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Molecular diagnosis for Hemophilia in Argentina: Updated and cost-effective scheme including inverse shifting-PCR, CSGE-screening and bioinformatics

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Hemophilia is an X-linked coagulopathy that affects one in 5,000 human males worldwide. Hemophilia is classified into hemophilia A (HA) due to deleterious mutations in the coagulation factor VIII gene (*F8*) (Xq28) and hemophilia B (HB) associated with factor IX (*F9*) (Xq27.1).

Our Laboratory of Molecular Genetics in Hemophilia, pioneer in Latin-America, achieves basic research. (e.g., addressing the mechanisms involved in genotype-phenotype associations in Hemophilia) and molecular diagnosis of affected families using an optimal scheme for *F8*-and *F9*-genotyping including development of new practical approaches.

Due to the characteristics of *F8* and *F9* (26 exons/186kb and 8 exons/33kb, respectively) and the heterogeneous spectrum of molecular defects, Hemophilia still challenges mutation detection in many laboratories, particularly from developing countries.

Our laboratory diagnostic algorithm covers the complete analysis of hemophilia genes including inverse shifting-PCR (IS-PCR), an original approach for genotyping inversions affecting intron 22 (type-1/type-2 patterns, and associated duplications and deletions) and intron 1 of *F8* that account for half of severe-HA patients worldwide; standard PCR-amplification of *F8* (or *F9*) for detection of large deletions and its specific analysis by long distance-PCR; small mutation screening by CSGE (conformation sensitive gel electrophoresis); conventional DNA sequencing of selected regions, followed by analysis of patient's genotype/phenotype correspondence by bioinformatics tools.

Moving forward, our goals still focus on investigating to provide, in a cost-effective manner, all the relevant genetic information to help physicians to adjust case-specific therapies and follow-up protocols and, ultimately, to improve the medical care of patients with hemophilia and their families.

Biography

Liliana Rossetti has completed her PhD in Molecular Biology from the University of Buenos Aires, Argentina, in 2002, and postdoctoral studies from the National Academy of Medicine of Buenos Aires. She is actually a Member of the Research Career of CONICET (National Scientific and Technical Research Council), Associate class, at the Genetics Department, at the National Academy of Medicine. She has published 15 papers in reputed journals. Her research is focused on understanding the genetics mechanisms involved in the origin of Hemophilia, development of new diagnosis approaches, and their importance in genetic counseling.

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