

MOLECULAR BASIS OF INHERITANCE

CLASS 12 BIOLOGY

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reviews the principles and progress of medical genetics and epigenetics related to bone disease including genome wide association studies GWAS genomic profiling copy number variation prospects of gene therapy pharmacogenomics genetic testing and counseling as well as the generation and utilizing of mouse models The third section details advances in the genetics and molecular biology of bone and joint diseases both monogenic and polygenic as well as skeletal dysplasias and rarer bone disorders The fourth section highlights the central role of the parathyroids in calcium and skeletal homeostasis by reviewing the molecular genetics of hyperparathyroidism hypoparathyroidism endocrine neoplasias and disorders of the PTH and calcium sensing receptors The fifth section details molecular and cellular advances across associated renal disorders such as vitamin D and rickets Identifies and analyzes the genetic basis of bone disorders in humans and demonstrates the utility of mouse models in furthering the knowledge of mechanisms and evaluation of treatments Demonstrates how the interactions between bone and joint biology physiology and genetics have greatly enhanced the understanding of normal bone function as well as the molecular pathogenesis of metabolic bone disorders Summarizes the clinical genetic molecular animal model molecular pathology diagnostic counseling and treatment aspects of each disorder

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Kück,2013-03-09 Mycology the study of fungi originated as a subdiscipline of botany and was a descriptive discipline largely neglected as an experimental science until the early years of this century A seminal paper by Blakeslee in 1904 provided evidence for self incompatibility termed heterothallism and stimulated interest in studies related to the control of sexual reproduction in fungi by mating type specificities Soon to follow was the demonstration that sexually reproducing fungi exhibit Mendelian inheritance and that it was possible to conduct formal genetic analysis with fungi The names Burgeff Kniep and Lindegren are all associated with this early period of fungal genetics research These studies and the discovery of penicillin by Fleming who shared a Nobel Prize in 1945 provided further impetus for experimental research with fungi Thus began a period of interest in mutation induction and analysis of mutants for bio chemical traits Such fundamental research conducted largely with *Neurospora crassa* led to the one gene one enzyme hypothesis and to a second Nobel Prize for fungal research awarded to Beadle and Tatum in 1958 Fundamental research in biochemical genetics was extended to other fungi especially to *Saccharomyces cerevisiae* and by the mid 1960s fungal systems were much favored for studies in eukaryotic

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