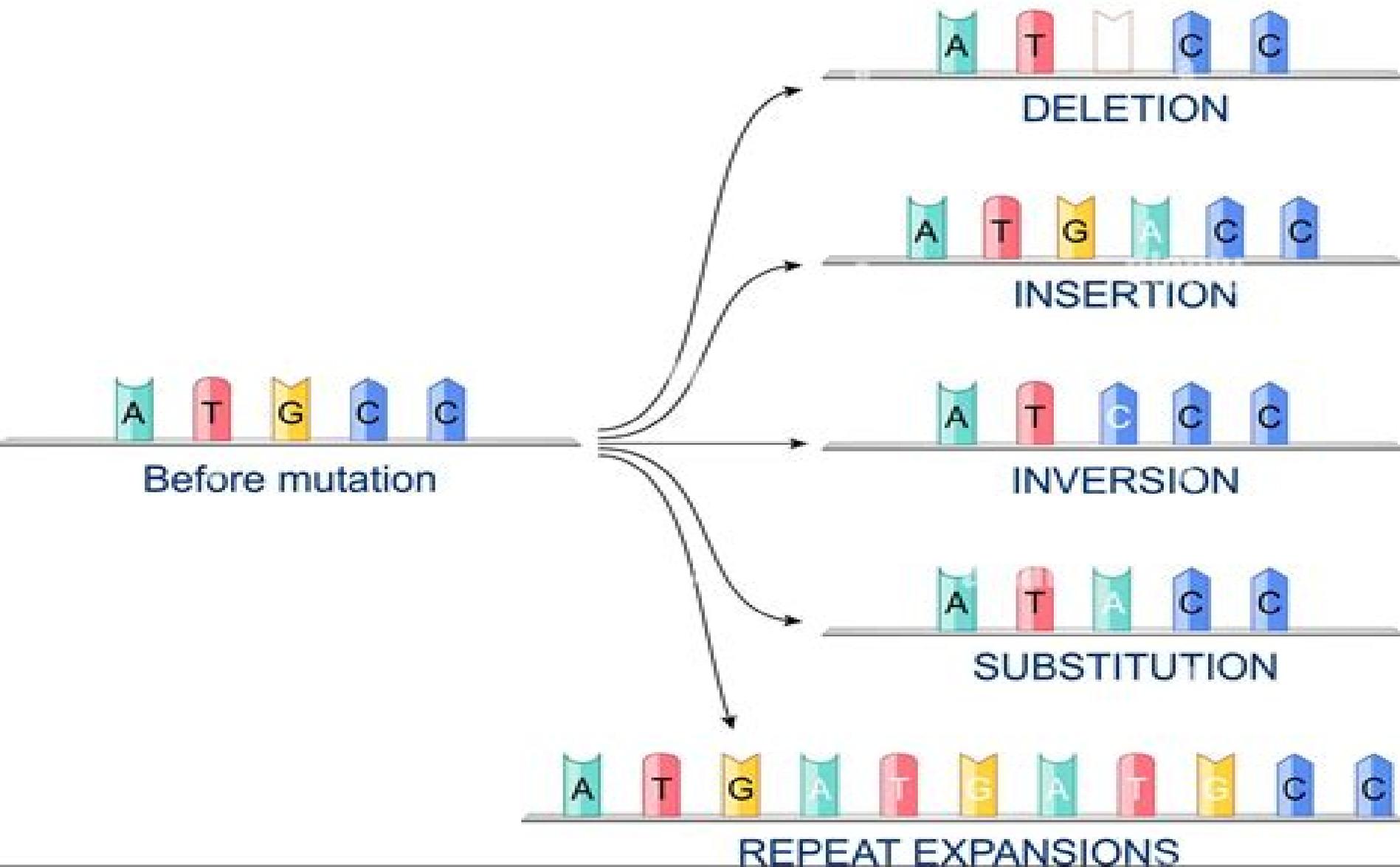


Genetic mutations



2012 Tesccc Mutations And Genetic Conditions

Armand Marie Leroi



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Stress-Induced Mutagenesis David Mittelman,2013-03-12 The discovery of stress induced mutagenesis has changed ideas about mutation and evolution and revealed mutagenic programs that differ from standard spontaneous mutagenesis in rapidly proliferating cells The stress induced mutations occur during growth limiting stress and can include adaptive mutations that allow growth in the otherwise growth limiting environment The stress responses increase mutagenesis specifically when cells are maladapted to their environments i e are stressed potentially accelerating evolution then The mutation mechanism also includes temporary suspension of post synthesis mismatch repair resembling mutagenesis characteristic of some cancers Stress induced mutation mechanisms may provide important models for genome instability underlying some cancers and genetic diseases resistance to chemotherapeutic and antibiotic drugs pathogenicity of microbes and many other important evolutionary processes This book covers pathways of stress induced mutagenesis in all systems The principle focus is mammalian systems but much of what is known of these pathways comes from non mammalian systems

Mutants Armand Marie Leroi,2005-01-25 Visit Armand Marie Leroi on the web <http://armandleroi.com/index.html> Stepping effortlessly from myth to cutting edge science Mutants gives a brilliant narrative account of our genetic code and the captivating people whose bodies have revealed it a French convent girl who found herself changing sex at puberty children who echoing Homer s Cyclops are born with a single eye in the middle of their foreheads a village of long lived Croatian dwarves one family whose bodies were entirely covered with hair was kept at the Burmese royal court for four generations and gave Darwin one of his keenest insights into heredity This elegant humane and engaging book captures what we know of the development of what makes us human Nature

Mobilizing Mutations Daniel Navon,2019-09-20 With every passing year more and more people learn that they or their young or unborn child carries a genetic mutation But what does this mean for the way we understand a person Today genetic mutations are being used to diagnose novel conditions like the XYY Fragile X NGLY1 mutation and 22q11.2 Deletion syndromes carving out rich new categories of human disease and difference Daniel Navon calls this form of categorization genomic designation and in Mobilizing Mutations he shows how mutations and the social factors that surround them are reshaping human classification Drawing on a wealth of fieldwork and historical material Navon presents a sociological account of the ways genetic mutations have been mobilized and transformed in the sixty years since it became possible to see abnormal human genomes providing a new vista onto the myriad ways contemporary genetic testing can transform people s lives Taking us inside these shifting worlds of research and advocacy over the last half century Navon reveals the ways in which knowledge about genetic mutations can redefine what it means to be ill different and ultimately human

Technologies for Detecting Heritable Mutations in Human Beings ,1986 *The Carriers* Anne Skomorowsky,2022 A tiny mutation on the X chromosome can shape a family s history Passed down from a carrier parent to a child fragile X syndrome is the most common inherited cause of intellectual disability

and autism Beyond that and a rarity among genetic disorders some fragile X carriers not only transmit the mutation but also experience related conditions themselves In such cases carriers can have tremors infertility and psychiatric disorders that complicate raising children with fragile X syndrome and all too often they suffer in silence The Carriers investigates this common but still little known genetic condition and its life altering consequences Anne Skomorowsky reveals how this disorder afflicts families across generations telling the stories of the mothers and grandparents of fragile X patients and considering how genes interact with family dynamics She interweaves the personal narratives and family histories of the people affected by fragile X disorders with clear and accessible explanations of the science behind them Skomorowsky unpacks the latest research on the fragile X mutation and explores the history of its discovery She highlights the roles of women as carriers caregivers and researchers who have made astonishing scientific breakthroughs over the last three decades The Carriers is an essential book for fragile X families including those just learning they are carriers and for all readers interested in the complexities of heredity the ethical dilemmas of genetic medicine and the relationship between genes and personality

Technologies for Detecting Heritable Mutations in Human Beings, 1986 *Mutation research*
Charlotte Auerbach, 1976-01-01 This book is intended for the senior undergraduate Honours student in genetics and for the postgraduate who wants a survey of the whole field or information on a special area within it In order to cater for readers with such different requirements I have made the list of references unusually large for a textbook It includes classical papers as well as very recent ones to the end of 1974 reviews as well as specialized articles elementary expositions from Scientific American as well as highly technical papers from journals on genetics and molecular biology In areas of active research I have given preference to the latest references which will lead the reader to earlier ones In addition to the references at the end of each chapter a bibliography at the end of the book lists relevant books and general reviews Apart from the first chapter the book is not written as a history of mutation research but throughout I have tried to emphasize the continuity of the problems concepts and ideas The reader will find many examples of this Muller s once famous and then almost forgotten classification of genes by their action has now been given biochemical reality by studies of gene action in vitro The problem of whether mutations can arise in non replicating genomes is one of the oldest in mutation research yet an unequivocal solution was obtained only recently with bacteriophage

Population and Biological Aspects of Human Mutation
Ernest B. Hook, 1981

Nonsense Mutation Correction in Human Diseases Fabrice Lejeune, Hana Benhabiles, Jieshuang Jia, 2016-03-14 Nonsense Mutation Correction in Human Diseases An Approach for Targeted Medicine provides an introduction on genetic diseases discusses the prevalence of nonsense mutations the consequences of a nonsense mutation for the expression of the mutant gene and the presentation of the nonsense mediated mRNA decay NMD It presents the mechanism of action and rationale associated with each strategy to correct nonsense mutations with the results of clinical trials to further support this basis In addition the book shows how it may be possible to combine several of these strategies to

ultimately improve the efficiency of correction also suggesting the future goals and objectives to improve treatment modalities in this evolving sphere of personalized medicine

Tay-Sachs Disease Robert J. Desnick, Michael M. Kaback, 2001 Tay Sachs disease is a rare hereditary disease caused by a genetic mutation that leaves the body unable to produce an enzyme necessary for fat metabolism in nerve cells producing central nervous system degeneration In infants it is characterized by progressive mental deterioration blindness paralysis epileptic seizures and death by age four Adult onset Tay Sachs occurs in persons who have a genetic mutation that is similar but allows some production of the missing enzyme There is no treatment for Tay Sachs A test to determine whether an infant is carrying the Tay Sachs disease was introduced in 1969 However work continues to be done to help find a cure Because there is no cure for this deadly disease genetic research is essential Advances in Genetics presents an eclectic mix of articles of use to all human and molecular geneticists They are written and edited by recognized leaders in the field and make this an essential series of books for anyone in the genetics field

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