THE STUDY OF CORRELATION BETWEEN LEIDEN V FACTOR AND METHYLEN TETRAHYDROFOLATE REDUCTASE ENZYME GENE MUTATIONS IN PRIMIGRAVIDA WITH MISSED ABORTION IN LATE FIRST TRIMESTER

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INTRODUCTION

Pregnancy is a hypercoagulable state secondary to an increase in coagulation factors, a reduction in naturally occurring anticoagulants, and impairment of fibrinolysis.

Pregnancy losses were divided into preclinical, first trimester clinical, and second trimester. A meaningfully increased rate of preclinical pregnancy failure in Leiden mutation carriers was found than in no activated protein C deficiency patients.

Another cause of miscarriages is inherited thrombophilia following damage to the maternal factor V gene G1691A (Leiden mutation) and prothrombin gene (G20210A mutation). These alterations are well studied and the test is part of the routine diagnostics of recurrent mis carriages. In the case of factor V, both the Leiden mutation G1691A and the T1328C mutation appear to be important in the pathogenesis of RM, mainly in cases observed before the 7th week of gestation.

The most common causes of inherited thrombophilia are polymorphisms in genes encoding factor V, prothrombin (factor II), factor VII, MTHFR, and plasminogen activator inhibitor, while protein C, protein S and ant thrombin deficiency is less common.

The finding of a link between FVL carrier state and early RPL would have significant implications for clinical practice, as it would provide a scientific rationale for screening for FVL mutation and targeted thrombo prophylaxis in affected women.

AIM OF THE WORK

This work aimed to evaluate the correlation of factor V Leiden (FVL) and methylenetetrahydrofolate reductase (MTHFR) C677T gene mutation with missed abortion in primigravida in the late first trimester to identify candidates for anticoagulation therapy to improve pregnancy outcome.

SUBJECTS AND METHODS

The present study was across-sectional study conducted on 40 primigravida females recruited from EL Shatby hospital. All participating women were primigravida in their late first trimester with missed abortion. Blood samples were collected from all pregnant women enrolled in the study for DNA extraction and genotype analysis based on polymerase chain reaction and reverse hybridization. The two mutations covered by the study are the FVL, and MTHFR C667T.

RESULTS

Table (1): Distribution of the studied cases according to MTHFR and gestational age by the US (n=40)

US	MTHFR						χ^2	MCp
	Normal		Heterozygous		Homozygous			
	No.	%	No.	%	No.	%		
7w-8w	0	0.0	0	0.0	0	0.0	–	–
8w-9w	1	2.5	3	7.5	1	2.5	2.076	0.340
9w-10w	4	10.0	3	7.5	2	5.0	0.733	0.777
10w-11w	2	5.0	2	5.0	0	0.0	0.639	1.000
11w-12w	5	12.5	4	10.0	2	5.0	0.340	1.000
12w-13w	4	10.0	2	5.0	0	0.0	1.176	0.602
13w-14w	3	7.5	1	2.5	1	2.5	1.037	0.679
Total	**19**	**47.5**	**15**	**37.5**	**6**	**15.0**		

χ^2: **Chi-square test** MC: **Monte Carlo**
p: p-value for comparing between the three categories

Table (2): Distribution of the studied cases according to factor V Leiden (n= 40)

Ultrasound	Factor V Leiden						χ^2	FEp
	Normal		Heterozygous		Homozygous			
	No.	%	No.	%	No.	%		
7w-8w	0	0.0	0	0	0	0.0	-	-
8w-9w	4	10.0	1	2.5	0	0.0	0.291	0.590
9w-10w	8	20.0	1	2.5	0	0.0	2.266	0.233
10w-11w	3	7.5	2	5	0	0.0	0.272	0.627
11w-12w	8	20.0	3	7.5	0	0.0	0.054	1.000
12w-13w	3	7.5	2	5	0	0.0	0.272	0.627
13w-14w	2	5.0	3	7.5	0	0.0	2.449	0.149
Total	**28**	**70.0**	**12**	**30.0**	**0**	**0.0**		

χ^2: **Chi-square test** FE: **Fisher Exact**
p: p-value for comparing between the three categories

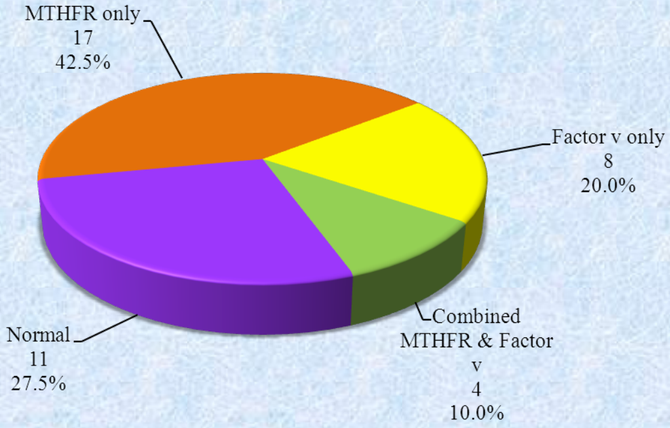

Figure: Distribution of the studied cases according to MTHFR & factor V (n = 40)

CONCLUSION

The prevalence of FVL mutations in our study did not appear significantly affect abortion in primigravida. MTHFR C667T mutations prevalence were appeared significantly have a relation to abortion.