Bleeding tendency and health-related quality of life in carriers of haemophilia

Akademisk avhandling

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av
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Avhandlingen baseras på följande delarbeten

I Olsson A, Hellgren M, Berntorp E, Ljung R, Baghaei F.
Clotting factor level is not a good predictor of bleeding in carriers of haemophilia A and B.
Blood Coagulation and Fibrinolysis 2014; 25: 471-475

II Olsson A, Hellgren M, Berntorp E, Baghaei F.
Association between bleeding tendency and health-related quality of life in carriers of moderate and severe haemophilia.
Haemophilia 2015; 21: 742-746

III Olsson A, Hellgren M, Berntorp E, Holmström M, Baghaei F.
Bleeding phenotype in carriers of haemophilia A does not correlate with thrombin generation.
Haemophilia 2015; 21: e111-e113

IV Olsson A, Ljung R, Hellgren M, Berntorp E, Baghaei F.
Phenotype and genotype comparisons in carriers of haemophilia A.
Haemophilia 2016; 1-3

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Abstract

Haemophilia A and B are X-linked disorders caused by impaired synthesis of coagulation factors VIII and IX, respectively. Women who carry the haemophilia trait have about 50 % of normal factor levels. Due to skewed X-chromosome inactivation, factors in carriers may however range from levels corresponding to those in men with haemophilia up to normal levels. One hundred and twenty six haemophilia carriers and 90 controls were included in the study. In **paper I**, bleeding tendency was evaluated with a standardised bleeding assessment tool. We found increased bleeding tendency among the carriers of haemophilia A and B, compared to the control group. The bleeding tendency was weakly correlated to FVIII levels. In **paper II**, health-related quality of life (HRQOL) in haemophilia carriers was compared to a control group and the normative population. HRQOL was evaluated with the Short-Form 36 questionnaire. Symptomatic carriers had lower scores in the General Health, Social Functioning and Mental Health domains, compared to the control group. These differences disappeared when comparisons were made with the normative population. **Paper III** demonstrates that thrombin generation potential, evaluated by the calibrated automated thrombography method (CAT), did not differ significantly in symptomatic and asymptomatic carriers of haemophilia A. The results of **paper IV** indicate that there was no association between bleeding tendency in haemophilia A carriers and genotype, evaluated by comparison of null and non-null mutations. In conclusion, the results suggest that carriers of haemophilia may have increased bleeding tendency, especially during haemostatic stress. Carriership did not affect HRQOL in comparison to the normative population. Factor levels as well as thrombin generation capacity appear to be inadequate for prediction of bleeding tendency in carriers. The bleeding tendency in haemophilia A carriers was not influenced by the genotype.

Keywords: Carriers of haemophilia, bleeding, SF-36, thrombin generation, genotype.

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