The Atlas of X-Linked Intellectual Disability Syndromes is a comprehensive summary of the clinically distinctive disorders caused by genes on the X chromosome. Each syndrome is defined and information is provided on somatic features, growth and development, neurological signs, cognitive performance, imaging and other laboratory findings, and when possible, the nature and localization of the responsible gene. Craniofacial and other somatic findings are extensively illustrated. A differential matrix accompanies each syndrome description to assist the reader in identifying other X-linked syndromes with overlapping features. Individual syndrome entries are supplemented with nineteen appendices that identify syndromes with common features and provide the location or mapping limits and function of the responsible genes.

The Bedside Dysmorphologist is a thoughtful clinical guide to common--and often quite subtle--congenital malformations in clinical exam. Organized by area of the body, each section provides the user with a concise, illustrated roadmap for assessment and differential diagnosis of highly nuanced elements of dysmorphology. Narrated by a world-class clinical geneticist and enriched with a library of original photos, it provides a safety net for physicians encountering abnormalities in physical exam.

Bone Dysplasias is an atlas of genetic disorders of skeletal development. Each disorder is defined and information is provided on somatic features, growth and development, neurological signs, cognitive performance, imaging and other laboratory findings, and when possible, the nature and localization of the responsible gene. Craniofacial and other somatic findings are extensively illustrated. A differential matrix accompanies each disorder description to assist the reader in identifying other bone dysplasias with overlapping features. Individual disorder entries are supplemented with twenty appendices that identify disorders with common features and provide the location or mapping limits and function of the responsible genes.
This is a unique atlas presenting age-related radiographs on more than 250 rare constitutional skeletal diseases (dysplasias, dysostoses, osteolyses, disorders of bone density, and more) focusing on diagnostically essential radiographic and clinical features. Each chapter is supplemented with prognostic and therapeutic information, a guide to differential diagnoses, and a short list of the most relevant publications. A major advantage is the systematic conformation of chapters, sparing the reader a cumbersome read-through of longer text. Presentation in accordance with the most recent International Nosology and Classification of Genetic Skeletal Disorders.

Ciliopathies A reference for clinicians
Thomas D. Kenny and Philip L. Beales (eds)

The ciliopathies are a group of rare diseases that often affect multiple systems within the body, and are caused by defects in the function or structure of cilia. This resource provides a clinical overview and reference to this newly emergent group of disorders ranging from Alström syndrome to putative ciliopathic disorders. Each chapter provides an in-depth discussion on a specific disorder, including the latest scientific research together with a description of its features, and practical guidelines on diagnosis.

Common Malformations
Lewis B. Holmes

This extensively illustrated resource describes the most common malformations and draws from many sources the information needed for a full diagnostic evaluation and discussion of treatment options and genetic counseling. The text also covers minor anomalies, birthmarks and includes dozens of charts of anthropologic measurements, material that is needed in the initial physical examination to describe an infant's physical features.

The Drama of DNA: Narrative Genomics
Karen H. Rothenberg and Lynn Wein Bush

Through the use of dramatic narratives, The Drama of DNA brings to life the complexities raised by the application of genomic technologies to health care and diagnosis. This creative, pedagogical approach shines a unique light on the ethical, psychosocial, and policy challenges that emerge as comprehensive sequencing of the human genome transitions from
research to clinical medicine. Narrative genomics aims to enhance understanding of how we evaluate, process, and share genomic information, and to cultivate a deeper appreciation for difficult decisions encountered by health care professionals, bioethicists, families, and society as this technology reaches the bedside. This innovative title includes both original genomic plays and theatrical excerpts that illuminate the implications of genomic information and emerging technologies for physicians, scientists, counselors, patients, blood relatives, and society. In addition to the plays, the authors provide an analytical foundation to frame the many challenges that often arise.

**Duchenne Muscular Dystrophy**

Alan E. H. Emery, Francesco Muntoni, and Rosaline C. M. Quinlivan

Duchenne Muscular Dystrophy, an inherited and progressive muscle wasting disease, is one of the most common single gene disorders found in the developed world. In this fourth edition of the classic monograph on the topic, Alan Emery and Francesco Muntoni are joined by Rosaline Quinlivan, Consultant in Neuromuscular Disorders, to provide a thorough update on all aspects of the disorder. Recent understanding of the nature of the genetic defect responsible for Duchenne Muscular Dystrophy and isolation of the protein dystrophin has led to the development of new theories for the disease's pathogenesis. This new edition incorporates these advances from the field of molecular biology, and describes the resultant opportunities for screening, prenatal diagnosis, genetic counselling and from recent pioneering work with anti-sense oligonucleotides, the possibility of effective RNA therapy. Although there is still no cure for the disorder, there have been significant developments concerning the gene basis, publication of standards of care guidelines, and improvements in management leading to significantly longer survival, particularly with cardio-pulmonary care. The authors also investigate other forms of pharmacological, cellular and gene therapies.

**Epstein's Inborn Errors of Development**

The Molecular Basis of Clinical Disorders of Morphogenesis

Robert P. Erickson and Anthony J. Wynshaw-Boris (eds)

Chapters provides overview of pathways of development and reviews of dysmorphic syndromes for which the causative gene has been identified. For each disorder, an analysis of the role of the gene in the relevant developmental pathway is provided, along with the mechanism by which mutations in the gene cause the developmental pathology. Emphasis is placed the developmental roles of genes in the causation of hereditary conditions affecting appearance and function.
Ethical Dilemmas in Genetics and Genetic Counseling
Principles through Case Scenarios
Janice Berliner (ed.)

Knowledge of the genetic basis of human diseases is growing rapidly, with important implications for pre-conception, prenatal, and predictive testing. While new genetic testing offers better insight into the causes of and susceptibility for heritable diseases, not all inherited diseases that can be predicted on the basis of genetic information can be treated or cured. By using a creative approach that focuses on a single extended family as a case example to illustrate each chapter’s key point, the authors elucidate ethical issues arising in the genetics clinic and laboratory surrounding many timely issues, including prenatal and pre-implantation genetic diagnosis, assisted reproductive technologies, incidental findings in genetic testing, gene patenting, testing children for adult onset disorders, and direct-to-consumer testing.

Females Are Mosaics
X Inactivation and Sex Differences in Disease
Barbara Migeon

Women can be described as genetic mosaics because they have two distinctly different types of cells throughout their bodies. Unlike males, who have one X chromosome, females have two X chromosomes in every cell. Much has been written about the Y chromosome and its role in inducing maleness. This is the only resource about the X chromosome as a key to female development and the role of X-related factors in the etiology of sex differences in human disease. This new edition reflects research advances since the widely praised first edition. New advances include knowledge of species differences in mammalian X inactivation processes and silencing of the inactive X chromosome.

Foundations of Perinatal Genetic Counseling
Amber Mathiesen and Kali Roy

Foundations of Perinatal Genetic Counseling provides an overview of the core concepts needed to practice perinatal genetic counseling, including the basics of pregnancy, the genetic counseling appointment, family and pregnancy history, prenatal screening, prenatal diagnosis, common indications, carrier screening, management of high-risk pregnancy, assisted reproductive technology, preimplantation genetic screening and diagnosis, and
common situations arising in perinatal genetic counseling. It discusses general obstetrical information as it pertains to perinatal genetic counseling, including topics such as calculating gestational age, understanding gravidity and parity, and reproductive options. This book reviews the key components of a perinatal genetic counseling session and how to take a perinatal family, medical, and pregnancy history, as well as a prenatal risk evaluation based on age, family and pregnancy history, testing results, and ultrasound findings. It includes a detailed description of both prenatal screening and diagnostic testing options, including maternal serum screening, cell-free DNA testing, amniocentesis, and chorionic villus sampling. It also provides an explanation of carrier testing, including methods of testing, types of conditions, and indications for testing. This text provides information on the indications for referral to a perinatal genetic counselor such as age-related risks, personal and family history, ultrasound anomalies, teratogen exposure, recurrent pregnancy loss, and preconception counseling. It also reviews the management and types of referrals made in a high-risk pregnancy. Assisted reproductive technology is reviewed as well as descriptions of preimplantation genetic diagnosis and screening. It also describes common psychosocial and ethical situations encountered in perinatal genetic counseling.

Gardner and Sutherland’s Chromosome Abnormalities and Genetic Counseling
R.J. McKinlay Gardner and David J. Amor

Medical geneticists and genetic counselors regularly see families attending the genetic counseling clinic with questions about chromosome abnormalities. These families may themselves have had a child affected with a chromosome condition; or, there may have been a history elsewhere in the family. The presentation may have been due to infertility or reproductive loss. Questions may include the following: What is known about this condition? What caused this to happen? Is it likely to happen again? If so, is there a way to prevent it from happening again? The power of molecular approaches to chromosome analysis, coming to be routinely available in this second decade of the twenty-first century, has brought to our knowledge many new “chromosomal syndromes” to add alongside those long known from the days of classical cytogenetics. This new knowledge has increased our ability to answer the questions that families may have; but equally, it has raised challenges in interpretation, as molecular karyotyping has revealed more complexity in the way the human genome is constructed. This book distils the knowledge that has evolved in recent and olden times, and it presents the information in a way that will be helpful to the practitioner. In particular, the risks of recurrence, or of occurrence, of a particular chromosome disorder are clearly set forth. The application of chromosomal knowledge to reproductive conditions, both diagnostically and in management, is rehearsed.

Genetic Consultations in the Newborn
Robin D. Clark and Cynthia J. Curry

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This book was written to assist clinicians who care for newborns with congenital abnormalities in their diagnosis, genomic testing, and management. The goal was to make the evaluation of common neonatal anomalies and genetic syndromes accessible and understandable. In addition, the book may serve as an initial guide for practitioners in areas in which clinical genetic expertise is not readily available. As the book was being written, the testing paradigm shifted to a genomic approach: Chromosome analysis gave way to microarrays, and single gene testing was largely replaced by gene panels and exome sequencing. Thus, this book, which was initially intended as a clinical primer, of necessity became a resource for gene-based information as well.

Genetic Counseling Research: A Practical Guide
Ian M. MacFarlane, Patricia McCarthy Veach, and Bonnie S. LeRoy

Genetic Counseling Research: A Practical Guide is an online resource devoted to research methodology in genetic counseling. It offers step-by-step guidance for conducting research, from the development of a question to the publication of findings. Genetic counseling examples and practical tips guide readers through the research and publication processes. With a highly accessible, pedagogical approach, this resource will help promote quality research by genetic counselors and research supervisors—and in turn, increase the knowledge base for genetic counseling practice, other aspects of genetic counseling service delivery, and professional education.

Genetic Diseases of the Eye
Elias I Traboulsi, MD (ed.)

This highly anticipated new edition brings together an expert group of authors to provide a comprehensive, systematic sourcebook on genetic diseases of the eye. Each chapter emphasizes the clinical aspects of disease, tying them to the underlying molecular mechanisms and outlining current therapy. While the molecular underpinnings, testing methods and therapy of genetic disorders continues to evolve, the clinical aspects are well established and are emphasized in this book. A large number of color figures are utilized to illustrate the various chapters.
This book is a readable, reliable guide to the diagnosis and differential of inherited skin disorders to which generalists, paediatricians, dermatologists, and geneticists can refer during an examination. The new edition reflects the most up-to-date understanding of the molecular and genetic bases of heritable skin diseases. Each chapter describes the signs and symptoms of heritable skin diseases and enumerates pertinent associated clinical features and differential diagnoses. Non-dermatological signs are symptoms round out the information on each condition. Where appropriate, descriptions of histopathology at both the light and electron microscopic levels are included. Over 800 full-colour photographs illustrate the concepts discussed in the text. Annotated bibliographies at the end of each section direct readers to more extensive sources, and an updated listing of support groups for patients and their families supplements the resources for medical professionals.

The role of genetics is becoming increasingly important in all aspects of healthcare and particularly in the field of cancer care. Genetics for Health Professionals in Cancer Care: From Principles to Practice equips health professionals with the knowledge and skills required for all aspects of managing cancer family history. This includes taking an accurate cancer family history and drawing a family tree; understanding cancer biology, basic cancer genetics and the genes involved in hereditary breast, ovarian, prostate, colorectal, gastric and related gynaecological cancers and rare cancer predisposing syndromes; assessing cancer risk and communicating risk information; early detection and risk reducing measures available for those at increased risk and managing individuals with hereditary cancer.

This resource addresses both the molecular and clinical features of the obesity syndromes, providing hard-core information for researchers and practical guidelines for clinicians caring for obese patients.
This completely revised second edition of Genomic Medicine reflects the rapidly changing face of applied and translational genomics in the medical and health context and provides a comprehensive coverage of principles of genetics and genomics relevant to the practice of medicine. New topics included include bioinformatics, proteomics, microbial genomics and genomic education. Detailed discussions of genetic/genomic testing and screening and the ethical, legal, and social issues (ELSI) crucially address genethics and genomethics in the practice of Genomic Medicine. This online resource all includes clinical practice-oriented chapters highlighting genomic applications (array comparative genomic hybridization, exome genome sequencing and new generation sequencing) in clinical diagnosis of congenital developmental malformations, Mendelian genetic disorders, and complex cardiovascular, neuro-psychiatric, ophthalmic, dermatologic, inflammatory and pediatric disorders. It also discusses microbial genomics with emphasis on the role of genomics in targeted antimicrobial therapy and development of genomic class of new vaccines. New developments in gene/ cell-based somatic therapy, regenerative medicine and targeted molecular therapy are also covered.

Genomics and Health in the Developing World

Genomics and Health in the Developing World provides detailed and comprehensive coverage of population structures, human genomics, and genome variation - with particular emphasis on medical and health issues - in the emerging economies and countries of the developing world. With sections dedicated to fundamentals of genetics and genomics, epidemiology of human disease, biomarkers, comparative genomics, developments in translational genomic medicine, current and future health strategies related to genetic disease, and pertinent legislative and social factors, this resource highlights the importance of utilizing genetics/genomics knowledge to promote and achieve optimal health in the developing world. Grouped by geographic region, the chapters in this volume address inherited disorders in the developing world, the progress of diagnostic laboratory genetic testing, prenatal screening, and genetic counseling worldwide, the ethical and legal concerns of medical genetics in the developing world, and social, cultural, and religious issues related to genetic diseases across continents.