Differentiation of Fetal and Maternal DNA in Chorionic Villus Samples by STRs Analysis.

Danish Zahid , Muhammad Nadeem ,Saad Ahmed Gul Sufaida , Saima Munzir, Tahir Sultan Shamsi

National Institute of Blood Disease and Bone Marrow Transplantation Karachi, Pakistan

Introduction:

Prenatal CVS DNA analysis in pregnant ladies with family history of Beta Thalassaemia is done in many centers to diagnose Beta chain defect in the sample. In case where heterozygous (minor) DNA mutation resembling mother's mutation is detected the possibility of mother's sample contamination cannot be ruled out.STR analysis is carried out to determine nature of DNA (Fetal or non-informative) in CVS.

Aims & Objective:

To perform the STR analysis on chorionic villus samples, (CVS) and confirm the fetal origin of DNA.

Material and Methods:

Prospectively those CVS samples showing one Beta chain mutation (Beta Thalassaemia Minor) resembling the mother's mutation were also tested for STR by ARMS PCR .DNA was extracted with Qiagen extraction Kit. The Primers used for the different STR markers included D13, D18, and D21.
PCR amplification was performed and result interpreted on Polyacrylamide Gel Electrophoresis (PAGE).

Results:

From 12th Aug 2014 to 31 Aug 2016 total 863 CVS mutation analysis for Beta Thalassaemia were performed. Beta Thalassaemia single mutation resembling mother's mutation was seen in 433 cases among STR analysis D13, D18 and D21 were informative in 222 (51%), 188(43%) and 242 (56%) samples respectively. In 07 (1.6%) sample STR analysis was non-informative.

Conclusion:

STR analysis should ideally be done in all prenatal Chorionic villus samples to confirm the fetal origin of DNA .Where STR analysis is non-informative repeat samples/re-extraction and amplification is recommended.

Key words : STR analysis, Beta Thalassaemia Minor, CVS.