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Metodi per l'analisi genetica di malattie rare

Dosaggio di enzimi nelle analisi di laboratorio

ESTRATTA

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Review BMJ. 2021 Jul 26;374:n1648. doi: 10.1136/bmj.n1648.

Long covid-mechanisms, risk factors, and management

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PMID: 34312178 DOI: 10.1136/bmj.n1648

Erratum in

Correction.
[No authors listed]
BMJ. 2021 Aug 3;374:n1944. doi: 10.1136/bmj.n1944.
PMID: 34344715 No abstract available.

Abstract

Since its emergence in Wuhan, China, covid-19 has spread and had a profound effect on the lives and health of people around the globe. As of 4 July 2021, more than 183 million confirmed cases of covid-19 had been recorded worldwide, and 3.97 million deaths. Recent evidence has shown that a range of persistent symptoms can remain long after the acute SARS-CoV-2 infection, and this condition is now coined long covid by recognized research institutes. Studies have shown that long covid can affect the whole spectrum of people with covid-19, from those with very mild acute disease to the most severe forms. Like acute covid-19, long covid can involve multiple organs and can affect many systems including, but not limited to, the respiratory, cardiovascular, neurological, gastrointestinal, and musculoskeletal systems. The symptoms of long covid include fatigue, dyspnea, cardiac abnormalities, cognitive impairment, sleep disturbances, symptoms of post-traumatic stress disorder, muscle pain, concentration problems, and headache. This review summarizes studies of the long term effects of covid-19 in hospitalized and non-hospitalized patients and describes the persistent symptoms they endure. Risk factors for acute covid-19 and long covid and possible therapeutic options are also discussed.

Supplementary concepts

post-acute COVID-19 syndrome

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Test genetici predittivi nella gestione del paziente oncologico

La spettrometria di massa nelle analisi di laboratorio

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2

Review Life Sci. 2018 Nov 1;212:194-202. doi: 10.1016/j.lfs.2018.09.035. Epub 2018 Sep 19.

Proteomics, metabolomics and metagenomics for type 2 diabetes and its complications

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PMID: 30243649 DOI: 10.1016/j.lfs.2018.09.035

Abstract

Type 2 diabetes mellitus (T2DM) is one of the most common diseases of endocrine and metabolic disorders, whose mechanism is still largely unknown. Fortunately, various "omics" tools have been employed to better understand the progression pathologies of T2DM and its complications. More specifically, proteomics, metabolomics and metagenomics have played crucial roles in advancing deeper understanding of the physiological processes and regulatory mechanisms of T2DM, such as regulation of signaling pathways perturbed by glucose levels, intestinal microorganism, and inflammation and so on. By analyzing the dynamic change and modification of proteins, proteomics has become an important tool in biology and medicine. Metabolomic analysis can amplify and quantify metabolites in living organisms to reveal the relative relationship between metabolites and physiological and pathological changes. There are also increasing evidences that the human microbiome, specifically the gastrointestinal microbiome have a potential role in the etiology and pathological outcomes of T2DM and its complications. This article summarized and discussed the recent applications of these "omics" tools in finding biomarkers for T2DM and its complications. We also reviewed employing multiple "omics" to further advance our understanding of this pathology. This review will benefit deeper understanding in new therapeutic and/or diagnostic biological target for the discovery of T2DM and its complications.

Keywords: Biomarkers; Metabolomics; Metagenomics; Proteomics; T2DM.

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Other Literature Sources
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Genetic Alliance
MedlinePlus Consumer Health Information
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Applicazioni in genetica medica della tecnologia NGS (next generation sequencing)

Marker plasmatici in medicina di laboratorio

NON ESTRATTA

3

Review Nat Rev Clin Oncol. 2018 Jun;15(6):353-365. doi: 10.1038/s41571-018-0002-6.

The emerging clinical relevance of genomics in cancer medicine

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PMID: 29599476 PMCID: PMC6658089 DOI: 10.1038/s41571-018-0002-6
Free PMC article

Abstract

The combination of next-generation sequencing and advanced computational data analysis approaches has revolutionized our understanding of the genomic underpinnings of cancer development and progression. The coincident development of targeted small molecule and antibody-based therapies that target a cancer's genomic dependencies has fuelled the transition of genomic assays into clinical use in patients with cancer. Beyond the identification of individual targetable alterations, genomic methods can gauge mutational load, which might predict a therapeutic response to immune-checkpoint inhibitors or identify cancer-specific proteins that inform the design of personalized anticancer vaccines. Emerging clinical applications of cancer genomics include monitoring treatment responses and characterizing mechanisms of resistance. The increasing relevance of genomics to clinical cancer care also highlights several considerable challenges, including the need to promote equal access to genomic testing.

Figures

Fig. 1. Clinical utility of genomic assays...

Fig. 2. Clinical trial designs invoking cancer...

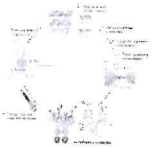

Fig. 3. NGS-based neoantigen discovery.
Neoantigen discovery...

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Test di primo e secondo livello in epoca neonatale

Metodi fluorimetrici nelle analisi di laboratorio

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4

Review Mass Spectrom Rev. 2012 Sep-Oct;31(5):583-605. doi: 10.1002/mas.20356. Epub 2012 Mar 15.

Cancer proteomics

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PMID: 22422534 DOI: 10.1002/mas.20356

Abstract

Cancer presents high mortality and morbidity globally, largely due to its complex and heterogenous nature, and lack of biomarkers for early diagnosis. A proteomics study of cancer aims to identify and characterize functional proteins that drive the transformation of malignancy, and to discover biomarkers to detect early-stage cancer, predict prognosis, determine therapy efficacy, identify novel drug targets, and ultimately develop personalized medicine. The various sources of human samples such as cell lines, tissues, and plasma/serum are probed by a plethora of proteomics tools to discover novel biomarkers and elucidate mechanisms of tumorigenesis. Innovative proteomics technologies and strategies have been designed for protein identification, quantitation, fractionation, and enrichment to delve deeper into the oncoproteome. In addition, there is the need for high-throughput methods for biomarker validation, and integration of the various platforms of oncoproteome data to fully comprehend cancer biology.

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Diagnosi prenatale genetica

Metodi di separazione di analiti da miscele complesse

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5

Review Nat Rev Mol Cell Biol. 2019 Jun;20(6):353-367. doi: 10.1038/s41580-019-0108-4.

Identification of bioactive metabolites using activity metabolomics

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PMID: 30814649 PMCID: PMC6613555 DOI: 10.1038/s41580-019-0108-4
Free PMC article

Abstract

The metabolome, the collection of small-molecule chemical entities involved in metabolism, has traditionally been studied with the aim of identifying biomarkers in the diagnosis and prediction of disease. However, the value of metabolome analysis (metabolomics) has been redefined from a simple biomarker identification tool to a technology for the discovery of active drivers of biological processes. It is now clear that the metabolome affects cellular physiology through modulation of other 'omics' levels, including the genome, epigenome, transcriptome and proteome. In this Review, we focus on recent progress in using metabolomics to understand how the metabolome influences other omics and, by extension, to reveal the active role of metabolites in physiology and disease. This concept of utilizing metabolomics to perform activity screens to identify biologically active metabolites - which we term activity metabolomics - is already having a broad impact on biology.

Figures

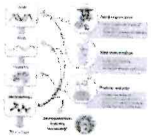

Figure 1. Metabolites – active modulators of...

Figure 2: Examples of macromolecule modification by...

Figure 3. Mechanisms for non-covalent modification of...

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Valutazione delle varianti di sequenza nelle malattie genetiche

Analisi di primo livello nello screening di malattie metaboliche rare

ESTRATTA

(6)

Review Annu Rev Biomed Eng. 2007;9:289-320.
doi: 10.1146/annurev.bioeng.9.060906.152037.

SNP genotyping: technologies and biomedical applications

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PMID: 17391067 DOI: 10.1146/annurev.bioeng.9.060906.152037

Abstract

Single nucleotide polymorphisms (SNPs) are the most frequently occurring genetic variation in the human genome, with the total number of SNPs reported in public SNP databases currently exceeding 9 million. SNPs are important markers in many studies that link sequence variations to phenotypic changes; such studies are expected to advance the understanding of human physiology and elucidate the molecular bases of diseases. For this reason, over the past several years a great deal of effort has been devoted to developing accurate, rapid, and cost-effective technologies for SNP analysis, yielding a large number of distinct approaches. This article presents a review of SNP genotyping techniques and examines their principles of genotype determination in terms of allele differentiation strategies and detection methods. Further, several current biomedical applications of SNP genotyping are discussed.

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NCI CPTAC Assay Portal

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Metodi di analisi di espressione genica

Interazione antigene-anticorpo nella diagnostica di laboratorio

ESTRATTA

Review Neurocrit Care. 2021 Jun;34(3):1062-1071. doi: 10.1007/s12028-020-01049-4.

Neurological Involvement in COVID-19 and Potential Mechanisms: A Review

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Affiliations

PMID: 32661794 PMCID: PMC7358290 DOI: 10.1007/s12028-020-01049-4
Free PMC article

Abstract

As the current understanding of COVID-19 continues to evolve, a synthesis of the literature on the neurological impact of this novel virus may help inform clinical management and highlight potentially important avenues of investigation. Additionally, understanding the potential mechanisms of neurologic injury may guide efforts to better detect and ameliorate these complications. In this review, we synthesize a range of clinical observations and initial case series describing potential neurologic manifestations of COVID-19 and place these observations in the context of coronavirus neuro-pathophysiology as it may relate to SARS-CoV-2 infection. Reported nervous system manifestations range from anosmia and ageusia, to cerebral hemorrhage and infarction. While the volume of COVID-19-related case studies continues to grow, previous work examining related viruses suggests potential mechanisms through which the novel coronavirus may impact the CNS and result in neurological complications. Namely, animal studies examining the SARS-CoV have implicated the angiotensin-converting-enzyme-2 receptor as a mediator of coronavirus-related neuronal damage and have shown that SARS-CoV can infect cerebrovascular endothelium and brain parenchyma, the latter predominantly in the medial temporal lobe, resulting in apoptosis and necrosis. Human postmortem brain studies indicate that human coronavirus variants and SARS-CoV can infect neurons and glia, implying SARS-CoV-2 may have similar neurovirulence. Additionally, studies have demonstrated an increase in cytokine serum levels as a result of SARS-CoV infection, consistent with the notion that cytokine overproduction and toxicity may be a relevant potential mechanism of neurologic injury, paralleling a known pathway of pulmonary injury. We also discuss evidence that suggests that SARS-CoV-2 may be a vasculotropic and neurotropic virus. Early reports suggest COVID-19 may be associated with severe neurologic complications, and several plausible mechanisms exist to account for these observations. A heightened awareness of the potential for neurologic involvement and further investigation into the relevant pathophysiology will be necessary to understand and ultimately mitigate SARS-CoV-2-associated neurologic injury.

Keywords: Cerebrovascular stroke; Coronavirus; Inflammation; Neurology.

Figures