OMICS Group Conferences
Accelerating Scientific Discovery

International Conference on
Hematology & Blood Disorders
September 23-25, 2013 DoubleTree by Hilton Hotel Raleigh-Durham Airport at RTP, NC, USA

Molecular characterization of factor V Leiden G1691A and Prothrombin G20210A mutations in Saudi females with recurrent pregnancy loss

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Recurrent pregnancy loss (RPL) is the most common complication of pregnancy in Saudi Female. Approximately 18% of all clinically recognized pregnancies are spontaneously aborted. Although several causes of RPL have been established, more than 50% of cases remain unexplained. Recently, thrombophilias have been suggested as a possible cause of RPL. Factor V Leiden (FVL) (G1619A) and Factor II (prothrombin) (G20210A) gene mutations are the most common types of hereditary thrombophilias, but are usually undiagnosed because most carriers are asymptomatic. The aim of this study was to investigate two common mutations of the factor V Leiden (G1619A) and factor II Prothrombin (G20210A) gene mutations and to assess whether recurrent pregnancy loss patterns among Saudi women differ according to causal/associated conditions. The study including 140 females, 72 had a history of 2 or more events of fetal loss in any of the 3 trimesters of pregnancy. The other 70 were clinically healthy women with a good obstetric history taken as a control group. Detection of FV Leiden (G1691A) and FII (Prothrombin G20210A) mutations were done using multiplex allele-specific PCR amplification. The results indicated that the total mutation carriage rate (AA and AG) among cases for FII was higher in frequency than FVL. Both were significantly higher than controls P>0.0001 while FIIP>0.0001. The frequencies of FVL & FII mutations related to the pregnancy loss stages showed that FVL mutation ratio was high among cases with early pregnancy loss (26%) followed by the late stage (25%) and controls (1.4%) that was statistically significant. On the other hand FII mutation ratio was high among cases with late pregnancy loss (50%) followed by early (38%) and controls (1.4%) that was statistically significant. We concluded that there are a strong association between the presence of thrombophilic mutations related to FVL and FII genes among Saudi women. Identifying heritable thrombophilia women might potentially prevent miscarriages, as well as serious maternal and neonatal complications.

Biography

Gihan E-H Gawish is an Assistant Professor Molecular Genetics. Also, she is a medical laboratory specialist since 2001. She earned her M.Sc. and Ph.D. at Mansoura University, Egypt. She is a Postdoc fellow, Oral Biological and Medical Science, University of British Columbia, Vancouver, Canada since 2011. She has published 12 peer-reviewed papers. Her research combines medical molecular genetics and advanced applications of techniques. Her research is based on the detection of biochemical signals that affect the Saudi women health and introduce these signals with modified protocols to be investigated using modern current routine equipments in clinical laboratories for aiding in the early diagnosis and development of new personalized treatment approaches. She has been included in Marquis Who's who in the world 2013.

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