

SCREENGEN – AN EURO-REGIONAL CROSS-BORDER PROJECT – DEDICATED TO THE BIO-MOLECULAR DIAGNOSIS OF HAEMOPHILIA ACHIEVEMENTS AND PERSPECTIVES

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The management of haemophilia is an ever-challenging health-problem in the world. Along with its condition of “rare disorder” haemophilia presents also the feature of “orphan disease” because of the high costs of its therapy, responsible for the unavailability and/ or unaffordability of factor concentrates, the pivotal treatment of the disease.

In this situation, prevention remains a key step in the medical approach of haemophilia.

Genetic counselling and family planning based on prenatal diagnosis and on the detection of carriers are of utmost importance. The prediction of the occurrence of inhibitors, a main posttherapeutical complication, is an additional meaningful goal.

The development of biomolecular assay is a key issue serving these objectives. That is the goal of our cross-border SCREENGEN project. Our preliminary results on 166 samples (159 patients and 7 potential carriers) investigated by PCR-based STRs polymorphism analysis are presented. They suggest the importance of bio-molecular investigations for the prediction of phenotypic severity, risk of inhibitors occurrence and genetic counselling.