

Preface

Congenital anomalies of the kidney and urinary tract (CAKUT) are a major cause of morbidity in children. They occur in 5–10% of the population and represent 25% of sonographically diagnosed fetal malformations. In addition, these anomalies occur in about a quarter of patients with chromosomal aberrations and two-thirds of patients with abnormalities of other organ systems. Some CAKUT are minor; others are major leading to obstruction, urinary tract infection, renal scarring, and chronic kidney disease (CKD). In fact, CAKUT is responsible for most cases of CKD in children.

Knowledge concerning terminology, pathogenesis, and treatment of CAKUT has improved significantly over the past two decades. Also, there have been significant advances in the prenatal diagnosis of these anomalies. Improved technology has contributed to better knowledge of the fetal renal function, renal cortex volume and corticomedullary differentiation, as well as prenatal treatment options. A unified position on prenatal urinary tract dilatation was recently adopted by a consortium of healthcare providers with a consensus on terminology, prenatal follow-up, and post-natal recommendations for imaging and institution of prophylactic antibiotics.

Although the great majority of CAKUT are sporadic and their causes are still unknown, genetic and environmental factors seem to play a major role in their etiology. Based on animal studies, it is believed that genetic mutations may emerge as the main etiologic cause of CAKUT. Mutations in several renal development genes produce defects in the morphogenesis of the kidney and urinary tract causing CAKUT. Molecular analysis of CAKUT-causing genes is now available for clinicians. In spite of continued technical and ethical issues, genetic testing has improved our diagnostic capabilities, allowing the prenatal diagnosis of certain renal diseases in at-risk fetuses, and identifying potential renal disease before it has become manifest. Identification of a specific gene mutation also holds the possibility of correction through gene therapy, although this remains experimental at the present time.

Advances in genetic testing, prenatal diagnosis, fetal surgery, organ transplantation, and surgical treatment of CAKUT have improved the prognosis and quality of life of affected patients. CAKUT have significant impact in clinical medicine and across various specialties, making the book an important reference to pediatricians,

primary care physicians, urologists, pediatric nephrologists, residents, medical students, and healthcare professionals who deal with children. The book is not meant to be a textbook, but rather a concise, easy-to-use clinical reference to help physicians diagnose and manage children with CAKUT and to advise them when to refer patients to the pediatric urologist or nephrologist.

To this end, we have assembled a panel of leading authorities in pediatric urology and nephrology to cover a complete scope of CAKUT and its clinical implications in children. The book stresses clinical presentation of various anomalies, workup, interpretation of imaging studies, genetics, prenatal diagnosis, and treatment. Pathogenesis, etiology, pathology, and surgical management are discussed briefly to help the reader understand the scope of the problem. Other system abnormalities associated with CAKUT are also discussed. Tables, figures, algorithms, and images are provided to assist physicians in the differential diagnosis and workup of different conditions. An extensive appendix listing conditions and syndromes associated with CAKUT is also provided.

We thank our distinguished authors for their authoritative contributions. We are also thankful to Elektra McDermott for her outstanding editorial assistance and to the publishing and editorial staff of Springer for their help and support. We sincerely hope that this book will help our readers to understand, diagnose, and manage CAKUT in children.

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Clinical Implications in Children

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