Abstract

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From Genetic Screening to Patient Management

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Background And Aims

Genetic screenings are increasingly present in the clinical field, not only for the identification of diseases, but especially as support in a more personalized therapeutic choice. Pharmacogenetic studies aim to identify "polymorphisms" capable of influencing an individual's response to a specific drug, making them crucial in therapeutic decision-making. Identifying polymorphisms that affect genes encoding enzymes, receptors, and other proteins involved in pharmacological response, as well as understanding their associated phenotypic consequences, will facilitate the selection of the most appropriate therapeutic protocol for each patient and greatly reduce adverse reactions, toxicity, or therapeutic failure.

Materials And Methods

The proposed approach involves implementing diagnostic technologies based on Next Generation Sequencing (NGS). A multidisciplinary pathway is proposed for studying genetic polymorphisms, aiming to evaluate the genes involved in clopidogrel metabolism to prevent rehospitalization due to coronary stent thrombosis and optimize pharmacological therapy in patients taking clopidogrel. In patients with acute coronary syndrome, current guidelines recommend 12 months of DAPT (dual antiplatelet therapy) with aspirin and an oral P2Y12 receptor inhibitor (preferably ticagrelor or prasugrel over clopidogrel) [1]. An alternative approach has recently been proposed involving Escalation and De-Escalation of antiplatelet therapy after ACS or PCI [2]. The guided approach involves the use of genetic tests or platelet function testing to identify carriers of mutated alleles of CYP2C19 (PM-Poor Metabolizer, two mutated alleles with loss of function, or IM Intermediate Metabolizer, one non-functioning allele).

Results

The implementation of this project would lead to:

Technological upgrades aimed at ensuring the application of the most advanced technologies currently available to support the correct prescription of pharmacological therapies.

Streamlining diagnostic pathways, allowing for case discussions and tailoring the examination to specific clinical needs, modifying the diagnostic and therapeutic pathway if necessary.

Increased availability of diagnostic tests for the benefit of users.

Clinical management of results within the same institution, where both clinical and laboratory expertise are guaranteed, within a precise diagnostic pathway that is completed with full patient care.

Discussion

When pharmacogenetics is introduced into clinical practice, we will witness a revolution in the way drugs are prescribed, resulting in several advantages, including improved therapeutic intervention efficacy, more accurate dosage selection, and a reduction in adverse reactions.

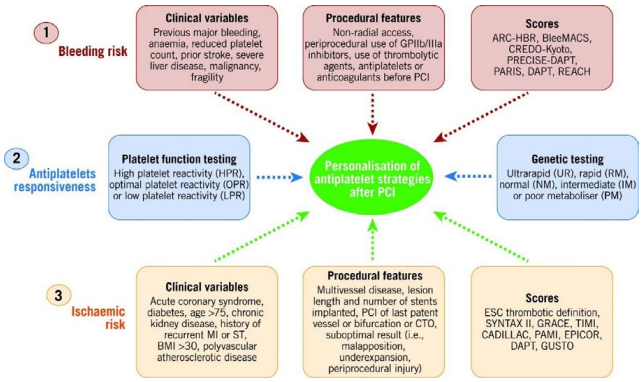

Figure 1. Risk assessment to guide antiplatelet therapy among percutaneous coronary disease patients

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