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Precision Cancer Medicine **Clinical Applications for Next-Generation Sequencing Genotyping and Bioforensics of Ricinus Communis** *Typing Highly Degraded DNA Using Target Enrichment* **Genetics and Genomics in Medicine** Mapping and Sequencing the Human Genome Pediatric Board Study Guide **Genomic Applications in Pathology Cytogenomics** *An Integrative DNA Sequencing Panel To Assess Mismatch Repair Deficiency* Next Generation Sequencing Technologies in Medical Genetics **Understanding Genetics** *Multicenter Validation of Cancer Gene Panel-based Next-generation Sequencing for Translational Research and Molecular Diagnostics* **JIMD Reports - Case and Research Reports, 2012/3 Droplet Microfluidics Treatment Strategies and Survival Outcomes in Breast Cancer** *Clinical Genomics* **AACR 2018 Proceedings: Abstracts 3028-5930** Hematopathology of the Young, An Issue of the Clinics in Laboratory Medicine, E-Book **Biochemical and Molecular Basis of Pediatric Disease** Protein-targeted DNA Scission **AACR 2019 Proceedings: Abstracts 1-2748 Targeting DNA Repair and the DNA Damage Response: Beyond the Standard PI3 Kinase-like Kinases Bacterial Pathogenesis** *Assessing Genetic Risks* **Clinical Genome Sequencing Molecular Biology of the Cell Problem Solving through Precision Oncology** *Lung Cancer, An Issue of Hematology/Oncology Clinics of North America, E-Book* Integrative analysis of single-cell and/or bulk multi-omics sequencing data **Targeted Therapies in Cancer: An Investigation of Sulfhydryl Cross-linked DNA Delivery Systems for Liver Targeted Gene Therapy** Early Phase Cancer Immunotherapy Prognostic Epigenetics **Protein Modifications in Epigenetic Dysfunctional Diseases: Mechanisms and Potential Therapeutic Strategies** *Handbook Of Forensic Genetics: Biodiversity And Heredity In Civil And Criminal Investigation Next Generation Sequencing Based Diagnostic Approaches in Clinical Oncology* Modern Techniques in Cytopathology Evolution of Translational Omics Characterizing the Utility of Cell-Free DNA in Prostate Cancer

Protein Modifications in Epigenetic Dysfunctional Diseases: Mechanisms and Potential Therapeutic Strategies May 30 2020

Molecular Biology of the Cell Feb 05 2021

Targeting DNA Repair and the DNA Damage Response: Beyond the Standard PI3 Kinase-like Kinases Jun 11 2021

An Investigation of Sulfhydryl Cross-linked DNA Delivery Systems for Liver Targeted Gene Therapy Sep 02 2020

An Integrative DNA Sequencing Panel To Assess Mismatch Repair Deficiency Jul 25 2022 Detecting mismatch repair (MMR) deficiency requires serial testing of both germline and tumour DNA using several assays to determine the underlying mechanism of MMR gene disruption. We have created an integrated targeted panel (MultiMMR) that tests for multiple sources of genome variation from a single aliquot of tumour or normal DNA. We have profiled 11 genes related to MMR deficiency or hereditary cancer syndromes on 82 individuals. For each sample, we performed hybrid capture of a single DNA sequencing library constructed using methylated adapters for parallel bisulfite and conventional sequence analysis. MultiMMR recapitulated clinical testing in 22/24 cases and was able to explain the mechanism of MMR loss in an additional 28 patients. This study has shown the utility of integrated mutation, copy number, and methylation profiling to detect hereditary and somatic causes of MMR deficiency. MultiMMR amalgamates the current step-wise and complex clinical testing workflow into a single assay.

AACR 2018 Proceedings: Abstracts 3028-5930 Nov 16 2021

Genomic Applications in Pathology Sep 26 2022 Genomic Applications in Pathology provides a state-of-the art review of the scientific principles underlying next generation genomic technologies and the required bioinformatics approaches to analyses of the daunting amount of data generated by current and emerging genomic technologies. Implementation roadmaps for various clinical assays such as single gene, gene panels, whole exome and whole genome assays are discussed together with issues related to reporting, including the pathologist's role in interpretation and clinical integration of genomic tests results. Genomic applications for site-specific solid tumors and hematologic neoplasms are detailed, as well as genomic applications in pharmacogenomics, inherited genetic diseases, and infectious

diseases. The latest iteration of practice recommendations and guidelines in genomic testing, put forth by stakeholder professional organizations such as the Association for Molecular Pathology and the College of American Pathologists, are also discussed in the volume, as well as regulatory issues and laboratory accreditation related to genomic testing. Written by experts in the field, Genomic Applications in Pathology provides a comprehensive resource that is of great value to practicing molecular pathologists, hematopathologists, other subspecialized pathologists, general pathologists, pathology trainees, oncologists, and geneticists.

Evolution of Translational Omics Jan 25 2020 Technologies collectively called omics enable simultaneous measurement of an enormous number of biomolecules; for example, genomics investigates thousands of DNA sequences, and proteomics examines large numbers of proteins. Scientists are using these technologies to develop innovative tests to detect disease and to predict a patient's likelihood of responding to specific drugs. Following a recent case involving premature use of omics-based tests in cancer clinical trials at Duke University, the NCI requested that the IOM establish a committee to recommend ways to strengthen omics-based test development and evaluation. This report identifies best practices to enhance development, evaluation, and translation of omics-based tests while simultaneously reinforcing steps to ensure that these tests are appropriately assessed for scientific validity before they are used to guide patient treatment in clinical trials.

Clinical Applications for Next-Generation Sequencing Apr 02 2023 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

Droplet Microfluidics Feb 17 2022 Droplet microfluidics offers tremendous potential as an enabling technology for high-throughput screening. It promises to yield novel techniques for personalised medicine, drug discovery, disease diagnosis, establishing chemical libraries, and the discovery of new materials. Despite the enormous potential to contribute to a broad range of applications, the expected adoption has not yet been seen, partly due to the interdisciplinary nature and the fact that, up until now, information has been scattered across the literature. This book goes a long way to addressing these issues. Edited by two leaders, this book has drawn together expertise from around the globe to form a unified, cohesive resource for the droplet microfluidics community. Starting with the basic theory of droplet microfluidics before introducing its use as a tool, the reader will be treated to chapters on important techniques, including robust passive and active droplet manipulations and applications such as single cell analysis, which is key for drug discovery. This book is a go-to resource for the community yearning to adopt and promote droplet microfluidics into different applications and will interest researchers and practitioners working across chemistry, biology, physics, materials science, micro- and nano-technology, and engineering.

JIMD Reports - Case and Research Reports, 2012/3 Mar 21 2022 JIMD Reports publishes case and short research reports in the area of inherited metabolic disorders. Case reports highlight some unusual or previously unrecorded feature relevant to the disorder, or serve as an important reminder of clinical or biochemical features of a Mendelian disorder.

Biochemical and Molecular Basis of Pediatric Disease Sep 14 2021 Biochemical and Molecular Basis of Pediatric Disease, Fifth Edition has been a well-respected reference in the field for decades. This revision continues the

strong focus on understanding the pathogenesis of pediatric disease, emphasizing not only the important role of the clinical laboratory in defining parameters that change with the disease process, but also the molecular basis of many pediatric diseases. Provides a fully-updated resource with more color illustrations Focuses on the biochemical and molecular basis of disease as well as the analytical techniques Defines important differences in the pathophysiology of diseases, comparing childhood with adult

Next Generation Sequencing Based Diagnostic Approaches in Clinical Oncology Mar 28 2020 Dr. Anton Buzdin (AB) is employed by Omicsway Corp. (USA). AB received grants from Amazon and Microsoft Azure to support cloud computations. Dr. Xinmin Li is director of JCCC Shared Genomics Resource, the University of California, Los Angeles, CA Dr. Ye Wang is Director of Gene testing Department (Core Lab) of Qingdao Central Hospital, the Second Affiliated Hospital of Qingdao University

Assessing Genetic Risks Apr 09 2021 Raising hopes for disease treatment and prevention, but also the specter of discrimination and "designer genes," genetic testing is potentially one of the most socially explosive developments of our time. This book presents a current assessment of this rapidly evolving field, offering principles for actions and research and recommendations on key issues in genetic testing and screening. Advantages of early genetic knowledge are balanced with issues associated with such knowledge: availability of treatment, privacy and discrimination, personal decision-making, public health objectives, cost, and more. Among the important issues covered: Quality control in genetic testing. Appropriate roles for public agencies, private health practitioners, and laboratories. Value-neutral education and counseling for persons considering testing. Use of test results in insurance, employment, and other settings.

Hematopathology of the Young, An Issue of the Clinics in Laboratory Medicine, E-Book Oct 16 2021 In this issue of Clinics in Laboratory Medicine, guest editor Vinodh Pillai brings considerable expertise to the topic of pediatric hematopathology. Provides in-depth, clinical reviews on pediatric hematopathology, providing actionable insights for clinical practice. Presents the latest information on this timely, focused topic under the leadership of experienced editors in the field; Authors synthesize and distill the latest

research and practice guidelines to create these timely topic-based reviews.

Mapping and Sequencing the Human Genome Nov 28 2022 There is growing enthusiasm in the scientific community about the prospect of mapping and sequencing the human genome, a monumental project that will have far-reaching consequences for medicine, biology, technology, and other fields. But how will such an effort be organized and funded? How will we develop the new technologies that are needed? What new legal, social, and ethical questions will be raised? Mapping and Sequencing the Human Genome is a blueprint for this proposed project. The authors offer a highly readable explanation of the technical aspects of genetic mapping and sequencing, and they recommend specific interim and long-range research goals, organizational strategies, and funding levels. They also outline some of the legal and social questions that might arise and urge their early consideration by policymakers.

Typing Highly Degraded DNA Using Target Enrichment Jan 31 2023 Forensic genetic profiling is the process of targeting unique positions within the human genome for identity testing of biological DNA evidence. Forensic profiling of highly degraded DNA samples is one of the primary challenges faced by forensic analysts. These compromised biological samples are difficult to genetically profile, due to the highly fragmented nature of the target molecules, using traditional methods which centers around the detection of short tandem repeats (STRs). For STR typing to be successful, DNA must be relatively intact in order to amplify by PCR. Molecular biology approaches have been developed that may be applied to severely degraded samples to increase the capability of DNA profiling. Targeting single nucleotide polymorphisms (SNPs) holds potential as their amplicons can be designed to be substantially smaller than those for STRs, making these markers a viable alternative for typing degraded (fragmented) DNA. Additionally, rolling circle amplification (RCA) can be exploited as a tool as it has the capacity to amplify all genomic DNA in a circular template present in a sample. A circular molecule essentially creates an infinitely long template for amplification. RCA generates linear tandem copies of the circular template sequence. However, nuclear DNA is not circular and thus RCA cannot be used to its full potential. CircLigase II is an enzyme that circularizes single-stranded DNA. Thus, it may be possible to generate circular DNA from the highly degraded fragments of challenged

samples. Molecular inversion probes (MIPs) are an alternative circle-based enrichment approach. A MIP is a single-stranded oligonucleotide that contains two target-specific arms flanking a SNP of interest (capture) and internal PCR primer binding sites for controlled amplification. The two target-specific arms hybridize to the target DNA, the gap is filled resulting in the complementary state of the SNP of interest, the MIP dissociates from the target, and the target site is amplified employing the internally incorporated primer binding sites. Coupled to massively parallel sequencing (MPS), both circle-based approaches were attempted with limited to no success. Reverse Complement PCR (RC-PCR) was pursued to address the same problem of analyzing degraded DNA. RC-PCR is an innovative, one-step PCR target enrichment technology adapted for the amplification of highly degraded (fragmented) DNA. It provides simultaneous amplification and tagging of a targeted sequence construct in a single, closed-tube assay. A human identification (HID) RC-PCR panel was designed targeting 27 identity SNPs generating targets only 50 base pairs in length. In a single reaction, the complete sequencing construct is produced which is essential for MPS library preparation. The RC-PCR approach produced reliable and concordant genotyping results as well as demonstrated a sensitivity of detection of a majority of alleles down to 60 pg of input DNA. In addition, RC-PCR showed robustness tolerating known PCR inhibitors, especially calcium and collagen. The RC-PCR system may be an effective alternative to current forensic genetic methods in the analysis of highly degraded DNA.

Treatment Strategies and Survival Outcomes in Breast Cancer Jan 19 2022 Treatment strategies for breast cancer are wide-ranging and often based on a multi-modality approach, depending on the stage and biology of the tumour and the acceptance and tolerance of the patient. They may include surgery, radiotherapy, and systemic therapy (endocrine therapy, chemotherapy, and targeted therapy). Advances in technologies such as oncoplastic surgery, radiation planning and delivery, and genomics, and the development of novel systemic therapy agents alongside their evaluation in ongoing clinical trials continue to strive for improvements in outcomes. In this Special Issue, we publish a collection of studies looking at all forms of therapeutic strategies for early and advanced breast cancer, focusing on their outcomes, notably survival.

Lung Cancer, An Issue of Hematology/Oncology Clinics of North America, E-Book Dec 06 2020 In this issue of Hematology/Oncology Clinics, guest editors Drs. Sarah B. Goldberg and Roy S. Herbst bring their considerable expertise to the topic of Lung Cancer. Top experts in the field cover key topics such as state-of-the-art pathologic and molecular testing; neoadjuvant vs. adjuvant therapy for early-stage NSCLC; new therapies on the horizon (immunotherapy, ADCs, etc.); supportive care and symptom management; smoking cessation; and more. Contains 15 relevant, practice-oriented topics including lung cancer screening; advances in surgical techniques for lung cancer; non-surgical therapy for early-stage lung cancer; locally advanced lung cancer; what's new in small cell lung cancer; and more. Provides in-depth clinical reviews on lung cancer, offering actionable insights for clinical practice. Presents the latest information on this timely, focused topic under the leadership of experienced editors in the field. Authors synthesize and distill the latest research and practice guidelines to create clinically significant, topic-based reviews.
Characterizing the Utility of Cell-Free DNA in Prostate Cancer Dec 26 2019 Motivation: Prostate cancer remains the most commonly diagnosed neoplasm in American men, with existing biomarkers (i.e. PSA, nomograms, MRI) having varying levels of sensitivity and specificity in identifying more advanced and potentially aggressive disease. Tumor tissue biopsies remain the gold standard for confirming the presence of prostate cancer, as well as evaluating the genomic heterogeneity and clonal architecture that may be predictive of poor outcomes (i.e. recurrence and metastasis). However, tissue biopsies are limited in their ability to comprehensively assess tumors, and may lead to underestimation of disease grade and stage. These hurdles may be overcome with cell-free DNA (cfDNA), which allows for minimally invasive, repeated sampling through blood draws. This is particularly important when tumor tissue is unavailable during active surveillance or disease monitoring for the detection of residual disease or progression. Additionally, genomic interrogation via cfDNA sequencing typically requires prior knowledge of existing mutations from a patient's tumor. The work presented here leverages a number of methods to ensure broad, yet sensitive detection of cfDNA variants for patients with localized prostate cancer, including sequencing with a machine-learning guided 2.5Mb targeted panel. In this dissertation, I

investigate the use of cfDNA concentration, fragment size, and sequencing to identify advanced prostate cancer, as well as detect somatic mutations present in patient-matched tumors.Methods: The patient cohort included in these studies are composed of 268 individuals: 34 healthy individuals, 112 men with localized prostate cancer who underwent radical prostatectomy (RP), and 122 men with metastatic castration-resistant prostate cancer (mCRPC). Plasma cfDNA concentration and fragment size were quantified with a Qubit fluorometer or Bioanalyzer utilizing a chip-based capillary electrophoresis method for nucleic acid analysis. Low-pass whole-genome and targeted sequencing were used to identify single nucleotide variants (SNVs), small insertions and deletions (indels), and copy number alterations (CNAs) for a subset of patients. Plasma cfDNA was barcoded with duplex Unique Molecular Identifiers (UMIs) to construct consensus reads and improve variant detection by leveraging duplicate reads and sequence complementarity of the two DNA strands. Extensive tissue sampling was used to capture tumor heterogeneity and provide a patient-specific gold standard for comparison of matched cfDNA.Results and Conclusions: Patients with advanced mCRPC had higher cfDNA concentration than men with localized disease or healthy controls, and those with localized disease had shorter average fragment sizes than controls. Importantly, cfDNA concentration and fragment size remained independent predictors after adjusting for age and PSA. We found that targeted sequencing of cfDNA-without a priori patient-specific tumor mutation information-identified somatic alterations found in matched tumor tissue from multiple regions, potentially allowing for dynamic monitoring of emerging resistant subclones throughout the course of disease. Detection of these concordant variants was associated with seminal vesicle invasion and the number of somatic variants found in the tumor tissue samples, predicating its use for patients with poor prognostic factors in a localized setting. Similar to cfDNA concentration, plasma cfDNA mutational burden was also found to increase with disease severity. The results from our studies demonstrate the ability of cfDNA to identify somatic variants in patients with heterogeneous, localized prostate cancer.

Clinical Genome Sequencing Mar 09 2021 Clinical Genome Sequencing: Psychological Aspects thoroughly details key psychological factors to consider

while implementing genome sequencing in clinical practice, taking into account the subtleties of genetic risk assessment, patient consent and best practices for sharing genomic findings. Chapter contributions from leading international researchers and practitioners cover topics ranging from the current state of genomic testing, to patient consent, patient responses to sequencing data, common uncertainties, direct-to-consumer genomics, the role of genome sequencing in precision medicine, genetic counseling and genome sequencing, genome sequencing in pediatrics, genome sequencing in prenatal testing, and ethical issues in genome sequencing. Applied clinical case studies support concept illustration, making this an invaluable, practical reference for this important and multifaceted topic area within genomic medicine. Features contributions from leading international researchers and practitioners versed in the psychosocial dimensions of genomic medicine implementation Presents clinical case studies that support concept illustration, making this an invaluable reference for students, researchers, and clinicians looking for practical guidance in this important and multifaceted topic area Details the current state of genomic testing, expectations of genome sequencing, patient consent, patient responses to sequencing data, uncertainties in genome sequencing, direct-to-consumer genome sequencing, and more

Cytogenomics Aug 26 2022 Cytogenomics demonstrates that chromosomes are crucial in understanding the human genome and that new high-throughput approaches are central to advancing cytogenetics in the 21st century. After an introduction to (molecular) cytogenetics, being the basic of all cytogenomic research, this book highlights the strengths and newfound advantages of cytogenomic research methods and technologies, enabling researchers to jump-start their own projects and more effectively gather and interpret chromosomal data. Methods discussed include banding and molecular cytogenetics, molecular combing, molecular karyotyping, next-generation sequencing, epigenetic study approaches, optical mapping/karyomapping, and CRISPR-cas9 applications for cytogenomics. The book's second half demonstrates recent applications of cytogenomic techniques, such as characterizing 3D chromosome structure across different tissue types and insights into multilayer organization of chromosomes, role of repetitive elements and noncoding RNAs in human genome, studies in topologically associated domains,

interchromosomal interactions, and chromoanagenesis. This book is an important reference source for researchers, students, basic and translational scientists, and clinicians in the areas of human genetics, genomics, reproductive medicine, gynecology, obstetrics, internal medicine, oncology, bioinformatics, medical genetics, and prenatal testing, as well as genetic counselors, clinical laboratory geneticists, bioethicists, and fertility specialists. Offers applied approaches empowering a new generation of cytogenomic research using a balanced combination of classical and advanced technologies Provides a framework for interpreting chromosome structure and how this affects the functioning of the genome in health and disease Features chapter contributions from international leaders in the field

Genotyping and Bioforensics of Ricinus Communis Mar 01 2023 The castor bean plant (Ricinus communis) is a member of the family Euphorbiaceae. In spite of its common name, the castor plant is not a true bean (i.e., leguminous plants belonging to the family, Fabaceae). Ricinus communis is native to tropical Africa, but because the plant was recognized for its production of oil with many desirable properties, it has been introduced and cultivated in warm temperate regions throughout the world (Armstrong 1999 and Brown 2005). Castor bean plants have also been valued by gardeners as an ornamental plant and, historically, as a natural rodenticide. Today, escaped plants grow like weeds throughout much of the southwestern United States, and castor seeds are even widely available to the public for order through the Internet. In this study, multiple loci of chloroplast noncoding sequence data and a few nuclear noncoding regions were examined to identify DNA polymorphisms present among representatives from a geographically diverse panel of Ricinus communis cultivated varieties. The primary objectives for this research were (1) to successfully cultivate castor plants and extract sufficient yields of high quality DNA from an assortment of castor cultivated varieties, (2) to use PCR and sequencing to screen available universal oligos against a small panel of castor cultivars, (3) to identify DNA polymorphisms within the amplified regions, and (4) to evaluate these DNA polymorphisms as appropriate candidates for assay development (see Figure 1). Additional goals were to design, test and optimize assays targeting any DNA polymorphisms that were discovered and to rapidly screen many castor cultivars to determine

the amount of diversity present at that particular locus. Ultimately, the goal of this study was to construct a phylogeographic tree representing the genetic relationships present among Ricinus communis cultivars from diverse geographic regions. These research objectives were designed to test the hypothesis that cultivated varieties of Ricinus communis from various geographic regions can be distinguished from one another based on differences present at the genetic level. In addition, the present study sought to determine the amount of diversity present among Ricinus communis cultivars.

Handbook Of Forensic Genetics: Biodiversity And Heredity In Civil And Criminal Investigation Apr 29 2020 The use of genetics for the resolution of legal conflicts has recently been gaining a higher profile, largely as a result of scientific and technological advancements and the substantial broadening of applications. The theoretical framework underlying forensic genetics is the same irrespective of the materials and technology involved, however a great divide still exists in the manner and processes related to human and non-human analyses.This advanced handbook intends to overcome the historical barriers between the scientific fields of legal medicine, biodiversity and conservation, and food analysis by presenting a unifying, global perspective on the implications of genetic analyses on forensic affairs. This global perspective is presented in three parts: modes of inheritance and reproduction and taxonomic implications; current technological approaches and future perspectives; and a comprehensive systematization of the types of applications and organisms. Finally, a critical revision of the current investigative/expert systems and future perspectives is undertaken.This book provides a collection of international research, thereby constituting a reference platform for the forensic community and an advanced textbook for graduate students. It encompasses the theoretical bases of the field, and presents in the context of both perspectives of forensic action — probative and investigative — a comprehensive coverage of the current applications and technological state of the art.

Understanding Genetics May 23 2022 The purpose of this manual is to provide an educational genetics resource for individuals, families, and health professionals in the New York - Mid-Atlantic region and increase awareness of specialty care in genetics. The manual begins with a basic introduction to genetics concepts, followed by a description of the different types and

applications of genetic tests. It also provides information about diagnosis of genetic disease, family history, newborn screening, and genetic counseling. Resources are included to assist in patient care, patient and professional education, and identification of specialty genetics services within the New York - Mid-Atlantic region. At the end of each section, a list of references is provided for additional information. Appendices can be copied for reference and offered to patients. These take-home resources are critical to helping both providers and patients understand some of the basic concepts and applications of genetics and genomics.

Integrative analysis of single-cell and/or bulk multi-omics sequencing data Nov 04 2020

Next Generation Sequencing Technologies in Medical Genetics Jun 23 2022 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 diagnoses 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.

Multicenter Validation of Cancer Gene Panel-based Next-generation Sequencing for Translational Research and Molecular Diagnostics Apr 21 2022 Abstract: The simultaneous detection of multiple somatic mutations in the context of molecular diagnostics of cancer is frequently performed by means of amplicon-based targeted next-generation sequencing (NGS). However, only few studies are available comparing multicenter testing of different NGS platforms and gene panels. Therefore, seven partner sites of the German Cancer Consortium (DKTK) performed a multicenter interlaboratory trial for targeted NGS using the same formalin-fixed, paraffin-embedded (FFPE) specimen of molecularly pre-characterized tumors (n = 15; each n = 5 cases of Breast, Lung, and Colon carcinoma) and a colorectal cancer cell line DNA dilution series. Detailed information regarding pre-characterized mutations was not disclosed to the partners. Commercially available and custom-designed cancer gene panels

were used for library preparation and subsequent sequencing on several devices of two NGS different platforms. For every case, centrally extracted DNA and FFPE tissue sections for local processing were delivered to each partner site to be sequenced with the commercial gene panel and local bioinformatics. For cancer-specific panel-based sequencing, only centrally extracted DNA was analyzed at seven sequencing sites. Subsequently, local data were compiled and bioinformatics was performed centrally. We were able to demonstrate that all pre-characterized mutations were re-identified correctly, irrespective of NGS platform or gene panel used. However, locally processed FFPE tissue sections disclosed that the DNA extraction method can affect the detection of mutations with a trend in favor of magnetic bead-based DNA extraction methods. In conclusion, targeted NGS is a very robust method for simultaneous detection of various mutations in FFPE tissue specimens if certain pre-analytical conditions are carefully considered

Bacterial Pathogenesis May 11 2021 This highly anticipated update of the acclaimed textbook draws on the latest research to give students the knowledge and tools to explore the mechanisms by which bacterial pathogens cause infections in humans and animals. Written in an approachable and engaging style, the book uses illustrative examples and thought-provoking exercises to inspire students with the potential excitement and fun of scientific discovery. Completely revised and updated, and for the first time in stunning full-color, Bacterial Pathogenesis: A Molecular Approach, Fourth Edition, builds on the core principles and foundations of its predecessors while expanding into new concepts, key findings, and cutting-edge research, including new developments in the areas of the microbiome and CRISPR as well as the growing challenges of antimicrobial resistance. All-new detailed illustrations help students clearly understand important concepts and mechanisms of the complex interplay between bacterial pathogens and their hosts. Study questions at the end of each chapter challenge students to delve more deeply into the topics covered, and hone their skills in reading, interpreting, and analyzing data, as well as devising their own experiments. A detailed glossary defines and expands on key terms highlighted throughout the book. Written for advanced undergraduate, graduate, and professional students in microbiology, bacteriology, and pathogenesis, this text is a must-have for anyone looking for a greater

understanding of virulence mechanisms across the breadth of bacterial pathogens.

AACR 2019 Proceedings: Abstracts 1-2748 Jul 13 2021 American Association for Cancer Research 2019 Proceedings: Abstracts 1-2748 - Part A

Prognostic Epigenetics Jul 01 2020 This volume provides comprehensive information on how mapping an individual's epigenome can be medically relevant and holds the potential to improve preventive medicine and precision therapeutics at an early-stage (prior to disease onset). In order to advance clinical adoption of the recently developed epigenetic approaches, it is necessary for translational scientists, clinicians, and students to gain a better understanding about epigenetic mechanisms that are associated with a particular disorder; and to be able to effectively identify biomarkers that can be applied in drug development and for better diagnosis and prognosis of diseases. Prognostic Epigenetics is the most-inclusive volume to-date specifically dedicated to epigenetic markers that have been developed for prognosis of diseases, recent advances in this field, the clinical implementation of this research, and the future outlook. Compiles all known information on prognostic epigenetics and its role in preventive medicine and drug discovery Covers the basic functionality of epigenetic mechanisms involved in early disease prognosis and diagnosis, and provides tools for the identification and development of these biomarkers for a wide range of diseases Enables clinicians, researchers, and pharmacologists to improve preventive medicine and precision therapeutics throughout a person's lifetime Features chapter contributions from leading international researchers

Genetics and Genomics in Medicine Dec 30 2022 The second edition of this textbook written for undergraduate students, graduate students and medical researchers, Genetics and Genomics in Medicine explains the science behind the uses of genetics and genomics in medicine today, and how it is being applied. Maintaining the features that made the first edition so popular, this second edition has been thoroughly updated in line with the latest developments in the field. DNA technologies are explained, with emphasis on the modern techniques that are revolutionizing the use of genetic information in medicine and indicating the role of genetics in common diseases. Epigenetics and non-coding RNA are covered in-depth as are genetic

approaches to treatment and prevention, including pharmacogenomics, genetic testing, and personalized medicine. A dedicated chapter charts the latest insights into the molecular basis of cancers, cancer genomics and novel approaches to cancer detection. Coverage of genetic testing at the level of genes, chromosomes and genomes has been significantly expanded and updated. Extra prominence has been given to additional genomic analyses, ethical aspects, and novel therapeutic approaches. Various case studies illustrate selected clinical applications. Key Features Comprehensive and integrated account of how genetics and genomics affect the entire spectrum of human health and disease Exquisite artwork illuminates the key concepts and mechanisms Summary points at the end of each chapter help to consolidate learning For each chapter, an abundance of further reading to help provide the reader with direction for further study Inclusive online question bank to test understanding Standard boxes summarizing certain key principles in genetics Clinical boxes summarizing selected case studies, pathogenesis mechanisms or novel therapies for selected diseases This book is equally suited for newcomers to the field as well as for engineers and scientists that have basic knowledge in this field but are interested in obtaining more information about specific future applications..

Protein-targeted DNA Scission Aug 14 2021

Early Phase Cancer Immunotherapy Aug 02 2020 This volume, a state-of-the-art review of early phase clinical trials for cancer immunotherapy, discusses biomarker selection, combinatorial strategies and their safety or toxicity, determination of Phase 2 dosing, endpoints in the setting of radiographic pseudoprogression, histology selection, and novel immunotherapeutics as they relate to early phase cancer immunotherapy.

Clinical Genomics Dec 18 2021 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

Pediatric Board Study Guide Oct 28 2022 Building upon the highly successful 1st edition, this book is a comprehensive review designed to prepare pediatric residents, fellows, and pediatricians for the General Pediatrics Certifying Examination, and for the American Board of Pediatrics Maintenance of Certification. Pediatric Board Study Guide: A Last Minute Review, 2nd edition, covers all aspects of pediatric medicine; each chapter has been updated according to the most recent content specifications provided by the ABP. The 2nd edition provides more illustrations, diagrams, radiology images, and clinical case scenarios to further assist readers in reviewing pediatric subspecialties. New chapter topics include nutrition, sports medicine, patient safety, quality improvement, ethics, and pharmacology. Finally, the book closes with a “Last Minute Review” of high-yield cases arranged in the same sequence as the chapters, providing readers with a concise study guide of critical cases and conditions. Pediatric residents and fellows preparing for the board examination, pediatricians, and pediatric subspecialists preparing for certification maintenance will find Pediatric Board Study Guide: A Last Minute Review, 2nd edition easy to use and comprehensive, making it the ideal resource and study tool.

Precision Cancer Medicine May 03 2023 Genomic sequencing technologies have augmented the classification of cancer beyond tissue of origin and towards a molecular taxonomy of cancer. This has created opportunities to guide treatment decisions for individual patients with cancer based on their cancer’s unique molecular characteristics, also known as precision cancer medicine. The purpose of this text will be to describe the contribution and need for multiple disciplines working together to deliver precision cancer medicine. This entails a multi-disciplinary approach across fields including molecular pathology, computational biology, clinical oncology, cancer biology, drug development, genetics, immunology, and bioethics. Thus, we have outlined a

current text on each of these fields as they work together to overcome various challenges and create opportunities to deliver precision cancer medicine. As trainees and junior faculty enter their respective fields, this text will provide a framework for understanding the role and responsibility for each specialist to contribute to this team science approach.

Problem Solving through Precision Oncology Jan 07 2021 Winner of the BMA Oncology Book of the Year Award. This practical learning and reference handbook provides an overview of the latest progress in the developing field of precision oncology, plus a ground breaking collection of case studies ("Problems") showing precision oncology in practice. The book includes a clear, readable summary of developments, alongside real-life case studies, providing a valuable update for all involved in the oncology community. The editors lead research and clinical teams at four UK centres of excellence in the field.

Targeted Therapies in Cancer: Oct 04 2020 Billions of dollars are spent every year on research into targeted therapies for cancer. That’s why it’s more than ever crucial for the thousands of scientists working in the field to keep right up to date with the cutting edge. This fascinating collection of material goes a long way to helping them do so, featuring as it does contributions to a crucial international meeting in Italy. The meeting provided a forum for scientists and clinicians working in cancer drug discovery and therapy to share their opinions and experiences. The text here offers readers an overview of diverse approaches, ranging from drug discovery to cellular therapy. Overall, the book addresses the key question of whether ultimately targeted therapy in cancer will be a myth or a reality.

Modern Techniques in Cytopathology Feb 26 2020 "The purpose of this book is to describe, illustrate, and review many of the most recent developments regarding modern techniques employed in cytopathology. It is intended for all cytologists, including cytopathologists, cytotechnologists, cytology lab assistants, trainees, research scientists, and anyone who is interested in the field of cytopathology"--

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- Cytogenomics
- An Integrative DNA Sequencing Panel To Assess Mismatch Repair Deficiency
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- Prognostic Epigenetics
- Protein Modifications In Epigenetic Dysfunctional Diseases Mechanisms And Potential Therapeutic Strategies
- Handbook Of Forensic Genetics Biodiversity And Heredity In Civil And Criminal Investigation
- Next Generation Sequencing Based Diagnostic Approaches In Clinical Oncology
- Modern Techniques In Cytopathology
- Evolution Of Translational Omics
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