



Molecular Biotechnology Programme
Uppsala University School of Engineering

UPTEC X 02 035		Date of issue 2002-06	
Author			
Ann-Charlotte Rönn			
Title (English)			
Analysis of sequence variations in the factor VIII gene in haemophilia A patients			
Title (Swedish)			
Abstract			
A 23kb region upstream of the factor VIII gene was screened for mutations in haemophilia A patients using solid phase fluorescent chemical cleavage of mismatch. The haplotypes found could not explain the difference in phenotype between patients sharing the same causative mutation. A putatively functional mutation was found in the promoter region. To further investigate the ancestry of the causative mutations in this study, the patients were haplotyped for several known intragenic and extragenic polymorphisms. This study shows that some of the causative mutations investigated probably have arisen on more than one occasion. No conclusive evidence can be given in this study regarding the functional role of the promoter mutation.			
Keywords			
Haemophilia A, factor VIII gene, polymorphism, haplotype, chemical cleavage of mismatch			
Supervisors			
Prof. Francesco Giannelli & Dr. Peter Green King's College, London, UK			
Examiner			
Dr. Matti Nikkola Uppsala University, Uppsala, Sweden			
Project name		Sponsors	
Language		Security	
English			
ISSN 1401-2138		Classification	
Supplementary bibliographical information		Pages	
		26	
Biology Education Centre Biomedical Center Husargatan 3 Uppsala Box 592 S-75124 Uppsala Tel +46 (0)18 4710000 Fax +46 (0)18 555217			