Molecular diagnosis of intron 22 mutation in Afghan female carriers with hemophilia A

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Introduction: A third world country like Afghanistan, which has been involved in war for more than three decades, women and girls have often been the worst victims of conflict. One of the most common hereditary hemorrhagic diseases is hemophilia A (HA). Prenatal detection in female carriers from families with HA is important to reduce the number of HA patients. It is very important to identify women who are carriers of the disease for controlling and reducing the disease.

Aim: The aim of this study was to identify women who have intron 22 inversion (INV22) in FVIII gene in families affected with severe hemophilia A.

Material & Methods: Whole blood samples were taken from 70 females from 21 unrelated families which were previously confirmed in INV22 mutation, coming from different provinces of northern, southern and central Afghanistan. Inverse shifting of PCR was used to detect intron 22 inversions (INV22), also for indirect detection, two methods of ARMS-PCR and STR analysis was used.

Results: Molecular analysis showed that 32/70 cases (46%) had INV22 and 38/70 (54%) normal. All families were informative in STR analysis, of the 70 females in 37 cases (53%) BCL1/Intron 18 T/A and 9 cases (13%) for IVS7 nt27 G/A were informative. Carriers of hemophilia A require special obstetric care with close liaison with the hemophilia center and management guidelines should be available and observed. This result represents a step for genetic counseling. Knowledge of fetal gender is very valuable to facilitate management in labour and for prenatal diagnosis.

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