Structural Fetal Abnormalities: The Total Picture. Edited by Roger C. Sanders. Mosby-Year Book, St. Louis, Missouri, 1996, 284 pp., $59.00 paperback

Structural Fetal Abnormalities: The Total Picture is an excellent reference tool for any genetic counselor involved in prenatal diagnosis counseling. Although it doesn’t really provide “the total picture,” it does provide an excellent starting point for the genetic counselor who is faced with counseling a patient whose fetus has just been diagnosed with a structural abnormality. Dr. Roger C. Sanders, Medical Director of the Ultrasound Institute of Baltimore and editor of this book, states in the preface that “this book is designed to provide a comprehensive picture of the genetic, epidemiologic, and ultrasonographic features; the obstetrical, neonatal, and surgical management; and the prognosis of the more common abnormalities discovered by ultrasound examination” (p. vii). To this end, the contributors include: a pediatric geneticist, perinatologist, neonatologist, pediatric cardiologist, pediatric urologist, and several surgeons. The book is divided into chapters based on categories of defects, such as chromosomal, central nervous system, cardiac, infections. Each chapter is further subdivided into specific fetal abnormalities, and each abnormality is reviewed in terms of the epidemiology, genetics, ultrasound findings, and pregnancy and neonatal management. A bibliography of additional resources is included at the end of each chapter. Abnormalities are illustrated with ultrasound photographs and many include clinical photographs of affected fetuses postdelivery. Appendix I consists of “The Differential Diagnoses of Abnormal In Utero Sonographic Findings” and Appendix II consists of “Sonographic Features of Less Common Fetal Abnormalities.” The organization of the book provides readers with quick access to information applicable to those patients whose fetuses have abnormalities described therein. Because the information provided is inclusive of many areas of concern, there may be a temptation to rely solely on the book. In many cases, however, such reliance may do the patient a disservice as there may be more current information available in the literature.
Genetic counselors are frequently asked by their patients, “What exactly is the defect?”, “What is the likely outcome after birth?”, “Can the defect be corrected in utero or after birth?”, “Is my pregnancy now considered high risk?”, “Is pregnancy termination an option?”, “How often does this defect occur?”, “Did I do something to cause the abnormality?”, and “Will it happen again?” Answers to these questions are provided in this book. In discussing each defect, the authors provide a simplified overview of the embryology of the defect and describe the ultrasound findings in the fetus as well as any associated findings in the placenta or amniotic fluid. Where applicable, measurement data are provided. The approximate gestational age at which each defect can be identified is included and is especially helpful for establishing an appropriate prenatal diagnosis plan for subsequent pregnancies. A helpful tool for describing congenital heart defects is the set of schematic diagrams illustrating various structural cardiac defects. Given the complexity of this organ, such diagrams are very useful. Schematic diagrams also would have been helpful to describe abnormalities in other complex organ systems such as the genitourinary tract or the gastrointestinal system, however this was not done in this book.

Within the discussion of each defect there are several sections. In the section called “Sonography,” there are three subcategories: “Pitfalls,” “Differential Diagnosis,” and “Where Else to Look.” Although there is some overlap in the information presented in these subcategories, they ultimately serve the purpose of describing other abnormalities whose sonographic appearances are difficult to distinguish from the defect being described as well as associated anomalies and possible causes of the defect. This section is followed by recommendations for further investigations and consultations and other pregnancy management issues. At this point there appears to be a glaring omission. Whereas Dr. Sanders comments in the preface that “clinical staff such as sonographers, nurses, and genetic counselors play a very significant role in both diagnosis and care” (p. vii), there is not a single recommendation for comprehensive genetic counseling in the entire book. Although evaluation by a geneticist is recommended for parents of fetuses found to have abnormalities often associated with dominantly inherited conditions, such recommendations appear sporadically in the book and do not fully articulate the overall importance of genetic counseling for fetal abnormalities. Supportive psychological care for the family is a frequent recommendation. The appropriateness of maternal/fetal monitoring is discussed as well as pertinent recommendations for delivery. Also included under pregnancy management for each defect is a discussion of pregnancy termination issues. For the most part, this is a nonjudgmental discussion focusing on whether pregnancy termination is likely to be an option given the usual gestational age at diagnosis and whether an intact fetus is re-