

## DNA

DNA is a molecule in the cell nucleus that contains instructions for making proteins. It is made of four different bases: adenine (A), thymine (T), guanine (G), and cytosine (C). A segment of DNA that contains the information for making a protein is called a gene. In the process of transcription, DNA that makes up a gene is copied into a complementary molecule called messenger RNA (mRNA).

## RNA

mRNA is also made of four bases: adenine (A), uracil (U), guanine (G), and cytosine (C). mRNA moves from the nucleus to the cytoplasm where it interacts with ribosomes, the protein factories of the cell. There, through a process called translation, mRNA is translated into amino acids. A sequence of three mRNA bases is called a codon, and each codon is translated into a specific amino acid. There are 20 different kinds of amino acids in humans.

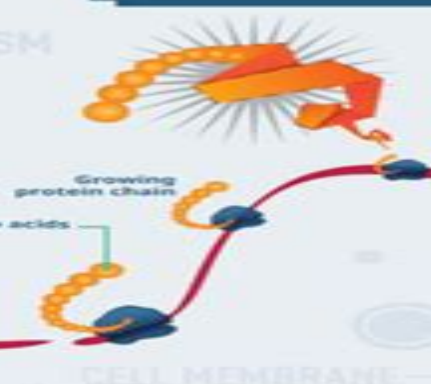


# GENETIC — CHANGES — AND CANCER

## HOW GENETIC INFORMATION CREATES PROTEINS

## PROTEIN

As an mRNA molecule is translated, a chain of amino acids is formed. The chain eventually folds into a three-dimensional protein. The shape of a protein determines its function. Proteins have millions of functions in cells.



## TYPES OF GENETIC MUTATIONS IN CANCER

DNA alterations can affect the structure, function, and amount of the corresponding proteins. All of these effects can change a cell's behavior from normal to cancerous. For example, a genetic alteration can intensify or eliminate the protein's function, which could make cells divide uncontrollably. Many different kinds of genetic mutations are found in cancer cells, including missense, nonsense, and frameshift mutations and chromosome rearrangements.

### MISSENSE MUTATION

|          |                         |                            |                        |                       |
|----------|-------------------------|----------------------------|------------------------|-----------------------|
| Original | CTA<br>LEU<br>(Leucine) | TGG<br>TRP<br>(Tryptophan) | GTA<br>VAL<br>(Valine) | DNA<br>Amino<br>Acids |
| Mutation | CTA<br>LEU<br>(Leucine) | TGC<br>CYS<br>(Cysteine)   | GTA<br>VAL<br>(Valine) | DNA<br>Amino<br>Acids |

A missense mutation is a change of a single DNA base that results in a change in the amino acid sequence. Sometimes a single amino acid change can greatly alter the protein's function.

### NONSENSE MUTATION

|          |                         |                            |                        |                       |
|----------|-------------------------|----------------------------|------------------------|-----------------------|
| Original | CTA<br>LEU<br>(Leucine) | TGG<br>TRP<br>(Tryptophan) | GTA<br>VAL<br>(Valine) | DNA<br>Amino<br>Acids |
| Mutation | CTA<br>LEU<br>(Leucine) | TGA<br>STOP                | GTA<br>VAL<br>(Valine) | DNA<br>Amino<br>Acids |

A nonsense mutation is a change of a single DNA base that creates a "stop" codon, which terminates translation. The result is a shortened protein that may not function or that may have an abnormal function.

### FRAMESHIFT MUTATION

|          |                         |                            |                         |                       |
|----------|-------------------------|----------------------------|-------------------------|-----------------------|
| Original | CTA<br>LEU<br>(Leucine) | TGG<br>TRP<br>(Tryptophan) | GTA<br>VAL<br>(Valine)  | DNA<br>Amino<br>Acids |
| Mutation | CTA<br>LEU<br>(Leucine) | ATG<br>MET<br>(Methionine) | GGT<br>GLY<br>(Glycine) | DNA<br>Amino<br>Acids |

A frameshift mutation results from the addition or removal of DNA bases that shifts the DNA sequence and the corresponding amino acid sequence. The result is a protein whose sequence, structure, and function are very different from those of the original protein.

### CHROMOSOME REARRANGEMENTS

DNA is wound tightly into structures called chromosomes. Chromosome rearrangements can occur when a piece of a chromosome breaks and is lost entirely (deletion), moves to a different chromosomal location (translocation), flips directions (inversion), or is repeated (duplication). These rearrangements can alter several genes at once. For example, they can generate fusion genes, in which parts of two separate genes are joined together. Proteins made from fusion genes sometimes cause cancer.



# Cancer Genetics

**Ros Eng, Douglas Easton, Bruce  
Ponder, Charis Eng**



## **Cancer Genetics:**

**Principles of Clinical Cancer Genetics** Daniel C. Chung, Daniel A. Haber, 2010-07-20 Advances in genetics are transforming estimates of an individual's risk of developing cancer and approaches to prevention and management of cancer in those who may have increased susceptibility. Identifying and caring for patients with hereditary cancer syndromes and their family members present a complex clinical, scientific, and social challenge. This textbook by leading experts at Massachusetts General Hospital Cancer Center highlights the current understanding of the genetics of hereditary cancers of the breast, ovary, colorectum, stomach, pancreas, kidney, skin, and endocrine organs. Practical guidelines for the use of genetic testing, cancer screening, and surveillance, prophylactic surgery, and promising targeted therapeutic agents are discussed. In addition, ongoing research involving genome-wide screens to identify novel modest risk-associated genetic loci are explored along with new approaches to the application of genetic markers in guiding therapeutic options. **Principles of Cancer**

**Genetics** Fred Bunz, 2022-06-25 This popular textbook, now in its third edition, provides a theoretical framework for understanding why cancers arise, how they develop, and how they can be treated. Particular attention is devoted to the origins of cancer and the application of evolutionary theory to explain how mutant cell populations tend to expand and spread. Focused on the genes and signaling pathways involved in the most common tumors, *Principles of Cancer Genetics* is a highly readable account that will be of interest to anyone who would like to attain a basic understanding of cancer biology. Students who have completed introductory coursework in genetics, biology, and biochemistry; medical students and medical house staff will find this book to be a useful starting point toward mastery of this complex but fascinating topic. This updated edition delves into the critical interactions between growing tumors and the immune system and introduces the concepts of T cell activation, immunoediting, and immune evasion. Novel strategies for cancer diagnosis and prognosis, including new roles for next-generation sequencing and liquid biopsies, as well as established and emerging therapeutic modalities, are now described in detail. For laypersons, students, and researchers in other fields with a general interest in cancer, this book provides an accessible overview enriched with many easy-to-understand illustrations. For advanced students considering future study in the field of oncology and cancer research, this concise book is a useful guide to the basic principles that underlie our understanding of cancer. **Cancer Genetics** Boris Pasche, 2010-06-02 Cancer Genetics is a collection of

chapters covering the key recent developments in cancer genetics which have an impact on clinical care. The target audience will be physicians and scientists who need to be apprised on the most recent developments in the field. *Genetic*

*Predisposition to Cancer, 2Ed* Ros Eng, Douglas Easton, Bruce Ponder, Charis Eng, 2003-10-29 Over the last 20 years it has become increasingly apparent that the occurrence of many cancers can have an inherited basis. This book examines the principles underlying genetic predisposition to cancer and will be relevant to practising oncologists, geneticists, and other professionals interested in this rapidly expanding field. Coverage is comprehensive. [A Guide to Cancer Genetics in Clinical](#)

Practice Sue Clark, 2009-05-01 FIRST PRIZE Winner in the Oncology category of the 2010 BMA Medical Book Competition Intensive research over the last fifteen years has yielded a vast expansion in our understanding of the role of inheritance and genetics in a variety of cancers Several inherited conditions have been identified which result in a high risk of various cancers some of these were previously recognized but the genetic basis underlying them has now been elucidated This knowledge is now entering the sphere of routine clinical care Surgeons gastroenterologists gynaecologists oncologists endocrinologists and many others need an awareness of these disorders an understanding of genetic testing and when it is indicated and how to manage patients with inherited cancer predisposition and their families This book covers the basic concepts of cancer genetics The common inherited cancer syndromes are each dealt with in greater depth with the current management outlined This book is aimed at all clinicians who may encounter these conditions in their practice It aims to facilitate identification of high risk individuals and families to inform interaction with geneticists and other subspecialists to provide a basis for patient management and to stimulate interest in these fascinating conditions **Cancer Genetics** Henry T. Lynch, 1976 *Clinical Cancer Genetics* Kenneth Offit, 2003-05-07 Written by a world recognized leader in this emerging field Clinical Cancer Genetics provides an updated and expanded treatment of Kenneth Offit's seminal text on the clinical management associated with syndromes of cancer predisposition with a thorough review of the relevant molecular genetics This second edition features new coverage of pharmacogenetics gene therapy trials high throughput genotyping and microarrays and includes a new focus on epigenetic events in carcinogenesis within background chapter on cancer genetics Expanded coverage highlights more uncommon and rare cancer predisposition syndromes **Cancer Genetics and Genomics for Personalized Medicine** Il-Jin Kim, 2017-04-11 This book covers almost all fields of cancer genetics and genomics for personalized medicine Targeted therapy or precision medicine or personalized medicine is becoming a standard treatment for many diseases including cancer However how much do we know about the personalized medicine approach This lucid book helps undergraduate and graduate students professional researchers and clinicians to better understand the key concept of personalized medicine The most up to date topics on personalized medicine in this book cover the recent trends in and updates on lung gastric liver breast and other types of cancers Circulating tumor cell cell free circulating DNA and microRNAs are discussed as new diagnostic and prognostic markers for cancer The avastin mouse model is also discussed for maximizing treatment efficacy and prognosis prediction and so is microenvironment as a drug resistance mechanism With classical and new pathological approaches the book provides a systemic overview of personalized immunotherapies and hyperthermic intraperitoneal chemotherapy followed by new emerging fields of hereditary cancer thereby equipping readers to eventually contribute in developing more advanced tools and therapies for curing cancer **Counseling About Cancer** Katherine A. Schneider, 2011-10-26 Important scientific discoveries and ever changing guidelines for how to identify and manage patients with hereditary cancer syndromes are constantly evolving This Third Edition of Counseling About Cancer is

completely updated and expanded to feature five entirely new chapters on breast cancer colon cancer other solid tumors clients and families and genetic test results and follow up This is the only reference and clinical book on the market for cancer genetics counselors and other healthcare providers who must quickly assimilate complex and ever changing data on the hereditary risk for cancer *Cancer Genetics for the Clinician* Gail L. Shaw, 2012-12-06 Cancer genetics is a quickly growing field within oncology The ability to identify individuals at high risk for cancer improves the chance of early prevention and detection of cancer The results of genetic testing affect quality of life employment and ability to be insured This volume will provide an overview of cancer genetics inherited cancer susceptibility and available services and testing including both the risks and benefits of testing Some of the topics covered include genetics of breast and ovarian cancer testing minors for inherited cancer risk chemoprevention of heritable cancer risk genetics of colorectal cancer insurance issues in genetic testing for cancer ethical and legal issues in genetic testing for cancer susceptibility testing for breast cancer risk in the Ashkenazim estimating individualized risk of breast cancer genetic counseling for the individual with inherited cancer susceptibility and components of a genetic cancer risk clinic While heritable cancers account for between five and ten percent of all cancer cases molecular alterations attributable to specific inherited cancer susceptibility may give us important clues into the mechanism by which cancer occurs not only in predisposed individuals but also for sporadic cases

*The Genetics of Cancer* Gajanan V. Sherbet, M. S. Lakshmi, 1997-06-22 Written by internationally recognized experts The Genetics of Cancer provides up to date information and insight into the genetic basis of cancer and the mechanisms involved in cancer invasion and its secondary spread This volume presents the deregulation of the cell cycle in tumor development and integrates the function of tumor suppressor genes oncogenes and metastasis associated genes in the pathogenesis and progression of cancer The Genetics of Cancer will be useful to all graduate students clinicians and researchers working in the fields of cancer biology genetics and molecular biology Clonal evolution of the metastasis phenotype Cell Cycle regulation Apoptosis in tumour growth and metastasis Angiogenesis in cancer Cell surface glycoproteins and their receptors Proteinases and their inhibitors in cancer invasion Oncogenes and cancer metastasis Developmental genes Tumour suppressor genes Metastasis suppressor genes Dominant metastasis associated genes Inherited Susceptibility to Cancer William D. Foulkes, Shirley V. Hodgson, 1998-06-28 Many cancers both common and rare are known to have a hereditary predisposition and advances in genetics have clarified the risks and in some cases the mechanisms of cancer developing in an individual First published in 1998 this important contribution to the literature of cancer genetics covers all the key issues reviewing both the technology behind genetic risk assessment and the ethical dilemmas it poses It is divided into two parts The first deals with ethical legal and social issues The second systematically outlines current knowledge of the inheritance patterns of many different cancer types both from a site by site perspective and for special groups This authoritative volume will be of interest to oncologists physicians and surgeons in other specialties and to health professionals in the areas of primary care

counselling and cancer risk assessment      *Pediatric Cancer Genetics* Nathaniel H. Robin, Meagan Farmer, 2017-08-22 Get a quick expert overview of the many key facets of pediatric cancer genetics with this concise practical resource by Dr Nathaniel H Robin and Meagan Farmer MS CGC MBA Ideal for pediatric oncologists and all providers who care for children this easy to read reference addresses the remarkable potential of genetic testing as well as the complexities of choosing the correct test understanding the results and counseling the family Features a wealth of information on pediatric cancer genetics including the epidemiology and biology of cancer and the genetic evaluation process and role of genetic counsellors Highlights examples of syndromes that present in childhood and increase susceptibility to cancer Discusses the genetic evaluation process in context of the multidisciplinary care of children with cancer Considers the ethical and legal issues of genetic testing in children and provides illustrative case examples Consolidates today's available information and guidance in this timely area into one convenient resource

**Genetics for Health Professionals in Cancer Care** Chris Jacobs, Pat A. Webb, Lorraine Robinson, Patricia Webb, 2014 Genetics for Health Professionals in Cancer Care equips health professionals with the knowledge and skills required for all aspects of managing cancer family history including discussing the challenges raised and provides practical guidance on setting up a cancer family history clinic in primary and secondary care

**Cancer Genetics: A Clinical Approach** Xavier Llor, Erin Wysong Hofstatter, 2021-11-19 The ultimate guide to caring for and communicating with patients suffering with hereditary cancer syndromes Providing the essential tools needed to understand clinical cancer genetics Cancer Genetics A Clinical Approach builds your mastery of differential diagnosis formulation all the available genetic testing options at your disposal interpreting results logically and accurately and educating patients and their families about effective cancer prevention methods This unparalleled resource walks you through the process of effectively and thoughtfully counseling patients from pre test counseling and relaying test results to navigating the legal implications of hereditary conditions and coping with the resulting psychological challenges Cancer Genetics A Clinical Approach is ideal for both specialized cancer genetics clinics or any healthcare professional seeking to improve their skills in identifying patients with possible hereditary cancer syndromes and recognizing which ones should be referred to a high risk clinical genetics specialty program

**Cancer** H. Leon Bradlow, Jack Fishman, Michael P. Osborne, 1997 This volume contains papers presented at a conference on Genetics and the Environment which was held by the Strang International Cancer Prevention Center on November 15th and 16th 1996 in New York The text seeks to describe the evolution and

current status of human cancer genetics and the current role of molecular diagnosis It elucidates the significance of mutations in specific cancer related genes and the ethical implications of genetic testing Environmental factors such as tobacco and toxic chemicals that have an important impact on cancer risk are also discussed A Cancer in the Family Theodora Ross, MD, PhD, Siddhartha Mukherjee, 2017-01-31 A Kirkus Best Book of 2016 Oncologist and cancer gene hunter Theo Ross delivers the first authoritative go to for people facing a genetic predisposition for cancer There are 13 million people with cancer in the United States and it s estimated that about 1 3 million of these cases are hereditary Yet despite advanced training in cancer genetics and years of practicing medicine Dr Theo Ross was never certain whether the history of cancers in her family was simple bad luck or a sign that they were carriers of a cancer causing genetic mutation Then she was diagnosed with melanoma and for someone with a dark complexion melanoma made no sense It turned out there was a genetic factor at work Using her own family s story the latest science of cancer genetics and her experience as a practicing physician Ross shows readers how to spot the patterns of inherited cancer how to get tested for cancer causing genes and what to do if you have one With a foreword by Siddhartha Mukherjee prize winning author of The Emperor of All Maladies this will be the first authoritative go to for people facing inherited cancer this book empowers readers to face their genetic heritage without fear and to make decisions that will keep them and their families healthy **A Practical Guide to Human Cancer Genetics** Shirley V. Hodgson, William D. Foulkes, Charis Eng, Eamonn Maher, 2013-11-27 This is a comprehensive and up to date guide to the diagnosis clinical features and management of inherited disorders conferring cancer susceptibility It is fully updated with much molecular screening and management information It covers risk analysis and genetic counselling for individuals with a family history of cancer It also discusses predictive testing and the organisation of the cancer genetics service There is information about the genes causing Mendelian cancer predisposing conditions and their mechanism of action It aims to provide such details in a practical format for geneticists and clinicians in all disciplines Cancer Principles and Practice of Oncology: Handbook of Clinical Cancer Genetics Ellen Matloff, 2013-03-25 This practical user friendly guidebook will allow the clinician to search under disease site for the hereditary cancer syndromes relevant for his her patient s cancer For example a gynecologist oncologist whose patient has ovarian cancer can turn to the Ovary chapter and quickly read a summary of all of the hereditary cancer syndromes that include ovarian cancer She can learn the questions she should be asking when expanding that patient s personal and family history which genes are most relevant whether to refer that patient on for genetic counseling and testing and how to manage that patient long term if the patient is mutation positive or negative The same holds true for the practicing oncologist surgeon urologist endocrinologist gynecologist primary care physician physician s assistant advanced practice nurse and any other clinician seeing a patient who has had cancer This guidebook also contains an overview article on genetic counseling and testing and several in depth articles on issues that are up and coming in the field of hereditary cancer *Cancer Genetics and Psychotherapy* Parvin

Mehdipour,2017-09-20 The aim of this book is to provide the readers with the most comprehensive and latest accounts of research and development in this field by emphasizing on the manner of relation between doctors and cancer patients in direction of improving the patients style of life This book partly will deal with psychotherapy by considering cancer patients benefits hazards and also social impacts including life style The social supports as the key and influential paradigms will be challenged as a comparative insight by considering the global unity in order to provide a reasonable model to improve the interaction between cancer and psychological nest In this book the real stories of cancer patient will be also provided The initial insight of sections includes 1 Brief classifications and key points of clinical and histopatological aspects of each organ 2 Brief view of genetic alterations in each organ 3 Therapeutic aspects 4 Brief classifications and key points of Psychology in cancer 5 The interactions of clinical aspects with psychological field



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