

PRENATAL DIAGNOSIS and
GENETIC COUNSELING of

OMPHALOCELE

An OVERVIEW and ATLAS of CASES

CHIH-PING CHEN, MD

ELSEVIER TAIWAN LLC

PREFACE

Omphalocele is a heterogeneous condition that may be associated with chromosomal abnormalities, epigenetic defects, and various syndromes and disorders. With the advent of ultrasonography and magnetic resonance imaging, omphalocele can be detected early in pregnancy. Once the prenatal diagnosis of omphalocele has been made, genetic counseling, evaluation of risks of genetic problems and differential diagnosis are required. Despite recent technological advances in pediatric surgery as well as neonatal care, the outcome of omphalocele may be less optimal if it is associated with inherent genetic disorders or various syndromes and disorders. This book discusses some of the more recent advances in knowledge of prenatal diagnosis and genetic counseling of omphalocele-related abnormalities including numerical chromosomal abnormalities, structural chromosomal abnormalities, uniparental disomy, Beckwith-Wiedmann syndrome, omphalocele-exstrophy-imperforate anus-spinal defects complex, pentalogy of Cantrell, and various syndromes and disorders associated with omphalocele.

The atlas contains comprehensive visual information that has been gathered by the author during a career devoted to fetal medicine. I believe that this book will be of benefit to a wide audience of medical students, genetic counselors, medical geneticists, obstetricians, pediatricians and healthcare professionals in the field of prenatal diagnosis. I am extremely grateful to Hsiao-En Cindy Chen, Wayseen Wang and Hsin-Hung Chen for their assistance in manuscript preparation.

Chih-Ping Chen
Mackay Memorial Hospital
Taipei, Taiwan

CONTENTS

| | | |
|------------------|---|------------|
| Chapter 1 | Numerical Chromosomal Abnormalities Associated with Omphalocele | <i>1</i> |
| Chapter 2 | Structural Chromosomal Abnormalities and Uniparental Disomy Associated with Omphalocele | <i>13</i> |
| Chapter 3 | Beckwith-Wiedemann Syndrome..... | <i>27</i> |
| Chapter 4 | Omphalocele-exstrophy-imperforate Anus-spinal Defects (OEIS) Complex | <i>47</i> |
| Chapter 5 | Pentalogy of Cantrell..... | <i>61</i> |
| Chapter 6 | Various Syndromes and Disorders Associated with Omphalocele..... | <i>71</i> |
| Chapter 7 | Atlas and Cases..... | <i>103</i> |
| Index | | |