CHAPTER 1

Introduction
INTRODUCTION

Ultrasound technology is capable to evaluate many anatomical structures of the fetus in great detail. Targeted ultrasound examination provides the possibility to diagnose a variety of structural malformations. Nowadays, screening for congenital anomalies is offered as standard obstetric care by ultrasound examination around 20 weeks’ gestation in most Western countries. Screening is the systematic application of a test, to identify individuals at sufficient risk for a specific disorder in order to benefit from further investigation or preventive action, among persons who have not sought medical attention on account of symptoms of that disorder. A prenatal diagnosis of a congenital heart defect (CHD), may lead to changes in obstetric management and allows for optimal neonatal care which may improve the neonatal outcome. The accuracy and success of detection of anomalies, depends strongly on the skills of the sonographer. CHD can be especially difficult to detect, and are still amongst the most commonly overlooked lesions in prenatal screening.

Figure 1 The four standard planes of the cardiac examination in prenatal screening are indicated.
The standard anomaly scan (SAS) was introduced in 2007 in the Netherlands. The program was introduced uniformly for all pregnant women. The national organisation makes the Dutch screening program unique, and comprises a scanning protocol and uniform training requisites of the sonographers. The quality of the screening program is continuously monitored biannually by assessment of the images of all ultrasonographers. Furthermore a minimum volume of scans per ultrasonographer per year is required. This is monitored by the 8 regional centers for prenatal screening.

The cardiac examination in a SAS comprises of 4 standard planes (Figure 1). After confirmation of the stomach and heart being situated on the left side of the fetus and the normal left sided arrangement of the aorta in the abdomen, the first plane is a transverse plane through the fetal thorax at the level of the ventricles and atria; the four-chamber view (Figure 2a). In this plane malformations of the ventricles, atria, atroventricular valves and septa are detectable. Although the four-chamber view is useful for identifying several abnormalities of the fetal heart, defects that involve the outflow tracts may not be demonstrated by an abnormal four-chamber view.

The outflow tracts are assessed in several planes. Just above the level of the four-chamber view the aorta exiting the left ventricle can be visualised by tilting and turning the transducer (Figure 2b). A bit more cranial, but again in a transverse plane, the pulmonary trunk can be visualised, arising from the right ventricle (Figure 2c). And then more cranial, the three-vessel view can been seen (Figure 2d). In this plane the spatial relationship between pulmonary trunk, ascending aorta and vena cava superior can be assessed.

The main two differences between a cardiac examination in a screening setting and a specialised echocardiographic examination are:

- The different expertise and training of the professionals performing the ultrasound (diagnostic fetal echocardiography is performed by a specialised cardiac team, including a pediatric (fetal) cardiologist. These professionals diagnose heart defects frequently and are capable to counsel the parents on the implication of the diagnosis and the treatment options)

- The heart is studied in far more detail. The examination is extended with, for example, evaluation of the valves, cardiac biometry, identification of the pulmonary and systemic veins, Doppler interrogation across the valves, assessment of the aortic and ductal arches, including neck vessels etc.
Figure 2a The four chamber view. This plane is indicated in green in Figure 1.*

Figure 2b The aorta arising from the left ventricle. This plane is indicated in red in Figure 1.*

Figure 2c The pulmonary trunk arising from the right ventricle. This plane is indicated in blue in Figure 1.*

Figure 2d The three-vessel view, with from top to bottom the superior vena cava, the cross-section of the ascending aorta and the full length of the pulmonary trunk. This plane is indicated in yellow in Figure 1.*

* Pictures from: Echoscopie in de verloskunde en gynaecologie, M.C. Haak. 2013
AIM OF THIS THESIS

The goal of this thesis was to gain insight in the performance of second trimester standard anomaly screening in detecting CHD. Although screening is performed in many countries for several years, reports on screening performance are only sparsely published. A comparison before and after introduction of a program was not possible because SAS was introduced gradually over several decades in most regions. The outcomes can be used to inform future parents about the performance of the SAS, and may provide options to improve the screening performance.

OUTLINE OF THIS THESIS

Chapter 2 is a systematic review of the literature and a meta-analysis on the prenatal detection rates of severe CHD by second trimester screening in unselected populations in Western countries. Severe CHD were defined as being potentially life threatening defects that require surgery or catheterization within the first year of life. Outcomes were evaluated and differentiated per type of heart defect, as well as in cases without additional extra-cardiac anomalies (isolated CHD).

Chapter 3 studies the effects of the introduction of the Dutch screening program in 2007 on the prenatal detection, pregnancy outcome and mortality of fetuses and neonates with CHD.

Within this cohort we focused on transposition of the great arteries in Chapter 4. Detection rates and the effect of a prenatal diagnosis on mortality and morbidity were studied in infants with a transposition.

If a CHD is suspected, the woman is referred to one of the tertiary centres for a fetal echocardiography. In Chapter 5 we studied the diagnostic accuracy of the third level echocardiography. We studied diagnostic errors in detail.

The complications in the pregnancies affected by a fetal CHD, in terms of preterm birth and fetal demise, were evaluated in Chapter 6. A rare postnatal complication in an infant with a prenatally detected left superior vena cava, which is usually not associated with complications, is described in Chapter 7. Finally, in the general discussion (Chapter 8) the current status of the SAS in relation to detection of CHD is described and future implications and recommendations are discussed. A summary of the thesis is given in Chapter 9.
REFERENCE LIST


