

Busta n. 1

1. Test di vitalità e di citotossicità delle colture cellulari.
2. Metodi di estrazione degli acidi nucleici.

Conoscenze informatiche: quale programma del pacchetto office permette di fare un'analisi statistica e quali altri programmi conosci al di fuori del pacchetto office.

Conoscenza della lingua inglese:

Abstract: Cardiorenal syndrome type 5 (CRS-5) includes conditions where there is a simultaneous involvement of the heart and kidney from a systemic disorder. This is a bilateral organ cross talk. Fabry's disease (FD) is a devastating progressive inborn error of metabolism with lysosomal glycosphingolipid deposition in variety of cell types, capillary endothelial cells, renal, cardiac and nerve cells. Basic effect is absent or deficient activity of lysosomal exoglycosidase α -galactosidase A. Renal involvement consists of proteinuria, isosthenuria, altered tubular function, presenting in second or third decade leading to azotemia and end-stage renal disease in third to fifth decade mainly due to irreversible changes to glomerular, tubular and vascular structures, especially highlighted by podocytes foot process effacement.

G

G.P.

G.P. M.P.

Busta n. 2

- 1. Allestimento di una coltura primaria.**
- 2. Analisi della espressione genica mediante RT-PCR.**

Conoscenze informatiche: cos'è un browser web e quale è il suo ruolo nel navigare su Internet?

Conoscenza della lingua inglese:

Combined heart and kidney dysfunction is common. A disorder of one of these two organs often leads to dysfunction or injury to the other. This is the pathophysiological basis for the clinical entity defined cardiorenal syndrome (CRS).

Generally defined as a condition characterized by the initiation and/or progression of renal insufficiency secondary to heart failure, the term CRS should also be used to describe conditions of renal dysfunction leading to heart dysfunction (renocardiac syndrome). The common view is that a relatively normal kidney is dysfunctional because of diseased heart. This concept, however, has been challenged, and the most recent definitions include a variety of conditions, either acute or chronic, where the primary failing organ can be either the heart or the kidney or both. Such advances in the definition and classification of CRS enabled the characterization of the complex organ cross talk and have proposed specific prevention strategies and therapeutic interventions to attenuate end organ injury.

J. G. e. SP 9/10

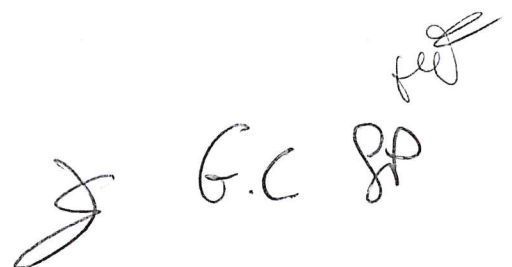
Busta n. 3

- 1. Confronto tra la tecnica di qPCR e ddPCR nello studio dell'espressione genica degli acidi nucleici.**
- 2. Descrizione della tecnica di Immunoprecipitazione delle proteine.**

Conoscenze informatiche: quale pacchetto office utilizzeresti per la preparazione di un poster o di una comunicazione orale?

Conoscenza della lingua inglese:

Early neural damage primarily involves small nerve fibers of the peripheral somatic and autonomic nerve systems] with onset of related symptoms generally occurring at an earlier age in boys than in girls. Pain is experienced by 60-80 % of classically affected boys and girls and is one of the earliest symptoms of FD. Two types of pain have been described: episodic crises ("Fabry's crises") characterized by agonizing burning pain originating in the extremities and radiating inwards to the limbs and other parts of the body, and chronic pain characterized by burning and tingling paraesthesias. Fabry's crises may be precipitated by fever, exercise, fatigue, stress and rapid changes in temperature. Pain may wane in adulthood, and it is important to search for a medical history of acroparesthesia in childhood during the first examination of a newly diagnosed adult patient.

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Busta n. 4

1. Allestimento di un campione da analizzare al microscopio a fluorescenza.
2. Controllo di qualità degli acidi nucleici.

Conoscenze informatiche: con quale carattere iniziano le formule utilizzate da Microsoft Excel?

Conoscenza della lingua inglese:

Light microscopy: the observation of biopsies with light microscopy does not usually contribute a great deal to diagnosis, but lipid staining of kidney biopsies can reveal storage cells within glomeruli, and when electron microscopy (EM) is not being done or not available, semi-thin sections stained with toluidine blue or Masson's trichrome can allow diagnosis. However, given the number of false negatives and the non-specificity of the results, this invasive procedure should not be used for diagnostic purpose. Electron microscopy: Ultrastructural studies of endomyocardial and kidney biopsies can reveal lysosomal storage in cardiomyocytes or in a variety of kidney cellular types, respectively. The ultrastructural appearance of the inclusions is of whorled layers of alternating dense and pale material "zebra bodies" or myelin figures). However, due to the invasive nature of the procedure and the availability of reliable biochemical or molecular methods, these procedures should be considered only in the rare instances where there is residual a-galactosidase A activity in males or doubts on the causality of a DNA sequence change in females. Skin biopsy observed by EM may be a useful additional diagnostic test when carefully interpreted by an expert pathologist. However, acquired metabolic disorders, such as the one induced by chloroquine therapy, may result in storage of ultrastructurally similar inclusions in many of the same cells as FD, leading to erroneous interpretation in addition, skin biopsies are often normal in heterozygous.

G.C. 80
G.C. 80

Busta n. 5

1. Tecniche di silenziamento genico.
2. Descrizione della tecnica di microscopia confocale e a fluorescenza.



Conoscenze informatiche: qual è la funzione principale della memoria RAM di un computer?

Conoscenza della lingua inglese:

Biomarkers

There is currently no proper plasma or urinary biomarker for FD. Mildly elevated plasma chitotriosidase levels have been reported in male patients, but not in heterozygous females. Globotriaosylsphingosine or lyso-Gb3 has been reported to be elevated in FD patients. This analyte is elevated in the plasma of hemizygous males and to a lesser extent in that of adult females with classical FD and lyso-Gb3 appears interesting to monitor enzyme replacement therapy. Lyso-Gb3 was shown to be an independent risk factor for the development of cerebrovascular white matter lesions in male patients with FD, while, in females, plasma lyso-Gb3 concentration correlated with overall disease severity. Lyso-Gb3 could be a potential biomarker since plasma lyso-Gb3 level in Fabry patients who had received ERT was shown to be elevated at baseline and to fall more dramatically on ERT than that of Gb3. Urinary lyso-Gb3 may also prove a potential biomarker. Lyso-Gb3 may have a role in glomerular injury in FD by promoting the release of secondary mediators of glomerular injury (transforming growth factor-beta 1 (TGF- β I) and the macrophage inhibitory factor receptor CD74) common to diabetic nephropathy.

Sphingosine-1-phosphate (S1P) was recently identified as a biologically active growth-promoting factor involved in cardiovascular remodeling in both males and females with FD. Male patients had significantly higher plasma S1P levels compared with healthy controls. Moreover, there was a strong correlation between plasma S1P levels and left ventricular mass (LVM) index and increased common carotid artery intimal-media thickening (IMT) in patients with FD. Sphingosine-1-phosphate has been shown to induce in vitro vascular smooth muscle cells proliferation by a variety of signal transduction pathways.

 G.C.  M.F.

Busta n. 6

1. Significato della curva di melting e cosa descrive l'efficienza di reazione in una reazione di PC
2. Metodi di processamento di campioni biologici: estrazione e purificazione delle proteine e degli esosomi.

Conoscenze informatiche: quale formato viene usato per salvare i files in Word?

Conoscenza della lingua inglese:

Genotyping

In female heterozygotes, α -galactosidase activity may be within the normal range, and therefore, the definitive diagnostic confirmation should be made by genetic analysis in suspected cases. The publication of the complementary (cDNA) and genomic DNA sequences of the GLA gene (Genbank XI4448) has paved the way toward understanding of the molecular basis of FD. Direct molecular analysis is easy because of the small size of the gene and allows the precise characterization of the mutation of the GLA gene. Denaturing high-performance liquid chromatography (DHPLC) has been shown to be useful as a screening method. Since direct sequencing limited to exons may miss deletions, the use of multiplex ligation-dependent probe amplification (MLPA) has been recommended in cases where a decreased enzyme activity is not associated with the identification of a pathogenic point mutation.


G.C. 

Busta n. 7

1. Descrivi le caratteristiche che deve avere una buona coppia di oligonucleotidi.
2. Metodi di quantificazione delle proteine

Conoscenze informatiche: che cos'è un documento PDF

Conoscenza della lingua inglese:

Biochemical diagnosis

Enzymatic assay: the demonstration of a deficiency activity of α -galactosidase activity in plasma or leukocytes is the reference laboratory method which should systematically be used to confirm the clinical diagnosis of FD in males in whom the result will be conclusive. Plasma assay may occasionally lead to false diagnosis and should be confirmed by a leukocyte assay. In contrast, affected girls and adult females may have their enzyme activity falling within the normal range. Therefore, all females should have their status determined by genotyping (analysis of the GLA gene mutation). A fluorimetric method that uses filter paper cards containing dried blood spots instead of the leukocyte pellet as the enzyme source was recently introduced for enzymatic diagnosis, allowing storage of the samples for up to 6 months due to stability of the enzyme. Globotriaosylceramide measurement: Plasma Gb3 has also been proposed and used in the biochemical diagnosis of FD, but this method is time-consuming, and in females, plasma Gb3 levels are generally lower than in males and usually in the normal range. Urinary Gb3 is a more reliable marker allowing diagnosis in the majority of both male and female patients. However, urinary Gb3 is not elevated in some patients with late-onset variants and/or particular mutations in the GLA gene.


G.C. GP MB