

# Diagnostic Imaging Of Fetal Anomalies

This is the most comprehensive book to be written on the subject of fetal MRI. It provides a practical hands-on approach to the use of state-of-the-art MRI techniques and the optimization of sequences. Fetal pathological conditions and methods of prenatal MRI diagnosis are discussed by organ system, and the available literature is reviewed. Interpretation of findings and potential artifacts are thoroughly considered with the aid of numerous high-quality illustrations. In addition, the implications of fetal MRI are explored from the medico-legal and ethical points of view. This book will serve as a detailed resource for radiologists, obstetricians, neonatologists, geneticists, and any practitioner wanting to gain an in-depth understanding of fetal MRI technology and applications. In addition, it will provide a reference source for technologists, researchers, students, and those who are implementing a fetal MRI service in their own facility.

This is the only book of its kind in prenatal diagnosis that details the most common sonographically detectable fetal syndromes. It has an easy-to-follow approach of using lists and patterns of malformations to generate a differential diagnosis of the possible syndrome involved. The reader is then led to the more detailed description of each syndrome to determine the exact final diagnosis. The new edition incorporates 3D ultrasound throughout the book, as well as 20 syndromes not previously

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covered. Cross-references sonographic fetal malformations with various syndromes to use patterns of malformation to arrive at the correct diagnosis. Shows the specific features of each syndrome to help you determine how detectable the syndrome is or how likely the fetus is to have the syndrome. Offers up-to-date information on how to manage borderline sonographic findings. Helps you to not only identify what syndrome a given set of malformations is likely to represent, but also understand the clinical implications of that syndrome. Includes more than 500 new images, many of which are 3D ultrasound images. Covers 20 new syndromes, including Perlman Syndrome, Cerebro-Costo-Mandibular Syndrome, Van der Woude Syndrome, Septo-Optic Dysplasia, Saethre-Chotzen Syndrome, Congenital Adrenal Hyperplasia, Congenital High Airway Obstruction Syndrome (CHAOS), Cloacal Extrophy Sequence, Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome, Opitz Syndrome, and DiGeorge Syndrome. Takes full advantage of a new full color design to make reference even easier. Presents ultrasound video clips as they would appear in practice on the bonus DVD.

The most trusted, all-in-one guide to fetal brain imaging—now in full color Edited and written by recognized experts, this acclaimed reference is a highly clinical text and visual atlas. It facilitates a thorough comprehension of the normal and abnormal fetal central nervous system—and helps you apply one of the most important advances in modern perinatology: the early detection of central nervous system anomalies. Here,

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you will find the full spectrum of prenatal sonography tools and insights, from using ultrasound and MRI to diagnose the fetal face, eye, and brain, to the neurobehavioral development of the fetal brain.

Featuring a new full-color presentation and an enhanced, reader-friendly design, the third edition of this unmatched guide is completely refreshed to mirror the significant advances made in imaging resolution and three-dimensional Doppler technology. In addition, the book reflects the growing interest in imaging the fetal nervous system as it pertains to the fetal brain. FEATURES New full-color design and additional figures, tables, and graphs New chapter on ventriculomegaly examines the most common presenting sonographic sign of brain pathology New chapters on the evaluation of the fetal cortex and posterior fossa shed light on diagnostically problematic areas of the fetal brain New chapters highlighting intrauterine insults, intrauterine infections, and metabolic disorders demonstrate the progress being made in areas that have become critical to fetal neuroscans Greater emphasis on the use of high frequency and deep penetrating ultrasound transducer probes clearly explain how they can yield high-resolution pictures of the fetal brain and spine Latest perspectives on dissemination of 3D ultrasound techniques and magnetic resource imaging are interwoven into individual chapters to encourage their adoption in daily clinical practice More detailed examination of imaging the fetal brain is based on leading-edge, peer-reviewed research from around the world SI units are included throughout Numerous new 2D and 3D ultrasound images and

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updated literature references contribute to the most current overview available of this dynamic specialty. This comprehensive guide to MRI of the pregnant patient presenting with fetal or maternal diseases provides a practical, hands-on approach to the use of state-of-the-art MRI techniques and the optimization of sequences. It analyzes data obtained from maternal and fetal MRI examinations, reviews fast imaging techniques, details pitfalls related to fetal MRI and assesses methods for improving image resolution. Fetal pathological conditions and methods of prenatal MRI diagnosis are discussed according to organ system and the literature is reviewed. Interpretation of findings and potential artifacts are thoroughly considered with the aid of numerous high-quality illustrations. The book will be a detailed resource for radiologists, obstetricians, neonatologists and geneticists, as well as any other practitioner who wishes to gain an in-depth understanding of fetal and maternal MRI. In addition, it will serve as a reference source for technicians, researchers and students, as well as for any specialist who is planning to set up a fetal and maternal MRI service.

The most authoritative guide to sonography in obstetrics and gynecology—completely revised to reflect the latest technologies and practices. Written by radiologists and ob/gyns to provide a balanced perspective, this standard-setting guide is both a clinically relevant reference text and atlas. Presented in full color and enhanced by more than 2000 illustrations, it expertly examines the full spectrum of disorders and conditions likely to be encountered in gynecologic and maternal-fetal care.

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Readers will find coverage of the latest procedures and diagnostic guidelines for the use of sonography in ob/gyn, including 3D and 4D image processing, transvaginal sonography, and color Doppler sonography. The book opens with general obstetric sonography, covering such pivotal topics as normal pelvic anatomy and fetal echocardiography, before moving into fetal anomalies and disorders. Risk assessment and therapy, including first trimester screening and amniocentesis, are explored in the next section, while the remaining parts of the book focus on maternal disorders, gynecological sonography, and the newest complementary imaging modalities.

- All-inclusive coverage of everything from sonographic operating instruments and screening the fetal patient for syndromes and anomalies, to diagnosing the female patient for cysts, infertility, and incontinence
- Learning aids include: a “key points” section in each chapter; skill-building clinical scenarios; a stronger focus on differential diagnosis; multiple visuals (figure, illustration, or table) on each page; and helpful chapter-opening summaries
- New chapters on enhanced myometrial vascularity, Doppler sonography of abnormal fetal condition, sonography of uterine scar pregnancies, ultrasound-guided fetal therapy, evaluation of acute and chronic pelvic pain, and contrast-enhanced

A mainstay for radiology trainees and practitioners, *Diagnostic Imaging: Genitourinary, Third Edition* features an image-rich, reader-friendly format that outlines the role of imaging in diagnosing and managing diseases of the GU tract. Concise chapters and spectacular imaging examples combine to make this medical reference book

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an all-inclusive resource for every member of the radiology team. State-of-the-art imaging — such as CT urography, DECT, MR urography, and DWI MR — addresses the rapidly changing diagnostic algorithm used for evaluation of diseases of the genitourinary tract Presents approximately 2,500 superior images for a greater visual understanding, while bulleted text expedites reference and review Includes an expanded table of contents, updated chapters and references, and brand new illustrations that highlight the roles of MR and ultrasound for evaluating diseases of the GU tract Covers important hot topics such as prostate carcinoma staging and surveillance, adrenal adenoma work-up and relevance, staging and subclassification of renal cell carcinoma, and the role of DECT for renal stone characterization.

Richly illustrated and comprehensive in scope, *Obstetric Imaging, 2nd Edition*, provides up-to-date, authoritative guidelines for more than 200 obstetric conditions and procedures, keeping you at the forefront of this fast-changing field. This highly regarded reference covers the extensive and ongoing advances in maternal and fetal imaging in a concise, newly streamlined format for quicker access to common and uncommon findings. Detailed, expert guidance, accompanied by superb, high-quality images, helps you make the most of new technologies and advances in obstetric imaging. Features more than 1,350 high-quality images, including 400 in color. Helps you select the best imaging approaches and effectively interpret your findings with a highly templated, bulleted, at-a-glance organization.

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Reflects all the latest developments in the field, including genetics, open fetal surgery, fetal echocardiography, Zika virus, and 3D imaging, so you can provide the safest and most responsive care to both mother and fetus. Includes new chapters on Limbs and Bones Overview; Open Fetal Surgery; Biophysical Profile; Ultrasound Physics; Elastography; Doppler; MRI; Echogenic Bowel; Pregnancy of Unknown Location (PUL), Failed Pregnancy and Ectopic Pregnancy, Cesarean Scar Pregnancy; Cytomegalovirus (CMG), Rubella, Toxoplasmosis, Herpes, Varicella; and Congenital Syphilis; plus a new chapter on Zika Virus written by imaging experts from the "hot zone." Keeps you up to date with the latest developments in multimodality imaging and optimizing diagnostic accuracy from ultrasound, 3D ultrasound, Doppler, MRI, elastography, image-guided interventions, and much more.

Stay up to date with recent advances in the use of ultrasound in early gestation with this comprehensive, full-color reference. *First Trimester Ultrasound Diagnosis of Fetal Abnormalities* is an authoritative, systematic guide to the role of first trimester ultrasound in pregnancy risk assessment and the early detection of fetal malformations. High-quality illustrations and numerous tables throughout enhance readability, making this text an excellent daily resource in clinical practice.

In recent years, Fetal MR has grown continually in importance, and the brain has become the main focus of investigation. However, we lack established standards and a good knowledge of the normal MR appearance. To fill this

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gap is the purpose of the first part of this book, which is an MR atlas of the cerebral development of the fetus. The second part is dedicated to cerebral pathologies. It includes, for each condition, a summary of the fundamental data, the imaging findings (US and MR) in correlation with neurofetopathology and/or postnatal imaging, and a brief perspective of the prognosis.

The newest edition of *Diagnostic Imaging: Obstetrics* provides radiologists with world-class content and instructions on the latest methodologies in this rapidly changing field. Featuring approximately 260 diagnoses highlighting the most recent information, references, and images, this title serves as a practical, highly formatted guide that's well suited for today's busy radiologists. Enhanced chapters on embryology, new reference tables, updated patient management guidelines, and much more ensure readers are current with the knowledge required for competent clinical practice. Guides practitioners through the intricacies of obstetric and pregnancy-related anomalies Features expanded embryology chapters delineating normal developmental anatomy An increased number of reference tables enables you to look up a normal measurement Includes new practice guidelines for patient management, a summary of consensus panels, and new standardized nomenclature Expanded syndrome section is rich in clinical pictures Brand new differential diagnoses section allows you to look up a finding and be guided to the correct diagnosis (e.g., absent cavum septi pellucidi) Richly colored graphics and fully annotated images highlight the most important diagnostic possibilities Highly templated and bulleted format makes it easier than ever to locate key information

About 21 years ago prenatal diagnosis became part of the physician's diagnostic armamentarium against genetic defects. My first monograph in 1973 (*The Prenatal Diagnosis*



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of Hereditary Disorders) critically assessed early progress and enunciated basic principles in the systematic approach to prenatal genetic diagnosis. Six years later and under the current title, a subsequent volume provided the first major reference source on this subject. The present second (effectively third) edition, which was urged in view of the excellent reception of the two earlier volumes, reflects the remarkable growth of this new discipline and points to significant and exciting future developments. Notwithstanding these advances, the use of the new tools and techniques for the benefit of at-risk parents has taken many more years than most anticipated. Key factors have been the lack of teaching of human genetics in medical schools in the preceding decades and the difficulty of educating practicing physicians in a new scientific discipline. Even today the teaching of genetics in medical schools leaves much to be desired and this will further delay the introduction of newer genetic advances to the bedside.

The quick-reference guide for the diagnosis and treatment of fetal anomalies – completely updated with a new full-color design *Structural Fetal Abnormalities* delivers practical, authoritative coverage of the detection and treatment of 125 of the most common fetal anomalies, grouped by organ system, and presented in concise bulleted text. This edition is highlighted by a new full-color design, hundreds of new high-quality images, and updated content that reflects the most current treatment methods. The book has been hailed for its consistent templated chapter structure (covering important topics such as Epidemiology/Genetics, Sonography, Pregnancy Management, Neonatal Management, Interventions and Surgical Options), hundreds of sonographic and gross clinical images show all stages of fetus development, and sonography findings that include pitfalls and differential diagnosis. The text has been completely

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revised under new lead editor W. Allen Hogge.

The most frequently asked questions that confront the fetal medicine trainee/expert on a daily basis are “Is the finding real or merely an artifact?” and “Is the diagnosis correct?”. However, to be able to find the description of an abnormal ultrasound finding in a textbook, one generally has to search by the definite diagnosis, which has not been done as yet. This uneasy feeling was the first factor that directed the layout of *Ultrasound of Congenital Fetal Anomalies: Differential Diagnosis and Prognostic Indicators, Second Edition*. Copiously illustrated, the book displays fetal anomalies by scanning view and descriptions of all major ultrasound planes, detailing what can be considered a normal view and what cannot. See What’s New in the Second Edition: Early detection of fetal anomalies (12-14 weeks) Ultrasound in fetal infections and in twins The nuchal translucency issue, the newest intracranial translucency as well as the range of congenital anomalies detectable at this gestational age Expanded coverage of heart anomalies, including arrhythmias and early fetal echocardiography The author’s mission continues to be to provide guidance on how to quickly recognize and diagnose congenital fetal anomalies, beginning at the beginning with ultrasound sign all the way through to final diagnosis.

This work covers the top imaging diagnoses in obstetrics, including both common and uncommon entities and includes an extensive image gallery for each entity, depicting common and variant cases with bulleted summaries of terminology. This atlas presents the sonographic features of the normal fetal abdomen at different gestational ages, and describes the anomalies of different organ systems in the fetal abdomen. It covers the gastrointestinal tract, genitourinary tract, and abdominal wall defects with the help of numerous ultrasound images, and also addresses differential diagnosis using

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various sonographic images of the fetal abdomen, as well as effective diagnostic approaches for these conditions. Based on the RCOG Training Module in Fetal Medicine, this book provides a knowledge base for practitioners in obstetrics and maternal-fetal medicine.

This text/atlas provides a multidisciplinary approach to fetal cardiac ultrasound. It covers all aspects of the subject, including how to perform a fetal echocardiogram--the scanning views involved, the correct equipment to use, and the technical and anatomical limitations; normal anatomy--views, variants, and common pitfalls in scanning and diagnosis; abnormalities--what they look like, which scanning views and modalities to use, differential diagnosis, and prognosis and treatment; and modalities--how and when to use the different types of ultrasound. It also serves as an all-inclusive reference of the drugs, chromosomal abnormalities, and syndromes associated with congenital heart disease. Presents the skills and perspectives of experts from a variety of disciplines that are necessary to diagnose and manage a wide array of complex cardiac problems presented by the fetus. Provides vital information regarding surgical techniques available for specific cardiac abnormalities and their success rates--useful when counselling parents on what to expect before and after birth. Reviews structural and rhythm abnormalities in the fetal heart, what they look like on ultrasound, and which scanning views are necessary for accurate diagnosis. Describes common pitfalls encountered in fetal echocardiography, including normal variants which are often mistaken for abnormalities. Contains the most extensive list in publication of drugs, syndromes, and chromosome abnormalities associated with congenital heart disease. Takes a step-by-step approach to the different scanning views involved, the correct equipment to use, the technical and anatomic limitations of fetal

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echocardiography, and much more. Explains how and when to use different aspects of ultrasound, including 2-D imaging, M-Mode, Pulsed Doppler, Color Flow Imaging, and Color Power. Features more than 320 clearly reproduced ultrasound images--18 in full color--of normal and abnormal cardiac anatomy. Makes specific guidance easy to find with separate chapters and tables for each kind of abnormality. This simple and easy-to-use guide to fetal echocardiography will help physicians and sonographers obtain a complete evaluation of the normal and abnormal fetal heart. The book is written in a user-friendly style and thoroughly illustrated with ultrasound images accompanied by schematic drawings. This edition presents a comprehensive approach to the examination of the fetal heart and covers all major cardiac malformations. Chapters include color Doppler in fetal echocardiography, three-dimensional ultrasound in fetal echocardiography, first and early second trimester imaging of the fetal heart, and an updated genetics section. This book, written by internationally recognized experts in fetal echocardiography, is a must-have for physicians and sonographers interested in this field.

This teaching atlas provides a comprehensive and practical overview of fetal ultrasound technology, providing up-to-date diagnosis and examination guidelines for all clinically important anomalies and diseases. Incorporating an impressive collection of sonographic images and plates, the book provides an invaluable visual aid in recognizing even the most difficult-to-interpret ultrasound findings.

This book describes how different diagnostic techniques may facilitate the prenatal identification of orofacial malformations. The role of standard two-dimensional ultrasound is documented and explained, but detailed attention is also paid to the use of three-dimensional (3D) ultrasound, including the novel 3D reslicing technique, and to the value of

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complementary magnetic resonance imaging. The malformations considered include cleft lip, cleft lip and palate, ocular malformations, abnormalities associated with fetal aneuploidy, and tumors. These conditions are depicted in numerous high-quality images accompanied by thorough and informative descriptions of imaging findings. The excellence of the illustrative material and the caliber of the authors, who are leading international experts in the field, ensure that Prenatal Diagnosis of Orofacial Malformations will be an invaluable tool for obstetricians and gynecologists.

Presented by an international team of experts, the new edition of Diagnostic Imaging: Gynecology features an exhaustive collection of imaging findings in gynecologic diseases. It pairs state-of-the-art images with extensive correlative imaging between ultrasound, sonohysterography, hysterosalpingography, MR, PET/CT, and gross specimens, while a dedicated Techniques section is designed to help optimize imaging protocols and enhance diagnostic specificity. Completely revised content, updated reference lists, and new high-quality images keep you abreast of the latest knowledge in the field. Features a brand-new section on pelvic floor imaging, accompanied by exquisite illustrations. Pelvic Floor section includes detailed anatomic drawings and patient examples. Diagnoses are grouped according to the organ involved: Uterus, cervix, vagina and vulva, ovary, fallopian tubes, and multi-organ disorders. Introduction and Overview chapters contain extensive illustrations of normal anatomy and variants, followed by a comprehensive series of chapters covering all pertinent pathologic entities, including congenital anomalies, infectious/inflammatory diseases, and benign and malignant neoplasms. Includes an expanded section on tumor staging with quick-reference tables, illustrations, and case examples covering TNM classifications, FIGO staging, and AJCC

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prognostic groups. Features illustrated differential diagnoses based on imaging findings. Essential information is distilled into a succinct, bulleted format with numerous high-quality images and "Key Facts" boxes to facilitate learning. Comes with Amirsys eBook AdvantageT, an eBook featuring expanded content, additional images, and fully searchable text.

Obstetric Imaging will help you detect fetal abnormalities with greater confidence and accuracy. Covering MRI as well as ultrasound and interventional procedures, it equips you with expert tips for recognizing and addressing problems that you might otherwise miss. Obstetric Imaging provides the advanced guidance you need to recognize fetal health challenges early and respond effectively! Get advanced clinical guidance from a preeminent team of international maternal-fetal medicine specialists and obstetrician/gynecologists. See perfect examples of normal and variant anatomy, as well as the full range of fetal syndromes, with 1,318 images, 361 in full color. Know how to get optimal diagnostic accuracy from ultrasound and when to use MRI instead. Effectively perform image-guided interventions including amniocentesis, fetal transfusion, selective laser photocoagulation, radiofrequency ablation, fetal shunt placement, and more. Master important nuances of sonography by watching 69 videos online. Access Obstetric Imaging online at [www.expertconsult.com](http://www.expertconsult.com), view all the videos, and download all the images.

Updated to reflect the recent advances in this fast-changing field, this highly illustrated text examines the latest imaging modalities for prenatal diagnosis of fetal abnormalities. A team of leading authorities provides practical, step-by-step guidance on everything from detection and interpretation...to successful management approaches. Algorithms and management strategies throughout not only describe the

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features of abnormalities, but also show you how to arrive at a correct diagnosis through the use of color Doppler, 3-D ultrasound, and fetal MR. This 2nd Edition offers the essential practice-proven guidance you need to arrive at confident diagnoses in critical situations. It's a resource you'll turn to time and again! Includes more than 700 illustrations that clearly depict a full range of conditions. Focuses on image interpretation and, wherever possible, correlation of radiographic features with pathologic findings to provide the most accurate and reliable diagnosis possible. Uses a reader-friendly format to facilitate quick access to specific information. Includes new chapters reflecting major advances in magnetic resonance imaging and 3-D ultrasound. Presents an increased use of lists of differential diagnosis.

Diagnostic Imaging of Fetal Anomalies Lippincott Williams & Wilkins

For more than 70 years, Caffey's Pediatric Diagnostic Imaging has been the comprehensive, go-to reference that radiologists have relied upon for dependable coverage of all aspects of pediatric imaging. In the 13th Edition, Dr. Brian Coley leads a team of experts to bring you up to date with today's practice standards in radiation effects and safety and head and neck, neurologic, thoracic, cardiac, gastrointestinal, genitourinary, and musculoskeletal pediatric imaging. This bestselling reference is a must-have resource for pediatric radiologists, general radiologists, pediatric subspecialists, pediatricians, hospitals, and more – anywhere clinicians need to ensure safe, effective, and up-to-date imaging of children. Includes separate chapters on radiation effects and safety, pre-natal imaging, neoplasms, trauma, techniques, embryology, genetic anomalies, and common acquired conditions. Takes an updated, contemporary approach with more focused and consistently formatted content throughout. Clinical content includes Overview; Etiologies,

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Pathophysiology, and Clinical Presentation; Imaging, including pros and cons, costs, evidence-based data, findings, and differential diagnostic considerations; and Treatment, including follow-up. Features 8,500 high-quality images – 1,000 new or updated. Provides expanded coverage of advanced imaging and diagnostics, including genetics and fetal imaging, MRI and advanced MR techniques, low-dose CT, ultrasound, nuclear medicine, and molecular imaging, as well as the latest quality standards, evidence-based data, and practice guidelines. Features new Key Points boxes and more tables and flowcharts that make reference faster and easier. Focuses on safety, particularly in radiation dosing, as part of the Image Gently® campaign to improve pediatric imaging while limiting radiation exposure and unneeded studies.

Designed primarily for the experienced sonographer who is confronted with obstetric patients having developmentally abnormal fetuses, the chapters are written by authors chosen because of their experience and expertise. Students and sonographers-in-training should also find this valuable. Access practical guidance on the radiologic detection, interpretation, and diagnosis of fetal anomalies with Twining's Textbook of Fetal Abnormalities. With fetal scanning being increasingly done by obstetricians, this updated medical reference book features a brand-new editorial team of radiologist Anne Marie Coady and fetal medicine specialist Sarah Bower; these authorities, together with contributions from many other experts, provide practical, step-by-step guidance on everything from detection and interpretation to successful management approaches. Twining's Textbook of Fetal Abnormalities is a resource you'll turn to time and again! Consult this title on your favorite e-reader, conduct rapid searches, and adjust font sizes for optimal readability. Quickly access specific information with a



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user-friendly format. Deliver a rapid, reliable diagnosis thanks to a strong focus on image interpretation, as well as the correlation of radiographic features with pathologic findings wherever possible. Clearly visualize a full range of conditions with help from more than 700 images. Stay abreast of the latest developments in detecting fetal abnormalities with 4 brand-new chapters: Fetal Growth; Haematological Disorders; Fetal Pathology; and Fetal Tumours. Access increased coverage of fetal growth, first trimester anomalies, DDX, and clinical management. Understand the major advances in today's hottest imaging technologies, including 3-D Ultrasound, Fetal MRI, and Colour Doppler. Effectively interpret the images you encounter with highly organized coordination between figures, tables, and imaging specimens. This unique book facilitates recognition and identification of the fetal abnormalities seen in ultrasound. It is the only resource that provides so much detail about all of the fetal abnormalities currently known. Enables readers to approach a given diagnostic challenge either by looking up its sonographic presentation, or by turning directly to the name of the syndrome that they believe is responsible. An easy-to-use organization makes it simple for readers to locate the most reliable information available today. Enables readers to approach a given diagnostic challenge either by looking up its sonographic presentation, or by turning directly to the name of the syndrome that they believe is responsible. Lists the syndromes associated with any sonographic finding and provides guidance on its differential diagnosis. Cross-references to discussions of individual syndromes make it easy to locate further information on each condition. Explores the full range of syndromes that are known to cause malformations in the fetus. Sonographic images are accompanied by concise discussions of prenatal diagnosis, differential diagnosis, detectability at various gestational

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ages, and common features. Cross-references point the way to other syndromes with similar sonographic characteristics. Examines sonographic findings that are potentially associated with chromosomal abnormalities, and discusses the issues that surround the management of these fetuses. Investigates the specific malformations and problems associated with monozygotic twins. Features over 582 crisp illustrations that depict the sonographic appearance of a full range of fetal abnormalities.

Birth defects are one of the major public health concerns in the world, as they cause approximately 20% of infant deaths. Genetic disorders, including chromosome abnormalities and single gene disorders, are the most common causes of birth defects for which there is no efficient treatment. Prenatal genetic screening and diagnosis allow early identification of affected conceptuses and facilitates reproduction planning or counseling. Molecular technologies have developed rapidly in recent years and have been widely used in screening and diagnosis of genetic disorders at all stages of prenatal development (e.g. pre-implantation, embryonic and fetal). However, their performance still needs to be validated and assessed as the balance between their advantages and disadvantages need to be discussed. With the ability to detect copy number variations (CNVs), polyploidy, uniparental disomy and maternal cell contamination, SNP-based chromosomal microarray analysis (CMA) is showing the unique importance in diagnosing chromosomal abnormalities. The interpretation of CNVs remains a challenge; however, ultrasound and biochemical screening improve the diagnosis of fetal chromosomal abnormalities. Whole exome sequencing (WES) and whole genome sequencing (WGS) play increasingly significant roles in prenatal and carrier screening for genetic disorders. NGS-based non-invasive prenatal screening (NIPS) is now widely used for detecting

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common autosomal aneuploidies and has shown the potential of detecting microdeletions and microduplications. However, further investigations of the sensitivity and accuracy are required and large-scale data is necessary to evaluate the performance and clinical applications of current and new methods. Recently, reports of application of newer technologies in prenatal setting became available. Examples include third generation sequencing (reading the nucleotide sequences at the single molecule level), digital PCR (used for direct quantification of DNA) and cell-based NIPT. In the followed listed papers, the authors showed their successful experiences in identifying novel mutation, detecting low-level mosaicism or de novo mutations limited in germline cells, investigating the association of the CNVs with specific phenotypic alterations by using WES, CMA, digital PCR and some other new-developed molecular techniques. More interesting, the authors also presented a report about the evaluation of diagnostic yield in fetal WES, which suggested a new tendency to apply WES or WGS directly for prenatal diagnosis. We believed that the efficiency of scanning causative mutations and prenatal or preimplantation genetic diagnosis for genetic disorders will further improved based on the technologies of whole genomic sequencing with further improved output and resolution. New techniques, such as quick-WES for the newborn in intensive care unit, direct-WGS for prenatal diagnosis and non-invasive test for fetal monogenic disorders, will become available in the near future. Extensively illustrated, this textbook covers the diagnostic imaging of fetal abnormalities. Taking a practical approach, the book guides reader step-by-step through the problems involved and explains the correct diagnosis.

A didactic, illustrated guide to the use of ultrasound as a diagnostic tool in clinical practice. Prepared by an international group of experts with wide experience in both

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developed and developing countries, the manual responds to the need for a basic reference text that can help doctors, sonographers, nurses, and midwives solve imaging problems when no experts are available. With this need in mind, the manual adopts a practical approach aimed at providing a thorough grounding in both the techniques of ultrasound and the interpretation of images. The need for extensive supervised training is repeatedly emphasized. Because the clinical value of ultrasound depends so greatly on the experience and skill of the operator, the manual makes a special effort to alert readers to common pitfalls and errors, and to indicate specific clinical situations where ultrasound may not be helpful or reliable as a diagnostic tool.

Explanatory text is supported by numerous practical tips, warnings, checklists and over 600 illustrations. The opening chapters explain how ultrasound works, outline the factors to consider when choosing a scanner, and introduce the basic rules of scanning, including advice on how to recognize and interpret artefacts. Guidance on the selection of ultrasound equipment includes clear advice concerning where costs can be spared and where investment is essential. The core of the manual consists of seventeen chapters providing guidance on scanning techniques and the interpretation of images for specific organs and anatomical sites, with the most extensive chapter devoted to obstetrics. Each chapter contains illustrated information on indications for scanning, preparation of the patient, including choice of transducer and setting of the correct gain, general scanning techniques, and specific techniques for identifying anatomical landmarks and recognizing abnormalities. The manual concludes with WHO specifications for a general-purpose scanner judged entirely suitable for 90-95% of the most common ultrasound examinations.

Nowadays, nobody can imagine practicing obstetrics without

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using obstetrical ultrasound. Working in the prenatal diagnosis field requires dedication, patience, skills, experience, caution, and empathy. The concept of this book was guided by the desire to provide some help to the ultrasound operators. On a daily basis, they are confronted with the challenging task of ruling out or suspecting/confirming the diagnosis of fetal anomalies, either structural or chromosomal. The chapters of this book contain objective and exhaustive updated reviews of the pertinent literature, so that the reader would have a wide reference basis on each subject. Yet, many authors scan the fetus themselves or are directly involved with managing pregnancies with structural malformations or chromosomal anomalies. They kindly shared their personal experience and lessons learned over the years. This book is beneficial for all the professionals working in the prenatal diagnosis.

Effectively evaluate obstetric patients with Fundamental and Advanced Fetal Imaging: Ultrasound and MRI! Written by an impressive roster of leading fetal radiologists and maternal-fetal medicine specialists, with additional input from cardiologists, geneticists, and Doppler specialists, this state-of-the-art reference explores how to obtain the maximum information from fetal ultrasound and magnetic resonance imaging, so you can rule out pathologies with confidence – or identify them early enough to initiate the most appropriate interventions.

**FETAL ANOMALIES** Advances in ultrasound technology are reshaping the field of health care for obstetricians and pediatric specialists. Detailed fetal imaging has enabled medical professionals to detect fetal structural anomalies and research practical guidelines for prenatal diagnosis and postnatal management. *Fetal Anomalies: Ultrasound Diagnosis and Postnatal Management* is a practical sourcebook with images of structural fetal malformations on a

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continuum that begins at the stage of ultrasonographic identification, progressing to characterization in the newborn period, and culminating in repair and postoperative follow up. This comprehensive text correlates pre- and post-natal images with the type of treatment appropriate to structural anomalies of the different organ systems. Numerous examples from each organ system are included and the material is clinically oriented.

**Fetal Anomalies: Ultrasound Diagnosis and Postnatal Management** reviews such topics as: Externally visible defects Skeletal dysplasia Central nervous, gastrointestinal, urinary, and genital systems Umbilical cord anomalies Abnormalities specific to multiple pregnancies Abnormalities of amniotic fluid volume Abnormalities that elude prenatal detection

Incorporating the pictorial strengths of an atlas with the didactic utility of a reference work, **Fetal Anomalies: Ultrasound Diagnosis and Postnatal Management** is a unique book bridging various specialties that comprise maternal-fetal medicine, such as obstetrics, diagnostic imaging, neonatology, perinatology, surgery, and urology. Here is a complete and convenient guide to the normal sonographic appearances of the embryo and fetus and its uterine environment. This handy atlas will provide you with a thorough knowledge of normal fetal anatomy and better enable you to promptly recognize and diagnose abnormalities. The images in this atlas were produced with state-of-the-art high-resolution ultrasound imaging systems and depict a spectrum of normal anatomy encountered during pregnancy. Coverage includes the fetal environment - the cervix, uterus, placenta, and umbilical cord, the successive stages of embryonic development; and the normal appearances of fetal organ systems. The appendix provides a set of basic biometry tables for easy reference and daily use. This pocket atlas is an essential resource for all health care

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professionals who perform or interpret obstetric ultrasound studies, including radiologists, obstetricians and gynecologists, sonographers, geneticists, nurses, and genetic counselors.

Written by the world's preeminent authorities on diagnostic ultrasound, the Second Edition of this bestseller guides readers through the use of ultrasound to detect and identify birth defects--including heart malformations, kidney obstructions, intestinal blockages, lung abnormalities, and more. The book offers up-to-date advice on what to look for, given a certain risk or clinical history, and how to perform and interpret the ultrasound examination. More than 1,600 images--including full-color throughout--provide a true-to-life view of ultrasound findings. Each anomaly is discussed in an easy-to-follow format that covers characteristic features...pathogenesis and etiology...differential diagnosis...prognosis...and management. This edition includes brief tables of teratogens and information on genetic markers.

Covers a broad spectrum of respiratory diseases during pregnancy, in order to improve successful management of both mother and fetus.

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