The use of *BclI* restriction fragment length polymorphism for Haemophilia A carrier detection in Sudanese families

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Objectives:

Haemophilia A is the most common X-linked inherited bleeding disorder caused by a deficiency in the activity of coagulation factor VIII (Hussein *et al.,* 2008), with an incidence of 1 in 5000 male births (Jayandharan *et al.,* 2012). The aim of this study was to investigate the usefulness of *BclI* restriction fragment length polymorphism (RFLP) in the linkage analysis for carrier detection in Sudanese families, in order to formulate an informative and accurate carrier diagnosis.

Methods:

The study included 20 families with at least one subject affected with Haemophilia A, attending the Haemophilia Center at Khartoum Teaching Hospital. Thirty unrelated normal females were included as control group. Polymerase chain reaction (PCR) and restriction enzyme analysis were used to study the polymorphism in *BclI*.

	No. of chromosomes (total)	No. of chromosomes (+/-)	Allele frequency* (+/–)	Heterozygosity rate_ (expected)	Heterozygosity rate (observed)
Patients (32)	32	25/7	0.78/0.21	0.34	
Controls(30)	60	20/40	0.33/0.67	0.46	0.66
females72	144	57/87	0.40/0.60	0.48	0.54

Results:

The incidence of *BclI* (+) allele was 78%, 39.5% and 33% in patients, female relatives and control group, respectively. Expected heterozygosity for *BclI* was 0.48 in female relatives compared with 0.46 in the female control group. However, observed heterozygosity was found to be 0.54 in female relatives compared with 0.66 in the control group. The defective X chromosome was tracked in 13/20 (65%) mothers, hence 65% of the studied families were found to be informative using this marker alone.

Conclusions:

This study demonstrated that the PCR-based analysis of the *BclI* RFLP was useful in the carrier detection of Haemophilia A in the Sudanese population.

References:

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