Prenatal Screening for Congenital Heart Defects

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INTRODUCTION

Congenital heart defects (CHD) that have an overall incidence of about 1% among liveborn infants constitute a major part of birth malformations and they account for about 20% of all stillbirths and 30% of neonatal death due to congenital abnormalities.[1] This figure obviously varies according to the population studied. Nevertheless, the true incidence among fetuses is difficult to estimate. The etiology of cardiovascular anomalies includes different known and unknown causes such us maternal diseases (diabetes mellitus, phenylketonuria), exposure to substances (anticonvulsants, lithium), infections (parvovirus, rubella), chromosomal abnormalities (trisomy 21,18) and specific mutant genes defects. The overall risk for aneuploidy in a fetus with CHD is estimated to be 30%.[2] In other words there seems to be an interaction of genetic factors, environment and coincidence for the development of the heart. An abnormal interaction of any of these processes may lead to a CHD. The recurrence risk of cardiac anomalies after the birth of one affected sibling, in the absence of a known genetic syndrome, is 2 to 4% depending on the type of the lesion and with two affected siblings the risk is about 10%.[3] Lethal cardiac defects and those requiring surgical repair or interventional catheterization within the first year of life are usually classified as major and have an estimated prevalence of 4/1000 live births.[4,5]

The revolutionary progress in ultrasound technology of the last decade and its successfully application to the prenatal examination has led to the diagnosis of most fetal abnormalities. In particular fetal echocardiography improved the identification *in utero* of congenital heart disease. The accurate diagnosis of a CHD in a fetus gives the potential of different management options to be considered, including counseling the parents about the natural history of the anomaly, delivery at a tertiary center reducing neonatal morbidity and mortality, termination of the pregnancy in complex uncorrectable anomalies or even, in some cases, intrauterine intervention. Prenatal detection of specific cardiac anomalies such as complete transposition of the great arteries[6] and hypoplastic left heart syndrome has been shown to improve neonatal morbidity and surgical outcome.[7] The overall detection rate of congenital heart disease is 25%, with a range of 19 to 48% in Western European countries.[8] Studies from specialist centers report the diagnosis of about 90% of defects (moderate to major defects in a high-risk population). However, due to widely different levels of the obstetric scanning expertise there is a considerable variation in reported study results. Cardiac anomalies are the most frequently missed lesions and this has important social, economic, psychological and legal implications.

Congenital heart disease being six times more common than chromosomal abnormalities and four times more common than neural tube defects is a main cause of mortality and morbidity in children. Historically, prenatal diagnosis of cardiac defects has been the point of interest of many developing screening programs. Risk factors and specific indications have been graded and patients defined as high-risk have been offered detailed fetal echocardiography. Contradictorily, most congenital heart disease (nearly 90%) is found in otherwise normal low-risk pregnancies. "Despite the high incidence of congenital heart disease, fetal echocardiography is not performed in all pregnancies at present because it requires advanced equipment (high-resolution ultrasound machine with pulsed and color Doppler facilities), an expert operator and detailed examination." Therefore, the concept of screening the normal obstetric population was introduced.

The object of a screening test is to identify, among a large population, a small group that is at sufficiently increased risk for certain disorders to justify offering more specific diagnostic test. The common criteria for screening (according to classical monographs of Wilson, Jungner and Hilliboe and World Health Organization) are easily fulfilled by fetal echocardiographic screening. Since screening the heart in a simplified fashion using the four-chamber view was first suggested by a French group in 1985,[9] evaluation of the cardiac structure during obstetric