Summary

Chapter 1 addresses the role of prenatal ultrasound in the detection of congenital anomalies. Accurate detection of a fetal anomaly allows the woman and physician to choose the best way to manage the pregnancy.

In contrast to screening-based ultrasound, indication-based ultrasound is performed because of risk factors known prior to pregnancy, or by risk factors found during the current pregnancy. In this thesis emphasis lies on the indication-based prenatal sonographic detection of (intra)thoracic and (intra)abdominal fetal anomalies.

The objectives of this thesis are (i) to review scientific literature on normal and abnormal fetal (intra)thoracic and (intra)abdominal development, as studied by conventional two-dimensional real-time ultrasound, (ii) to assess the role of conventional two-dimensional real-time and colour coded Doppler ultrasound in congenital cystic adenomatoid malformation of the lung, congenital diaphragmatic hernia, fetal abdominal wall defects, gastrointestinal tract obstructions, and renal tract anomalies, and (iii) to evaluate the presence of fetal (intra)thoracic and (intra)abdominal structural anomalies in small-for-gestational age fetuses.

Chapter 2 describes five sequential phases of normal lung development in the human fetus. Subsequently, literature data are presented on the normal sonographic appearance and anatomy of the fetal lungs using conventional two-dimensional real-time ultrasound with emphasis on fetal lung biometry and lung echogenicity.

With modern colour Doppler ultrasound equipment also the peripheral pulmonary vasculature can be evaluated.

Finally, the normal development and sonographic anatomy of the fetal thoracic wall are discussed.

Chapter 3 presents a review of abnormal lung development and thoracic morphology. Pulmonary hypoplasia can be a life-threatening complication of long-term renal or non-renal oligohydramnios, of intrathoracic space-occupying abnormalities, and of neuromuscular and skeletal disorders. Pulmonary hypoplasia may be detected with two-dimensional and (colour) Doppler ultrasound. With respect to abnormal intrathoracic morphology, fetal hydrothorax, congenital cystic adenomatoid malformation, bronchopulmonary sequestration, bronchogenic cyst, mediastinal teratoma, congenital

diaphragmatic hernia, and fetal thoracic wall abnormalities are discussed.

Apart from congenital heart disease, the role of colour coded Doppler imaging in the prenatal diagnosis of abnormal (intra)thoracic anatomy is limited to pulmonary hypoplasia, bronchopulmonary sequestration, and congenital diaphragmatic hernia.

In a retrospective study, our experience with six cases of macrocystic and one case of microcystic congenital cystic adenomatoid malformation is discussed. Only two fetuses survived. In general, associated polyhydramnios and fetal hydrops suggest a poor outcome.

Furthermore, a retrospective analysis of congenital diaphragmatic hernia (n=28) is presented. Congenital heart disease constituted the majority of associated anomalies. An abnormal karyotype was found in 10.5 per cent. Overall mortality was 86 per cent. Polyhydramnios and intrathoracic position of the stomach were poor predictors of fetal outcome.

Chapter 4 discusses the normal development of the fetal abdominal wall, digestive tract, and renal tract and presents a literature review on their normal sonographic appearance.

The clinical significance of colour Doppler imaging was prospectively studied in patients with an increased risk of an anomaly in their offspring (n=90) and in patients with a suspected fetal anomaly in the present pregnancy (n=76). With colour coded Doppler ultrasound, additional information could be obtained regarding the visualization of the normal intra-abdominal vasculature, in particular the renal artery and vein, common iliac vein, and ductus venosus.

Chapter 5 discusses developmental, sonographic and clinical aspects of anomalies of the fetal abdominal wall, gastrointestinal tract, and renal tract. Also, the additional value of colour coded Doppler in the presence of these anomalies is addressed.

In the first subchapter a retrospective analysis is presented of the two most common types of fetal anterior abdominal wall defects, omphalocele (n=31) and gastroschisis (n=11). An abnormal karyotype was established in 20 per cent in case of an omphalocele, the majority consisting of trisomy 18. No abnormal karyotypes were diagnosed in the gastroschisis group. Approximately the same rate of associated anomalies was encountered in both groups (32 and 36 per cent). No cases of gastroschisis were terminated, while termination of pregnancy was performed in six cases of omphalocele because of the associated anomalies (n=3) or trisomy 18 (n=3). Preterm delivery occurred in 48 per cent in the omphalocele subgroup versus 73 per cent in the gastroschisis

subgroup. The Caesarean section rate was almost identical in the two subgroups (19 and 18 per cent). The survival rate within the omphalocele group was 39 per cent; in all surviving infants this was the sole congenital anomaly and in each instance there was a normal karyotype. In the gastroschisis group 72 per cent survived; two children also displayed unilateral hydronephrosis.

In the second subchapter, 29 cases of suspected obstructive bowel disease are retrospectively studied. Polyhydramnios was present in 69 per cent. An abnormal karyotype was diagnosed in 24 per cent, and was always associated with the ultrasonic "double bubble" sign. A gastrointestinal obstruction was confirmed postnatally in 69 per cent, i.e. oesophageal atresia (n=1), duodenal obstruction (n=12), and small bowel obstruction (n=7). Other congenital malformations were seen in 21 per cent, whereas the overall perinatal mortality was 35 per cent.

In the third subchapter, literature data on renal tract anomalies are presented.

In the fourth subchapter, 33 fetuses with postnatally proven renal pathology are studied with combined colour flow mapping and Doppler evaluation. Renal Doppler flow studies in hydronephrosis do not seem to provide convincing information regarding renal function. Colour flow mapping may be helpful in confirming the diagnosis of renal agenesis.

Chapter 6 focusses on the association between fetal growth retardation and congenital anomalies, with emphasis on fetal thoracic and abdominal pathology. Historically, two types of growth retardation, symmetrical (proportionate) and asymmetrical (disproportionate), have been described. Recent data question this distinction, as well as its clinical relevance, as both categories seem to be at-risk of structural and chromosomal abnormalities. Also, the pathogenetic mechanism underlying the association between congenital malformations and growth retardation remains to be determined. As only few literature data are available on the association between growth-retarded or smallfor-gestational age (SGA) fetuses and structural abnormalities, we retrospectively studied 461 pregnancies which were referred to our centre between 1981 and 1991 because of SGA (n=442), or because of combined SGA and fetal structural abnormality (n=19). In our centre, SGA was confirmed by ultrasound in 75 per cent (344/461), whilst combined SGA and structural anomaly was substantiated in only 16 per cent (3/19). However, a structural anomaly was diagnosed in 34 fetuses who were referred because of SGA alone. Postnatally, SGA (birth weight below the 10th centile) was established in 89 per cent (295/332), while a structural abnormality was confirmed in 65 per cent (24/37) SGA

infants, the majority consisting of renal and cardiac defects. Overall, an abnormal karyotype was established in 7 per cent (20/295).