encountered at our Center.

Twenty-one cardiopathies (77.7%) were isolated, six (22.2%) were associated with malformations shown ie Table 4. In 18 (66,6%) of the 27 fetuses examined a karyotyping was performed; none of these was pathological.

A case of atrial septal-defect, another one of double outlet right ventricle and one more of pulmonary stenosis were associated with bradycardia; an atrioventricular block was associated with a left ventricular hypoplasia and a ventricular extra-

Table 3. – *Spectrum of CHD observed in our Centre.*

Type of CHD	No.
Ventricular septal defect	6
Atrial septal defect	4
Atrio-ventricular canal defect	3
Cardiomyopathy	2
Single Ventricle	2
Truncus arteriosus	2
Pulmonary stenosis	2
Transposition of the great arteries	1
Tetralogy of Fallot	1
Double outlet right ventricle	1
Situs inversus	1
Left ventricle hypoplasia	1
Tricuspid dysplasia	1

systolia was associated with a tricuspid valve dysplasia.

The frequency of fetal cardiopathy related to the specific risk-factors ranged from 1.2% (teratogen exposure) to 6.5% (fetal arrhytmias).

The prenatal echocardiographic diagnosis was correct in all but 4 cases. There were in fact three false negative cases (2 atrial septal defects and an atrio-ventricular canal defect) and a false positive case (1 septal ventricular defect). Therefore the sensitivity and specificity of fetal echocardiography at our Center were 90% (27/27+3) and 99% (709/709+1) respectively. The positive predictive value was 96% (27/27+1) and the negative predictive value was 99% (709/709+3).

Table 5 shows the correlation among echographic diagnosis, outcome and pathologic results.

DISCUSSION

In our study, the frequency of fetal heart defects was sensibly higher than that evaluated in the general population, probably because we focussed our examinations on pregnancies with an elevated risk for congenital cardiac anomalies. The fetal cardiopathies observed were evenly distributed in the risk groups considered (Table 2); in any case the fetal factors risk group was characterized by a higher frequency of cardiac anomalies according to the expectation of genetic counselling (Table 1). The distribution of indications for sonography, showed in Table 2, should not be taken as representative of the general obstetric population, rather as a reflection of the referral pattern of our institution. For this reason the table cannot be used to estimate the frequency of cardiac abnormalities among all patients with the above mentioned conditions.

Crowford *et al.*, in their study reported an incidence of fetal cardiopathies higher than ours (7.5%, 74/989) probably because they also included cardiopathies suspected on the occasion of routinary scan ([5]). On the other hand Broshtein *et al*, in their study did not find any significant difference in fetal heart defect frequency between high risk (0.5%) and lor risk (0.3%) pregnancies ([6]).

Within the group of anomalous fetuses the distribution of congenital heart disease observed confirms that a more severe spectrum of pathologies is detected prenatally than in postnatal referrals by paediatric cardiologists ([7]). It also suggests that many of the fetuses identified antenatally may not survive to the point of inclusion in paediatric series.

Fetal echocardiography at our centre has allowed a detection of fetal heart defects in 90% of cases, using a multiple

Table 4. – *Extracardiac anomalies associated with fetal congenital heart disease.*

Type ofCHD	Assiciated anomalies
Atrial septal defect	ano-rectal atresia
Truncus arteriosus	esophageal atresia
Truncus arteriosus	renal agenesis
Atrial ventricular canal defect	hydrocefaly
Atrial ventricular canal defect	renal agenesis
Situs inversus	cistic higroma

Table 5. – *Outcome of fetuses with congenital heart disease detected prenatally.*

Type of CHD	G.A.	Outcome	Pathologic results
ASD	21	alive	same as ultrasound
ASD	21	alive	same as ultrasound
VSD	20	alive	same as ultrasound
VSD	21	alive	same as ultrasound
VSD	22	alive	same as ultrasound
VSD	21	alive	same as ultrasound
VSD	20	alive	same as ultrasound
VSD	21	alive	same as ultrasound
TGA	20	alive	same as ultrasound
DORV+VSD	22	PGE2	Troncus+ASD+VSD
Single ventricle	23	PGE2	same as ultrasound
Pulmonary stenosis	28	alive	same as ultrasound
Pulmonary stenosis	21	alive	same as ultrasound
Tetralogy fallot	21	alive	same as ultrasound
Cardiomyopathy	20	PGE2	same as ultrasound
Cardiomyopathy	22	PGE2	same as ultrasound
Single ventricle	23	PGE2	same as ultrasound
AVCD	22	PGE2	AVCD+DORV
Tricuspid dysplasia	22	ID	same as ultrasound
Hypoplastic left heart syndrome	22	PGE2	same as ultrasound
Situs inversus	14	PGE2	same as ultrasound
AVCD	19	PGE2	AVCD
Hypoplastic left heart syndrome	20	ID	Troncus
DORV	22	alive	DORV

ASD: atrial septal defect, VSD: ventricular septal defect, TGA: transposition of major arteries. DORV: double outlet right ventricle, AVCD: atrial-ventricular canal defect.

cardiac view and sensitivity and specificity compare favorably with data in the literature ([1, 5, 8, 9]).

In our experience, 3 of 4 diagnostic mistakes regarded septal defects: 2 atrial-septal defects (false negative cases) and 1 ventricular septal defect (false positive case). Such a defect, usually, considered a minor abnormality with a good prognosis, falls in to the category of cardiac lesions frequentely missed by fetal echocardiography ([10, 11, 12]).

CONCLUSIONS

The high incidence of fetal cardiopathy encountered in our study group reflects the important role of genetic counselling in order to select pregnancies presenting specific risk factors. The multiple cardiac view, as part of the detailed targeted examination, permits the increase of prenatal diagnosis of congenital heart diseases and the study of their possible evolution.

Early detection would allow patients to be referred at a stage when various options are open. Termination of pregnancy can be discussed if the cardiac lesion has a poor prognosis. If the pregnancy continues the parents can be prepared for the problem and for likely postnatal events consequent on the defect.

In any case an important feature in prenatal diagnosis concerns the prospects for long term survival. Transferring antenatal care to a centre with paediatric

cardiac facilities is often indicated when the neonate is likely to need specialised emergency care or when interventional procedures are appropriate. Intrauterine, rather than postnatal transfer, ensures an infant of optimum conditions for surgery and should reduce early morbidity and mortality.

It would be interesting to verify our results in a low risk population, but the specialized training that would be required makes this evaluation difficult to realize.

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