Relationship between coagulometric tests, clinical symptoms and the PCR test on fVIII haemophilia carriers

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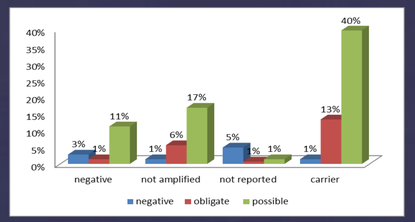
INTRODUCTION AND OBJECTIVES

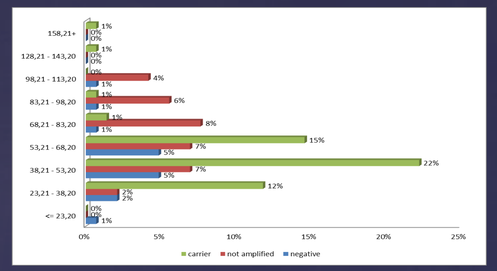
For the first time in 2006, haemophilia carriers were detected using coagulometric tests. Nine years later in our country, pilot tests were done to PCR carriers, seeking to relate the results of aPTT and fVIII with those obtained in the test of molecular biology, as well as detecting asymptomatic carriers. Patients´ symptoms were checked and family trees were built; these results allowed us to guide families with haemophilia.

METHODS

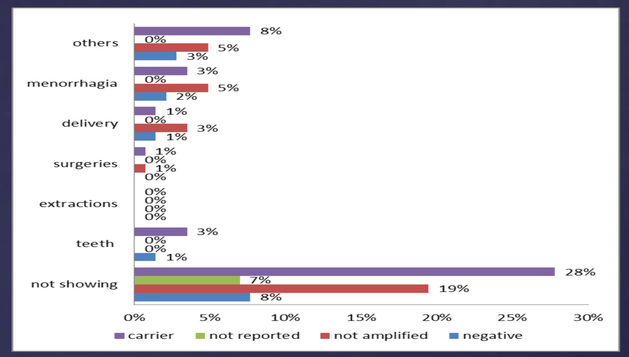
144 plasma samples were obtained with sodium citrate from carriers who were classified by age. An aPTT (VR = until 36 sec) and the determination of the VIII factor (VR = 50- 150%) was done on the semiautomatic team Stago-Start 4. The long-PCR test of intron 1 and 22 inversion (most common mutations) was performed. DNA was extracted with the Promega Wizard Genomic DNA kit protocol and the beta globin was used as a positive control of the samples of patients with EDTA



76 patients (53%) presented positive mutation of intron 22; according to the studied literature this mutation is the most common in this type of haemophilia A (severe) 40-45%



26 patients (19%) show levels of factor VIII higher than 53% and are positive carriers of the intron 22.



The result seen in relation to the carrier bleeding reflects that most asymptomatic carriers are 40 (27.8%), 24 patients (16.7%) have other kind of bleeding (included in the questionnaire) such as bruising, injuries that take time to clot, etc

RESULTS

76 patients (53%) were positive carriers of the 22 mutation. No positive results were obtained from the intron 1 mutation. 26 carriers (19%) had a fVIII value higher than 53%. 24 carriers (16.7%) said they had bleeding evidence in their daily lives and 40 patients (27.8%) were asymptomatic but were positive carriers of the intron 22 mutation.

CONCLUSIONS

48.58% of the patients being studied were in a fertile age stage, which shows that results must go along with genetic counseling. It was found that coagulation tests are a diagnostic aid key in countries with limited resources such as Ecuador. It was concluded that fVIII reference value in carriers must be reconsidered, since the 60% percentage was regarded as negative. The family tree allowed us to recognize possible carriers as well as obligate carriers. Carriers with low fVIII levels (34%) also showed a different bleeding type in their daily lives. The PCR was an essential diagnostic test for asymptomatic patients.

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