

Contents

Introduction

1 Standard views for examination of the fetus

2 Central nervous system

- Normal sonographic anatomy
- Neural tube defects
- Hydrocephalus and ventriculomegaly
- Holoprosencephaly
- Agenesis of the corpus callosum
- Dandy-Walker complex
- Microcephaly
- Megalencephaly
- Destructive cerebral lesions
- Arachnoid cysts
- Choroid plexus cysts
- Vein of Galen aneurysm

3 Face

- Normal sonographic anatomy
- Orbital defects
- Facial cleft
- Micrognathia

4 Cardiovascular system

- Introduction to congenital heart disease
- Assessment of the fetal heart
- Atrial septal defects
- Ventricular septal defects
- Atrioventricular septal defects
- Univentricular heart
- Aortic stenosis
- Coarctation and tubular hypoplasia of the aorta
- Interrupted aortic arch
- Hypoplastic left heart syndrome
- Pulmonary stenosis and pulmonary atresia
- Ebstein's anomaly and tricuspid valve dysplasia
- Conotruncal malformations
- Transposition of the great arteries
- Tetralogy of Fallot
- Double-outlet right ventricle
- Truncus arteriosus communis
- Cardiosplenic syndromes
- Echogenic foci
- Cardiac dysrhythmias: premature contractions
- Cardiac dysrhythmias: tachyarrhythmias
- Cardiac dysrhythmias: complete atrioventricular block

- 5 Pulmonary abnormalities
 - Normal sonographic anatomy
 - Cystic adenomatoid malformation
 - Diaphragmatic hernia
 - Pleural effusions
 - Sequestration of the lungs

- 6 Anterior abdominal wall
 - Normal sonographic anatomy
 - Exomphalos
 - Gastroschisis
 - Body stalk anomaly
 - Bladder exstrophy and cloacal exstrophy

- 7 Gastrointestinal tract
 - Normal sonographic anatomy
 - Esophageal atresia
 - Duodenal atresia
 - Intestinal obstruction
 - Hirschsprung's disease
 - Meconium peritonitis
 - Hepatosplenomegaly
 - Hepatic calcifications
 - Abdominal cysts

- 8 Kidneys and urinary tract
 - Normal sonographic anatomy
 - Renal agenesis
 - Infantile polycystic kidney disease (Potter type I)
 - Multicystic dysplastic kidney disease (Potter type II)
 - Potter type III renal dysplasia
 - Obstructive uropathies

- 9 Skeleton
 - Normal sonographic anatomy
 - Skeletal anomalies
 - Osteochondrodysplasias
 - Limb deficiency or congenital amputations
 - Split hand and foot syndrome
 - Clubhands
 - Polydactyly
 - Fetal akinesia deformation sequence (FADS)

- 10 Features of chromosomal defects
 - Phenotypic expression
 - Risk for chromosomal defects

- 11 Fetal tumors
 - Introduction
 - Intracranial tumors
 - Tumors of the face and neck
 - Tumors of the thorax
 - Tumors of the abdomen and retroperitoneum
 - Tumors of the extremities
 - Tumors of the skin
 - Sacroccygeal teratoma
- 12 Hydrops fetalis
- 13 Small for gestational age
- 14 Abnormalities of the amniotic fluid volume
 - Oligohydramnios/Anhydramnios
 - Polyhydramnios
- Appendix I Risk of major trisomies in relation to maternal age and gestation
- Appendix II Antenatal sonographic findings in skeletal dysplasias
- Appendix III Fetal biometry at 14-40 weeks of gestation

ACKNOWLEDGEMENTS

The authors gratefully acknowledge the contributions from Roberto Romero to the chapter on the skeleton, from Rosalinde Snijders to the chapter on the features of chromosomal defects and from Israel Meizner to the chapter on fetal tumors.



Introduction

Ultrasound is the main diagnostic tool in the prenatal detection of congenital abnormalities. It allows examination of the external and internal anatomy of the fetus and the detection of not only major defects but also of subtle markers of chromosomal abnormalities and genetic syndromes. Although some women are at high risk of fetal abnormalities, either because of a family history or due to exposure to teratogens such as infection and various drugs, the vast majority of fetal abnormalities occur in the low-risk group. Consequently, ultrasound examination should be offered routinely to all pregnant women. The scan, which is usually performed at 18–23 weeks of pregnancy, should be carried out to a high standard and should include systematic examination of the fetus for the detection of both major and minor defects.

The Fetal Medicine Foundation, under the auspices of the International Society of Ultrasound in Obstetrics and Gynecology and the International Society of Perinatal Medicine, has introduced a process of training and certification to help establish high standards of scanning on an international basis. The Certificate of Competence in the 18–23-week scan is awarded to those sonographers that can perform the scan to a high standard and can demonstrate a good knowledge of a wide spectrum of fetal abnormalities.

This book, which summarizes the prevalence, etiology, prenatal sonographic features and prognosis for both common and rare fetal abnormalities, provides the basis of learning for the theoretical component of the Certificate of Competence in the 18–23-week scan.