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Next Generation Sequencing Technologies in Medical Genetics



Springer

Next Generation Sequencing Technologies In Medical Genetics Springerbriefs In Genetics

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Next Generation Sequencing Technologies In Medical Genetics Springerbriefs In Genetics:

Next Generation Sequencing Technologies in Medical Genetics C. Alexander Valencia, M. Ali Pervaiz, Ammar Husami, Yaping Qian, Kejian Zhang, 2013-10-16 This book introduces readers to Next Generation Sequencing applications in medical genetics. The authors discuss the direct application of next generation sequencing to medicine specifically laboratory medicine or molecular diagnostics. The first part of the book contains chapters on sanger sequencing NGS technologies targeted amplification and capture and exome sequencing. The second part of the book focuses on genetic disorders diagnosed by NGS prenatal diagnosis muscular dystrophies mitochondrial disorders diagnosis and challenges in molecular diagnosis. Recent developments and potential future trends in NGS sequencing applications are highlighted as well. Next Generation Sequencing Technologies and Challenges in Sequence Assembly Sara El-Metwally, Osama M. Ouda, Mohamed Helmy, 2014-04-19 The introduction of Next Generation Sequencing NGS technologies resulted in a major transformation in the way scientists extract genetic information from biological systems revealing limitless insight about the genome transcriptome and epigenome of any species. However with NGS came its own challenges that require continuous development in the sequencing technologies and bioinformatics analysis of the resultant raw data and assembly of the full length genome and transcriptome. Such developments lead to outstanding improvements of the performance and coverage of sequencing and improved quality for the assembled sequences. Nevertheless challenges such as sequencing errors expensive processing and memory usage for assembly and sequencer specific errors remain major challenges in the field. This book aims to provide brief overviews of the NGS field with special focus on the challenges facing the NGS field including information on different experimental platforms assembly algorithms and software tools assembly error correction approaches and the correlated challenges. Computational Intelligence Techniques in Health Care P.V. Lakshmi, Wengang Zhou, P. Satheesh, 2016-04-20 This book presents research on emerging computational intelligence techniques and tools with a particular focus on new trends and applications in health care. Healthcare is a multi-faceted domain which incorporates advanced decision making remote monitoring healthcare logistics operational excellence and modern information systems. In recent years the use of computational intelligence methods to address the scale and the complexity of the problems in healthcare has been investigated. This book discusses various computational intelligence methods that are implemented in applications in different areas of healthcare. It includes contributions by practitioners technology developers and solution providers. **Next Generation Sequencing and Sequence Assembly** Ali Masoudi-Nejad, Zahra Narimani, Nazanin Hosseinkhan, 2013-07-09 The goal of this book is to introduce the biological and technical aspects of next generation sequencing methods as well as algorithms to assemble these sequences into whole genomes. The book is organized into two parts: part 1 introduces NGS methods and part 2 reviews assembly algorithms and gives a good insight to these methods for readers new to the field. Gathering information about sequencing and assembly methods together helps both biologists and

computer scientists to get a clear idea about the field Chapters will include information about new sequencing technologies such as ChIp seq ChIp chip and De Novo sequence assembly

Microbiome-Host Interactions D. Dhanasekaran,Dhiraj Paul,N. Amaresan,A. Sankaranarayanan,Yogesh S. Shouche,2021-03-31 Microbiota are a promising and fascinating subject in biology because they integrate the microbial communities in humans animals plants and the environment In humans microbiota are associated with the gut skin and genital oral and respiratory organs The plant microbial community is referred to as holobiont and it is influential in the maintenance and health of plants which themselves play a role in animal health and the environment The contents of Microbiome Host Interactions cover all areas as well as new research trends in the fields of plant animal human and environmental microbiome interactions The book covers microbiota in polar soil environments in health and disease in *Caenorhabditis elegans* and in agroecosystems as well as in rice root and actinorhizal root nodules speleothems and marine shallow water hydrothermal vents Moreover this book provides comprehensive accounts of advanced next generation DNA sequencing metagenomic techniques high throughput 16S rRNA sequencing and understanding nucleic acid sequence data from fungal algal viral bacterial cyanobacterial actinobacterial and archaeal communities using QIIME software Quantitative Insights into Microbial Ecology FEATURES Summarizes recent insight in microbiota and host interactions in distinct habitats including Antarctic hydrothermal vents speleothems oral skin gut feces reproductive tract soil root root nodules forests and mangroves Illustrates the high throughput amplicon sequencing computational techniques involved in the microbiota analysis downstream analysis and visualization and multivariate analysis commonly used for microbiome analysis Describes probiotics and prebiotics in the composition of the gut microbiota skin microbiome impact in dermatologic disease prevention and microbial communities in the reproductive tract of humans and animals Presents information in a reachable way for students teachers researchers microbiologists computational biologists and other professionals who are interested in strengthening or enlarging their knowledge about microbiome analysis with next generation DNA sequencing in the different branches of the sciences

Clinical Applications for Next-Generation Sequencing Urszula Demkow,Rafal Ploski,2015-09-10 Clinical Applications for Next Generation Sequencing provides readers with an outstanding postgraduate resource to learn about the translational use of NGS in clinical environments Rooted in both medical genetics and clinical medicine the book fills the gap between state of the art technology and evidence based practice providing an educational opportunity for users to advance patient care by transferring NGS to the needs of real world patients The book builds an interface between genetic laboratory staff and clinical health workers to not only improve communication but also strengthen cooperation Users will find valuable tactics they can use to build a systematic framework for understanding the role of NGS testing in both common and rare diseases and conditions from prenatal care like chromosomal abnormalities up to advanced age problems like dementia Fills the gap between state of the art technology and evidence based practice Provides an educational opportunity which advances patient care through the transfer of NGS to real

world patient assessment Promotes a practical tool that clinicians can apply directly to patient care Includes a systematic framework for understanding the role of NGS testing in many common and rare diseases Presents evidence regarding the important role of NGS in current diagnostic strategies Clinical Genomics Shashikant Kulkarni,Somak Roy,2014-11-10 Clinical Genomics provides an overview of the various next generation sequencing NGS technologies that are currently used in clinical diagnostic laboratories It presents key bioinformatic challenges and the solutions that must be addressed by clinical genomicists and genomic pathologists such as specific pipelines for identification of the full range of variants that are clinically important This book is also focused on the challenges of diagnostic interpretation of NGS results in a clinical setting Its final sections are devoted to the emerging regulatory issues that will govern clinical use of NGS and reimbursement paradigms that will affect the way in which laboratory professionals get paid for the testing Simplifies complexities of NGS technologies for rapid education of clinical genomicists and genomic pathologists towards genomic medicine paradigm Tried and tested practice based analysis for precision diagnosis and treatment plans Specific pipelines and meta analysis for full range of clinically important variants **Next Generation Sequencing** Lee-Jun C. Wong,2013-05-31 In recent years owing to the fast development of a variety of sequencing technologies in the post human genome project era sequencing analysis of a group of target genes entire protein coding regions of the human genome and the whole human genome has become a reality Next Generation Sequencing NGS or Massively Parallel Sequencing MPS technologies offers a way to screen for mutations in many different genes in a cost and time efficient manner by deep coverage of the target sequences This novel technology has now been applied to clinical diagnosis of Mendelian disorders of well characterized or undefined diseases discovery of new disease genes noninvasive prenatal diagnosis using maternal blood and population based carrier testing of severe autosomal recessive disorders This book covers topics of these applications including potential limitations and expanded application in the future **Next Generation Sequencing Technologies and Challenges in Sequence Assembly** M. Sc Sara El-Metwally,Osama M. Ouda,Mohamed Helmy,2014-05-31 **Next Generation Sequencing** Jerzy Kulski,2016-01-14 Next generation sequencing NGS has surpassed the traditional Sanger sequencing method to become the main choice for large scale genome wide sequencing studies with ultra high throughput production and a huge reduction in costs The NGS technologies have had enormous impact on the studies of structural and functional genomics in all the life sciences In this book Next Generation Sequencing Advances Applications and Challenges the sixteen chapters written by experts cover various aspects of NGS including genomics transcriptomics and methylomics the sequencing platforms and the bioinformatics challenges in processing and analysing huge amounts of sequencing data Following an overview of the evolution of NGS in the brave new world of omics the book examines the advances and challenges of NGS applications in basic and applied research on microorganisms agricultural plants and humans This book is of value to all who are interested in DNA sequencing and bioinformatics across all fields of the life sciences **Next Generation Sequencing (NGS) for**

Rare Diseases Diagnosis Xiu-An Yang,Hu Hao,Yanling Yang,Can Liao,2022-01-28 Challenges and Opportunities of Next-Generation Sequencing for Biomedical Research ,2012-10-04 Published continuously since 1944 the Advances in Protein Chemistry and Structural Biology serial has been a continuous essential resource for protein chemists Covering reviews of methodology and research in all aspects of protein chemistry including purification expression proteomics modeling and structural determination and design each volume brings forth new information about protocols and analysis of proteins while presenting the most recent findings from leading experts in a broad range of protein related topics This volume features articles on Challenges and Opportunities of Next Generation Sequencing for Biomedical Research Covers reviews of methodology and research in all aspects of protein chemistry Brings forth new information about protocols and analysis of proteins while presenting the most recent findings from leading experts in a broad range of protein related topics

Assessing Genomic Sequencing Information for Health Care Decision Making Institute of Medicine,Board on Health Sciences Policy,Roundtable on Translating Genomic-Based Research for Health,2014-08-19 Rapid advances in technology have lowered the cost of sequencing an individual s genome from the several billion dollars that it cost a decade ago to just a few thousand dollars today and have correspondingly greatly expanded the use of genomic information in medicine Because of the lack of evidence available for assessing variants evaluation bodies have made only a few recommendations for the use of genetic tests in health care For example organizations such as the Evaluation of Genomic Applications in Practice and Prevention working group have sought to set standards for the kinds of evaluations needed to make population level health decisions However due to insufficient evidence it has been challenging to recommend the use of a genetic test An additional challenge to using large scale sequencing in the clinic is that it may uncover secondary or incidental findings genetic variants that have been associated with a disease but that are not necessarily related to the conditions that led to the decision to use genomic testing Furthermore as more genetic variants are associated with diseases new information becomes available about genomic tests performed previously which raises issues about how and whether to return this information to physicians and patients and also about who is responsible for the information To help develop a better understanding of how genomic information is used for healthcare decision making the Roundtable on Translating Genomic Based Research for Health of the Institute of Medicine held a workshop in Washington DC in February 2014 Stakeholders including clinicians researchers patients and government officials discussed the issues related to the use of genomic information in medical practice Assessing Genomic Sequencing Information for Health Care Decision Making is the summary of that workshop This report compares and contrasts evidence evaluation processes for different clinical indications and discusses key challenges in the evidence evaluation process **Next Generation Sequencing Based Clinical Molecular Diagnosis of Human Genetic Disorders** Lee-Jun C. Wong,2017-05-15 Next Generation Sequencing technology has been applied to clinical diagnoses in the past three to five years using various approaches including target gene panels

and whole exomes The purpose of this book is to summarize the experiences the results advantages and disadvantages along with future development in the area of NGS based molecular diagnosis This up to date volume will not only provide the readers working with Next Generation Sequencing the basics on how to apply the technology to molecular diagnosis but will present the results and experience of practical application Next Generation Sequencing in Cancer Research, Volume 2 Wei Wu,Hani Choudhry,2015-04-25 Latest generation sequencing revolutionizes the fields of cancer research and oncology This follow up volume focuses more extensively on single cell sequencing of cancer and trials in drug resistance Another exciting feature is the bioinformatics tools given that can be used on cancer genome studies Scientists around the world are attempting to find the root cause of cancer A reasonable cancer treatment plan and potential cure is more optimistic now with the unfolding of the cancer genome The collective knowledge of how to leverage next generation sequencing in cancer research is paving the way The important information provided in this volume will move the field forward in developing novel targeted cancer therapies **Clinical Genomics, Second Edition** Shashikant Kulkarni,Somak Roy,2023-05-01 Clinical Genomics Second Edition provides a full overview of next generation sequencing NGS technologies that are currently used in clinical diagnostic laboratories The book presents key bioinformatic challenges and solutions that must be addressed by clinical genomicists and genomic pathologists such as specific pipelines for identification of the full range of variants that are clinically important It focuses on the challenges of diagnostic interpretation of NGS results in a clinical setting with later sections devoted to emerging regulatory issues governing the clinical use of NGS and reimbursement paradigms that will affect the way in which laboratory professionals get compensated for testing Following the wide use of the current edition by clinicians and researchers alike this fully revised new edition addresses the evolving landscape of genomics medicine along with recent advances in our understanding of molecular mechanisms of human diseases and developmental biology In addition the book updates on sequencing chemistry and platforms bioinformatics algorithms clinical informatics IT infrastructure and emerging applications of artificial intelligence in genomics **High-Throughput Next Generation Sequencing** Young Min Kwon,Steven C. Ricke,2011-08-25 Due to their novel concepts and extraordinary high throughput sequencing capacity the next generation sequencing methods allow scientists to grasp system wide landscapes of the complex molecular events taking place in various biological systems including microorganisms and microbial communities These methods are now being recognized as essential tools for a more comprehensive and deeper understanding of the mechanisms underlying many biological processes In High Throughput Next Generation Sequencing Methods and Applications experts in the field explore the most recent advances in the applications of next generation sequencing technologies with an emphasis on microorganisms and their communities however the methods described in this book will also offer general applications relevant to the study of any living organisms Written in the highly successful Methods in Molecular Biology™ series format chapters include introductions to their respective topics lists of the necessary materials

and reagents step by step readily reproducible laboratory protocols and key tips on troubleshooting and avoiding known pitfalls Comprehensive and cutting edge High Throughput Next Generation Sequencing Methods and Applications is an excellent collection of chapters to aid all scientists who wish to apply these innovative research tools to enhance their own pursuits in microbiology and also biology in general Next-Generation Sequencing and Sequence Data Analysis Kuo Ping Chiu,2015-11-04 Nucleic acid sequencing techniques have enabled researchers to determine the exact order of base pairs and by extension the information present in the genome of living organisms Consequently our understanding of this information and its link to genetic expression at molecular and cellular levels has lead to rapid advances in biology genetics biotechnology and medicine Next Generation Sequencing and Sequence Data Analysis is a brief primer on DNA sequencing techniques and methods used to analyze sequence data Readers will learn about recent concepts and methods in genomics such as sequence library preparation cluster generation for PCR technologies PED sequencing genome assembly exome sequencing transcriptomics and more This book serves as a textbook for students undertaking courses in bioinformatics and laboratory methods in applied biology General readers interested in learning about DNA sequencing techniques may also benefit from the simple format of information presented in the book **Advancing Development of Synthetic Gene Regulators** Anandhakumar Chandran,2017-09-19 This book focuses on an outside the box notion by utilizing the powerful applications of next generation sequencing NGS technologies in the interface of chemistry and biology In personalized medicine developing small molecules targeting a specific genomic sequence is an attractive goal N methylpyrrole P N methylimidazole I polyamides PIPs are a class of small molecule that can bind to the DNA minor groove First a cost effective NGS ion torrent platform based Bind n Seq was developed to identify the binding specificity of PIP conjugates in a randomized DNA library Their biological influences rely primarily on selective DNA binding affinity so it is important to analyze their genome wide binding preferences However it is demanding to enrich specifically the small molecule bound DNA without chemical cross linking or covalent binding in chromatinized genomes Herein is described a method that was developed using high throughput sequencing to map the differential binding sites and relative enriched regions of non cross linked SAHA PIPs throughout the complex human genome SAHA PIPs binding motifs were identified and the genome level mapping of SAHA PIPs enriched regions provided evidence for the differential activation of the gene network A method using high throughput sequencing to map the binding sites and relative enriched regions of alkylating PIP throughout the human genome was also developed The genome level mapping of alkylating the PIP enriched region and the binding sites on the human genome identifies significant genomic targets of breast cancer It is anticipated that this pioneering low cost high through put investigation at the sequence specific level will be helpful in understanding the binding specificity of various DNA binding small molecules which in turn will be beneficial for the development of small molecule based drugs targeting a genome level sequence Next Generation Sequencing in Cancer Research Wei Wu,Hani Choudhry,2013-08-04 This volume

provides an interdisciplinary perspective of applying Next Generation Sequencing NGS technology to cancer research It aims to systematically introduce the concept of NGS a variety of NGS platforms and their practical implications in cancer biology This unique and comprehensive text will integrate the unprecedented NGS technology into various cancer research projects as opposed to most books which offer a detailed description of the technology This volume will present true experimental results with concrete data processing pipelines discuss the bottleneck of each platform for real project in cancer research In additional single cancer cell sequencing as the proof of concept will be introduced in this book along with cutting edge information provided will help the intended audience to develop a comprehensive understanding of the NGS technology and practical whole genome sequencing data analysis and rapidly translate into their own research specifically in the field of cancer biology

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