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Combined analysis of the *Msp*I and *Xba*I polymorphisms in intron 22 of the factor VIII gene for detection of hemophilia A in a Korean population

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ABSTRACT. To determine the usefulness of *Msp*I/int22h-1 (intron 22 homologous region-1) polymorphism of the factor VIII gene for molecular genetic diagnosis of hemophilia A in the Korean population, *Msp*I/intron 22 and *Xba*I/intron 22 polymorphisms were analyzed in 101 unrelated Korean families with severe hemophilia A. The expected heterozygosity rates of *Msp*I/int22h-1 and *Xba*I/int22h-1 polymorphisms were 49.5 and 43.6%, respectively; these polymorphisms were not in complete linkage disequilibrium. Combined analysis using both polymorphisms provided an informative rate of 66.3%. These results suggest that PCR analysis of the *Msp*I/int22h-1 polymorphism of the factor VIII gene would be useful for

carrier detection and prenatal diagnosis of hemophilia A in the Korean population.

Key words: Factor VIII gene; Hemophilia A; Intron 22; *Msp*I; *Xba*I; Molecular genetic diagnosis

