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Intron 1 Inversion among Hemophilia a Patients in Basrah at the South of Iraq

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Abstract

Background/Aim: Hemophilia A is an inherited bleeding disorder that results from a mutation in FVIII gene, intron 1 inversion is one of the mutations of FVIII gene. We aimed to detect the prevalence of Intron 1 inversion among hemophilia A patients in Basrha/ south of Iraq

Subjects and Methods: A screen for the presence of intron 1 inversion at the F8 gene in 95 hemophilia A patient from Basrah at the south of Iraq. All patients have A total genmic DNAwas extracted from EDTA fresh whole blood samples drawn from all participants,then detection of intron 1 inversion by conventional PCR and use of touchdown technique.

Results: Not there any one of Hemophilia A patients has Intron one Inversion

Conclusions: Absent of intron 1 inversion compare to other studies may be referred to geographic distribution of patients in spite of low prevalence percentage of intron 1 inversion, also more number of patients increase the accuracy of the research.

Keywords: intron 1 inversion, Basrah, Hemophilia, FVIII mutation, touchdown PCR.

Introduction

The most common congenital bleeding disorder is hemophilia A, which is caused by a lack of the clotting protein Factor VIII (Callaghan, Sidonio and Pipe, 2018).

Factor VIII (FVIII) deficiency is an X-linked recessive condition that affects one out of every 5000 male births, regardless of ethnicity.(Witmer and Young, 2013).

F8 genotype mutations included large deletions (single or multiple exons), intron 1 and 22 inversions, nonsense mutations (light chain or non-light chain), small deletions/insertions/combined deletions and insertions (in poly-A runs or outside poly-A runs), splice site (conserved or non-conserved nucleotide positions), and missense mutations.(Gouw *et al.*, 2012).