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Application of indirect linkage analysis and direct genotyping to hemophilia A carrier detection in Sichuan, China

P. Sun, L. Ma, G. Diao, C.Q. Li and F.Z. Lin

Blood Biochemistry and Molecular Biology Research Center,
Institute of Blood Transfusion, Chinese Academy of Medical Sciences,
Chengdu, Sichuan, China

Corresponding author: F.Z. Lin
E-mail: lfangzhao@126.com / sunpan_1@163.com

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ABSTRACT. Hemophilia A (HA) is an inherited X-linked bleeding disorder caused by mutations in the factor VIII gene. Prenatal detection in female carriers from families with HA is important to reduce the number of HA patients. The purpose of this study was to detect carriers in families with HA from Sichuan, China, using linkage analysis and a direct genotyping method. A total of 18 HA families were studied. Using a combination of intron 22 inversion, intron 1 inversion, the *Bcl*I polymorphic site in intron 18, the *Hin*dIII polymorphic site in intron 19, and dinucleotide CA-repeat markers in introns 1, 13, 22, and 24, we were able to detect HA in 88.9% (16/18) of the families studied. HA was detected in the remaining two families by direct genotyping. This study gave the participants a good understanding of their genetic condition and gave us a preliminary understanding of the prevalence of each mutation in Sichuan HA patients.

Key words: Hemophilia A; Carrier detection; Linkage analysis; Direct genotyping

