Informativeness of St14 VNTR Polymorphic Marker in the Carrier Detection of Hemophilia A

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Hemophilia A is the most common hereditary severe disorder of blood clotting. In families affected with hemophilia, genetic analysis provides opportunities to prevent recurrence of the disease. This study establishes a diagnostic strategy for carrier-ship determination in Pakistani population using an extragenic polymorphic marker for the first time. The analysis of St14 VNTR (DXS52) was carried out by polymerase chain reaction (PCR), in order to determine its informativeness in terms of heterozygosity in Pakistani population. This may be a milestone for further analysis of other polymorphic markers for carrier detection and prenatal diagnosis of hemophilia. Seventy eight blood samples (Hemophiliac = 23, Normal = 55) from 15 families were analyzed for determining informativeness of St14 VNTR in carrier detection of hemophilia A. A total of nine alleles (2400, 2100, 1750, 1690, 1630, 1570, 1390, 1300, 1220 bp) was detected in the pool of subjects. 19 out of 40 females were found to be carriers with respect to the St14 VNTR polymorphic marker. The marker was informative in 73.33% of families. The expected heterozygosity rate of the St14 VNTR was 0.86 while the observed heterozygosity was 0.7. This shows that St14 VNTR is **70%** informative in our population, allowing it to be a useful marker in carrier detection, as informativeness is the direct reflection of heterozygosity of a polymorphic marker.

Key words: Hemophilia, carrier, extragenic polymorphic marker, St14 VNTR (DXS52), informativeness.