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Principles Of Human Biochemical Genetics

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The Principles of Human Biochemical Genetics Harry Harris, 1974 Gene mutations and single amino acid substitutions One gene one polypeptide chain Duplications and deletions and their effects on protein structure Gene mutations affecting rates of protein synthesis Quantitative and qualitative variation of enzymes The inborn errors of metabolism The blood group substances Enzyme and protein diversity in human populations Gene mutations and inherited disease The principles of human biochemical genetics Harry Harris, 1975 **The principles of human biochemical genetics, 2., rev. and enl** Harry Harris, 1975 Fundamentals of Genetic Epidemiology Muin J. Khoury, Terri H. Beaty, Bernice H. Cohen, 1993 This text integrates the principles methods and approaches of epidemiology and genetics in the study of disease aetiology The authors define the central theme of genetic epidemiology as the study of the role of genetic factors and their interaction with environmental factors in the occurrence of disease in populations **A Clinical Guide to Inherited Metabolic Diseases** Joe T. R. Clarke, 2005-12-08 This user friendly clinical handbook provides a clear and concise overview of how to go about recognizing and diagnosing inherited metabolic diseases The reader is led through the diagnostic process from the identification of those features of an illness suggesting that it might be metabolic through the selection of appropriate laboratory investigation to a final diagnosis The book is organized into chapters according to the most prominent presenting problem of patients with inherited metabolic diseases neurologic hepatic cardiac metabolic acidosis dysmorphism and acute catastrophic illness in the newborn It also includes chapters on general principles laboratory investigation neonatal screening and the principles of treatment This new edition includes much greater depth on mitochondrial disease and congenital disorders of glycosylation The chapters on neurological syndrome and newborn screening are greatly expanded as are those on laboratory investigation and treatment to take account of the very latest technological developments Basic Genetics Gurbachan S. Miglani, 2000 An exploration of basic genetics It features discussion of cell division and its significance chromosomes multiple alleles gene gene interactions genetic analysis in diploid and haploid eukaryotes mutations quantitative inheritance sex determination and genetic engineering **One Hundred Years of Chromosome Research and What Remains to be Learned** A. Lima-de-Faria, 2013-06-29 One Hundred Years of Chromosome Research What Remains to be Learned offers the reader a critical analysis of the observations and experiments that shaped the last 100 years of chromosome research as well as the ideas which prevailed during this period Emphasis is placed on what remains to be learned particularly in light of reality of the sequencing of DNA which leaves the previous era of chromosome research as a prehistoric event It is at this turning point that well formulated questions can be asked about many of the chromosome's properties which remain to be unveiled The author Lima de Faria is Professor Emeritus of Molecular Cytogenetics at Lund University Sweden previously Head of the Institute of Molecular Cytogenetics Lund University A Short History of Medical Genetics Peter S. Harper, 2008-10-24 An eminent geneticist veteran author OMMG

Series Editor and noted archivist Peter Harper presents a lively account of how our ideas and knowledge about human genetics have developed over the past century from the perspective of someone inside the field with a deep interest in its historical aspects Dr Harper has researched the history of genetics and has had personal contact with a host of key figures whose memories and experiences extend back 50 years and he has interviewed and recorded conversations with many of these important geneticists Thus rather than being a conventional history this book transmits the essence of the ideas and the people involved and how they interacted in advancing and sometimes retarding the field From the origins of human genetics through the contributions of Darwin Mendel and other giants the identification of the first human chromosome abnormalities and up through the completion of the Human Genome project this Short History is written in the author's characteristic clear and personal style which appeals to geneticists and to all those interested in the story of human genetics

The Genetics of Osteoporosis and Metabolic Bone Disease Michael J. Econs, 2000-05-12 The explosive growth in the field of molecular biology over the last two decades has started to make a great impact on clinical medicine Genes have been cloned for diseases that were poorly understood only a decade ago Additionally investigators are increasingly aware that there are strong genetic components to complex disorders such as osteoporosis that are not classically thought of as genetic disorders New insights into the pathogenesis of metabolic bone diseases have been obtained from investigations into the molecular biology of these diseases and new therapies will become available based on these new insights In The Genetics of Osteoporosis and Metabolic Bone Disease I have assembled an internationally renowned group of experts to write the various chapters Each of the authors is an expert in his/her field who is currently performing research on the content of their chapter and have made important contributions to the understanding of the clinical features and pathophysiology of metabolic bone disease and genetics The first part of The Genetics of Osteoporosis and Metabolic Bone Disease addresses issues related to genetic contributions to the development of osteoporosis and the many factors that must be considered when searching for genes that predispose to osteoporosis The second section addresses recent advances in the clinical and molecular biological aspects of inherited metabolic bone disorders The last section reviews the latest techniques for finding genes that predispose to metabolic bone diseases

Schizophrenia Irving I. Gottesman, James Shields, Daniel R. Hanson, 1982-06-30

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