Chapter 1
Introduction and definition of research objectives

1.1 Introductory remarks

Perinatal mortality is in the range of 10 per 1000 births in the Western world. Reduction in perinatal mortality is predominantly determined by improved intrapartum and neonatal care. Intrauterine growth retardation, preterm delivery, and congenital disease are still major determinants of perinatal mortality and morbidity. Therefore, the prevention and early detection of congenital anomalies has become an important part of prenatal care. About 2-3 per cent of newborns have detectable congenital structural anomalies[1]. Major fetal structural anomalies are potentially associated with preterm delivery, perinatal mortality and morbidity, unwarranted obstetric surgery, and prolonged postnatal hospitalization, all of which place an emotional, social, and financial burden upon the involved families and society. Prenatal detection of a malformed fetus allows the woman a range of options, varying from pregnancy termination to possible intrauterine treatment and adjustment of obstetric management. The latter concerns the immediate availability of sophisticated neonatal care of the structurally abnormal infant. The potential advantage of prenatal diagnosis of a major, but non-lethal malformation is modification of the timing, mode, and geographic location of the delivery.

Indication-based ultrasound investigations apply to selective -level III- ultrasound imaging, aimed at women at increased risk of fetal structural anomalies. They entail a more detailed examination of fetal morphology and physiologic function than the screening-based scan. All fetal organ systems are subjected to scrupulous sonographic investigation.

Diagnostic investigations for suspected fetal pathology are based on abnormal obstetric findings that become manifest during the index pregnancy, such as polyhydramnios, oligohydramnios, severe intrauterine growth retardation, and preterm labour. These findings are associated with fetal structural anomalies. Women with pregnancies thus affected are often referred to tertiary centres for diagnostic evaluation by means of detailed ultrasonography, and invasive prenatal diagnosis if indicated.

The prevalence of fetal structural anomalies in this category approximates 35-40 per cent[2]. The anomalies are usually detected after 24 weeks of gestation. In approximately 20 per cent of cases more than one organ system is involved. This is often based on the presence of a particular syndrome or chromosomal abnormality. In advanced pregnancies, swift information on the fetal chromosome pattern is essential for optimal obstetric management.

The overall sensitivity of an indication-based ultrasound examination is approximately 85 per cent[3]. This is mainly explained by the higher level of scanning expertise in referral centres, better knowledge of genetic syndromes, the availability of superior ultrasound equipment, and the availability of diverse pediatric subspecialists for consultation.

1.2 Definition of research objectives

In this thesis emphasis lies on the indication-based prenatal sonographic detection of (intra)thoracic and (intra)abdominal fetal anomalies. Next to conventional two-dimensional real-time ultrasonography the role of colour coded Doppler ultrasonography in the evaluation of normal and abnormal fetal organ development was assessed, as part of an "Ontwikkelingsgeneeskunde" Committee of the Dutch Sickness Benefit Council (Ziekenfondsraad) during the period 1990-1993 (OG 89-023).

The following research objectives were addressed:

1. To review the literature on normal and abnormal fetal (intra)thoracic anatomy (excluding the heart), as studied by conventional two-dimensional real-time ultrasonography;
2. To determine the role of conventional two-dimensional real-time ultrasonography and/or colour coded Doppler ultrasonography in the prenatal detection and management of: (i) congenital cystic adenomatoid malformation of the lung, and (ii) congenital diaphragmatic hernia;
3. To review the literature on normal and abnormal fetal (intra)abdominal anatomy, as studied by conventional two-dimensional real-time ultrasonography;

4. To establish the role of conventional two-dimensional real-time ultrasonography and/or colour coded Doppler ultrasonography in the prenatal detection and management of: (i) fetal abdominal wall defects; (ii) obstructive bowel disease; (iii) congenital renal tract anomalies;
5. To study the relationship between prenatally diagnosed fetal (intra)thoracic and (intra)abdominal structural anomalies and fetal growth retardation.

1.3 References

1. Morrison, I. (1985). Perinatal mortality: basic considerations, Semin.Perinatol., 9, 144-150.

2. Wladimiroff, J.W., Sachs, E.S., Reuss, A., Stewart, P.A., Pijpers, L., Niermeijer, M.F. (1988). Prenatal diagnosis of chromosome anomalies in the presence of fetal structural defects, Am.J.Med.Genet., 29, 289-291.

3. Wladimiroff, J.W. (1996). Routine ultrasonography for the detection of fetal structural anomalies. In: When to screen in Obstetrics & Gynecology, Wildschut, H.I.J., Weiner, C.P., Peters, T.J.(Eds), WB Saunders, London, 108-117.