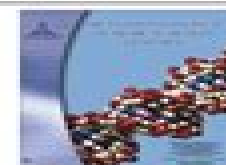




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EDUCATIONAL CORNER OF THE ISSUE

Basic concepts of medical genetics. Formal genetics, part 4

Mohammad Saad Zaghloul Salem

Faculty of Medicine, Ain-Shams University, Cairo, Egypt

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Non-traditional patterns of inheritance

Non-traditional/non-classical/non-Mendelian patterns of inheritance refer to different modes of transmission of genetic diseases that are not caused by single mutant genes. These diseases include a wide variety of genetically-determined disorders, e.g., polygenic diseases/chromosomal aberrations/mitochondrial disorders and multifactorial diseases. Non-traditional inheritance patterns are not compatible with the rules of inheritance that characterize the transmission of single gene disorders since they differ from each other in many respects like rates of occurrence, recurrence risk, sex predilection, spectrum of phenotypic variation and many others. The characteristics of these non-traditional patterns are dependent solely on the nature of the disease as regards its etiology and the specific pathogenetic mechanism(s) underlying its development. Recognition of these different inheritance patterns is important for the provision of proper counseling advice because they have different recurrence risks and different clinical and management implications. Non-traditional patterns of inheritance characterize diseases caused by specific pathogenetic mechanisms that include: defective genetic imprinting, uniparental disomy, nucleotide repeat expansion (tri/tetra/penta/hexa-nucleotide repeat expansion), mutations of mitochondrial genome, mutations caused by combined multifactorial (genetic/environmental) effects, mosaicism, chromosomal aberrations, microdeletion/microduplication/microtriplication defects and polygenic defects [1] (Table 1).

1. Genetic imprinting

Genetic imprinting refers to the predetermined functional status of a gene, a group of genes, part or most of a chromosome. This imprint might be imposed by different factors. For instance, it might be imposed by the parental origin, i.e. specific genes or sets of genes transmitted by the mother or the father may be expressed or suppressed according to their parent of origin, thus resulting in a specific monoallelic gene expression profile. This type of imprinting might be referred to as **parental imprinting** [2]. Alternatively, a gene might be silenced/suppressed/turned off or kept functioning following the completion of the critical stages of embryogenesis/differentiation/growth and development of the offspring. This type of imprinting might be referred to as **temporal imprinting**. A third type of imprinting that might be referred to as **spatial imprinting** is determined by the location of the gene, where a gene is suppressed or activated by regulatory mechanisms imposed by adjacent chromatin modifications (Table 2).

In diploid organisms, somatic cells possess two copies of the genome and each autosomal gene is represented by two copies, or alleles, with one copy inherited from each parent at fertilization. For the vast majority of autosomal genes, expression occurs from both alleles simultaneously. In mammals, however, a small proportion (<1%) of genes are parentally imprinted, meaning that gene expression occurs from only one allele. The expressed allele is dependent upon its parental origin. For example, the gene encoding insulin-like growth factor 2 (IGF2/Igf2) is only expressed from the allele inherited from the father [3].

Imprinting is a fundamental genomic regulatory mechanism during development and differentiation whereby overexpression of specific sets of maternal or paternal genes and silencing of other sets of maternal or paternal genes is mandatory for

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Basic Concepts In Medical Genetics

**Reed E. Pyeritz, Bruce R. Korf, Wayne
W. Grody**



Basic Concepts In Medical Genetics:

Basic Concepts in Medical Genetics Marshall Horwitz, Mary Beth Dinulos, Kris Carroll, 2000 **Basic Concepts in Medical Genetics** Marshall Horwitz, Mary Beth Dinulos, 2000 The main focus of this text is on medical genetics with a general approach to explanations It offers key concepts by the way of example to common problems and illustrations of concepts through working through some of the classic and prototypical problems that students encounter Introduction To Basic Concepts Of Medical Genetics Mohammad Salem, 2012 Medical genetics a branch of human genetics spans a wide spectrum of sub branches including basic clinical diagnostic therapeutic prophylactic and applied genetics This booklet I hope it be the first in a series is confined to two disciplines of basic genetics viz molecular genetics and pathogenetics that are concerned with studying the structure and function of the genetic material in health and disease states It aims at offering the basic concepts of these two subjects to clinicians and research workers in the field For clinicians beginning their specialization as medical geneticists I tried my best to offer as simplified as possible definitions and illustrations of the main points of both subjects concerning the structure and function of genes mutagens and disease causing mutations as well as the anti mutation mechanisms of the human genome For research workers I pointed out some of the most enigmatic aspects of the field that still await more research to disclose in order to go a step further in our way to understand what we think we know about the role of our genetic constitution in shaping our life Medical Genetics E-Book Lynn B. Jorde, John C. Carey, Michael J. Bamshad, 2019-09-27 Up to date and extensively revised to reflect recent advances in the genetics of common diseases as well as current progress in gene therapy Medical Genetics 6th Edition delivers easy to read highly visual coverage of this rapidly changing field This accessible practical text integrates key concepts with clinical practice highlighted by numerous illustrations tables concept summaries and more all designed to enhance effective learning and retention of complex material Discusses current topics including polygenic risk scores and their potential applications for diabetes cancer and heart disease and the latest sequencing technologies and their clinical application in genetic testing and diagnosis Offers a completely updated discussion of genetic testing modalities and applications Includes convenient concept summaries more than 230 photographs illustrations and tables as well as patient family vignettes that present valuable perspectives on disease and treatment Features Clinical Commentary boxes that demonstrate how the hard science of genetics has real applications to everyday patient problems preparing you for problem based integrated courses Illustrates key concepts with disease examples to demonstrate relevance to medicine Provides study questions for self assessment as well as 200 additional USMLE style questions online Enhanced eBook version included with purchase Your enhanced eBook allows you to access all of the text figures and references from the book on a variety of devices Medical Genetics Lynn B. Jorde, PhD, John C. Carey, MD, MPH, Michael J. Bamshad, MD, 2015-09-04 Popular for its highly visual clinical approach Medical Genetics delivers an accessible yet thorough understanding of this active and fast changing field Key updates in this new

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Medical Genetics Lynn B. Jorde, 1999 The emphasis of this book is on those aspects of medical genetics most useful in a modern clinical practice Clinical aspects of molecular genetics research have been incorporated throughout the spectrum of genetically determined diseases As such the book covers not only chromosomal and single gene disorders but also the more challenging multifactorial diseases

Medical Genetics Lynn B. Jorde, John C. Carey, Michael J. Bamshad, 2020-02-03

American Journal of Medical Genetics, 1986

Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics Reed E. Pyeritz, Bruce R. Korf, Wayne W. Grody, 2018-11-20 For decades Emery and Rimoin s Principles and Practice of Medical Genetics and Genomics has served as the ultimate resource for clinicians integrating genetics into medical practice With detailed coverage in contributions from over 250 of the world s most trusted authorities in medical genetics and a series of 11 volumes available for individual sale the Seventh Edition of this classic reference includes the latest information on seminal topics such as prenatal diagnosis genome and exome sequencing public health genetics genetic counseling and management and treatment strategies to

complete its coverage of this growing field for medical students residents physicians and researchers involved in the care of patients with genetic conditions This comprehensive yet practical resource emphasizes theory and research fundamentals related to applications of medical genetics across the full spectrum of inherited disorders and applications to medicine more broadly This volume Foundations summarizes basic theories concepts research areas and the history of medical genetics providing a contextual framework for integrating genetics into medical practice In this new edition clinically oriented information is supported by full color images and expanded sections on the foundations of genetic analytics next generation sequencing and therapeutics With regular advances in genomic technologies propelling precision medicine into the clinic Emery and Rimoin s Principles and Practice of Medical Genetics and Genomics Seventh Edition bridges the gap between high level molecular genetics and practical application and serves as an invaluable clinical tool for the health professionals and researchers Introduces genetic researchers students and health professionals to basic theories concepts research areas and the history of medical genetics offering a contextual framework for integrating genetics into medical practice Completely revised and up to date this new edition highlights traditional approaches and new developments in the field of medical genetics including cancer genetics genomic technologies genome and exome sequencing prenatal diagnosis public health genetics genetic counseling and single cell analysis for diagnosis Includes color images supporting identification concept illustration and method processing Features contributions by leading international researchers and practitioners of medical genetics

Genetics for the Health Sciences Heather Skirton,Christine Patch,2009 Based on their extensive experience of clinical work the authors emphasize the practical issues related to the healthcare of individuals and families Genetics for the Health Sciences takes an holistic approach from preconception to adulthood and addresses the false notion that clinical genetics is of relevance only to those who are planning a family The book enables nurses midwives genetic counselors and doctors to apply the general principles of genetics in their routine clinical practice As well as discussing the basic principles Genetics for the Health Sciences also describes the latest technologies and shows how these can be applied to clinical practice This is an essential text which helps all those in clinical healthcare understand the genetics they need in their professional roles It is also an ideal coursebook for students in the healthcare professions seeking an understanding of core genetic principles and how these are applied in practice

Reviews The text is a compact concise presentation of the basic concepts in genetics science and the impact of genetics across the lifespan The organization of the text increases its usefulness to clinicians as each specialty area could easily locate the information most pertinent to their work The text is well illustrated throughout again providing summarized information that is easily accessed An especially helpful feature of this text is the presence of multiple case studies in each chapter making this text particularly useful for teaching I think faculty teaching students in advanced practice nursing programs and allied health courses would find this text a succinct addition to their course However I could also see this text being useful in RN preparation curriculum It could easily be a text that would

carry over for several specialty areas and provide additional material specific to genetics in each area Genetics for Health Sciences A Handbook for Clinical Healthcare is a welcome addition to the rather sparse choices available for presenting genetics content in clinical practice curriculum Rebekah Hamilton ISONG June 2010 Tremendous progress in recent years has shaped the field of medical genetics which continues to expand to involve almost every aspect of human health Hence it is necessary for every healthcare professional to acquire a basic understanding of this science This is clearly the objective of this well edited and structured book by Skirton and Patch in its second updated edition The authors considerable experience in genetics through direct clinical practice in addition to organisational and academic roles can be appreciated in the practical aspect of their writing The book starts with useful definitions and alerts the reader to the wide applicability of genetics through clinical cases raising questions that are answered and discussed when relevant in later chapters Important aspects of genetic counselling from basic skills such as risk estimation using family trees to more complex ones related to effective ethical communication with the clients in the light of their needs and the different models of counselling are then discussed in two chapters However given their intrinsic function in medical genetics these issues are also efficiently tackled in the clinical cases outlined in each chapter The authors then describe important scientific concepts and techniques that are necessary for a good clinical understanding of genetics Although this section may seem short the additional resources provided at the end are very useful Likewise for all topics covered in the book the updated links provided serve as a handy catalogue for the health professional seeking extra information Next the concepts and techniques described in the first part of the book are applied directly to common genetic conditions and issues faced by different age groups from prenatal care to older adulthood in separate chapters This organisation puts into perspective the importance of genetics in different medical specialities and settings This book with its concise yet comprehensive explanation of a complex rapidly growing field should prove to be a valuable resource for a diverse audience of health professionals including midwives nurses and physicians who confront genetic issues during their daily practice without being specialists in medical genetics Nadine Taleb Journal of Medical Genetics When first asked to write this book review I thought to myself how it would be important to give a rounded report on both the book s strengths and weaknesses After reading the book however I realised that it is difficult to find weaknesses The authors Heather Skirton and Christine Patch between them have over 30 years of experience in clinical genetics Their experience of explaining genetics in their professional roles shines through in the way they have written the book They make complex genetic principles interesting and understandable The book does not baffle or patronise The book is easily navigated It is broken down into clear chapters that are ordered in a pleasingly logical way The first chapter sets the scene by introducing the reader to important concepts related to genetic health care including issues such as ethical practice the different forms of genetic testing and the impact of genetic conditions on families The next chapters then discuss the family tree counselling issues genetic science and public health genetics The remaining chapters then explore the core topics

relating to particular life stages from preconception to older adulthood It engages the reader from the start when it introduces seven core families that reappear throughout the book to highlight a number of key issues The text is peppered with clear illustrations useful step by step guides practical checklists and test yourself sections making the book lively and dynamic The test yourself sections make the book student friendly and would also make good teaching aids to lecturers and tutors At the start of the book there is a list of helpful websites and at the end of each chapter there is a list of resources for those who wish to extend their knowledge One of the main strengths of this book is its refreshingly practical approach All too often books can be written in a way in which it is difficult for the reader to transfer the knowledge that they have gained into their day to day practice The introduction states that the aim of this book is to enable those in healthcare to update their knowledge on topics related to genetics and genomics that have an impact in their daily work and apply it usefully in patient care Given the fact that genetics has increasingly become a core component in a number of specialties one would think that meeting this aim for all would be a tall order This book however eloquently does so using case examples and key practice points boxes throughout to demonstrate the clinical application of the topic under discussion Another of the book's main strengths is the way in which it promotes individualised care by enabling the reader to consider the impact of genetic conditions from the patient's point of view Genetics for the Health Sciences brings genetics into common healthcare settings It is highly recommended as an essential text for health care professionals in roles across all specialties It is also recommended to students lecturers social scientists to anyone who has an interest in genetics and wishes to extend their knowledge It is a joy to read and could be read from cover to cover Helen Thistlewood *Medical Genetics* Genetics is at the forefront of medicine and nurses are expected to have a basic understanding of the subject This handbook is well written and the authors do a good job of making this complex topic understandable The book lists prenatal childhood and adult genetic conditions that may involve testing and counselling Genetic services types of genetic testing and the impact of genetic conditions on families are discussed in depth and case studies highlight the emotional and psychological needs of patients and their families Guidelines for drawing family trees and practical examples of risk assessment are well explained The perspective of the patient regarding risk and lay knowledge is also considered All departments should have a copy of this book on their shelves Audrey Ardern Jones *Nursing Standard* *Progress in Medical Genetics* Arthur Gerald Steinberg, 1985 **Journal of Medical Genetics**, 1989 *Genetics in Medical Practice* Mihály Bartalos, 1968 **Medical Genetics and Society** Norio Fujiki, Victor Bulyzhenkov, Zbigniew Bańkowski, 1991 **Clinical Evaluation and Diagnostic Tests for Neuromuscular Disorders** Tulio E. Bertorini, 2002 This book provides expert advice from leading authorities on how and when to use the numerous evaluation tests now available for diagnosis and management of neuromuscular disorders Its practical approach will appeal to practicing neurologists as well as neurology residents and fellows All methods of testing and assessment are covered and an invaluable section on protocol summarizes the material A practical source of information

on how and when to select a test written by experts in the field Includes all methods of neuromuscular disorder tests and assessments Assists the practitioners in narrowing down the number of tests necessary to diagnose a condition

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