Factor X Deficiency

**Alternative Names**
Stuart-Prower Factor Deficiency
Coagulation Factor X
F10

**WHO International Classification of Diseases**
Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism

**OMIM Number**
227600

**Mode of Inheritance**
Autosomal recessive

**Gene Map Locus**
13q34

**Description**
Factor X deficiency is a rare inherited bleeding disorder that causes abnormal blood coagulation, resulting from a shortage of the plasma protein factor X. It is estimated that Factor X deficiency affects 1 individual per 500,000-1,000,000 population worldwide. Inheritance is in an autosomal recessive trait; with heterozygotes most often remaining clinically asymptomatic. The clinical phenotype is of variable hemorrhagic symptoms in homozygous individuals, including easy bruising, hematuria, hemarthroses, soft tissue hemorrhages, menorrhagia, and recurrent epistaxis.

**Molecular Genetics**
Factor X is one of the vitamin K-dependent serine proteases, playing a crucial role in the coagulation cascade, as the first enzyme in the common pathway of thrombus formation. The gene for factor X maps to the long arm of chromosome 13, and spans approximately 25 kb of genomic DNA. The gene consists of eight exons, each of which encodes a specific functional domain within the protein: exon 1 codes for the signal peptide, exon 2 for the propeptide and the Gla-rich domain, exon 3 for the short aromatic acid-rich stack, exon 4 for the first epidermal growth factor (EGF) domain, exon 5 for the second EGF domain, exon 6 for the activation peptide, and exons 7 and 8 for the catalytic domain. Both the gene structure and the amino acid sequence show homology to other vitamin K-dependent clotting factors, suggesting their origin in a common ancestral protein.

**Epidemiology in the Arab World**

**Morocco**
Boxus et al. (1997) described a 3 year-old boy, born to consanguineous Moroccan parents, with the rare combination of congenital factors VII and X deficiency. Originally, the child had a prolonged partial thromboplastin time discovered fortuitously. This finding led to the diagnosis of combined factors VII and X deficiency. His siblings had the same deficiencies.

**Sudan**
El Kalla and Menon (1991) described a baby boy who was born to second-degree consanguineous couple from Sudan. He was admitted at 8 days old with a history of hematemesis and skin hematoma since the first day of life. After treatment, he remained symptom free until he was 8 months old.

**United Arab Emirates**
El Kalla and Menon (1991) described four neonates with congenital Factor X deficiency who presented soon after birth with bleeding episodes. The first case is of a full-term baby boy who was born to a first-degree consanguineous couple. At 3 days old, the infant developed anemia and jaundice with irritability and bulging fontanel. Initially, he was on replacement schedule every third day
with a post-infusion Factor X level of more than 80%. At four years of age, the child was normal clinically and neurologically with no bleeding manifestations, except for occasional skin petechia and ecchymoses. The second case was for a full-term baby girl who was born to a first-degree consanguineous couple. At her third day of life, she started to have spontaneous bleeding and bleeding from injection sites. In spite of treatment, she sustained massive intraventricular and intracerebral hemorrhage and expired at 4 months. The third case was of a baby boy who was admitted with a history of hematemesis and skin hematoma since the first day of life [See also: Sudan > El Kalla and Menon, 1991]. The fourth case was of a baby girl born to a first-degree consanguineous couple. The infant was admitted at 4 days old with a history of bleeding from the umbilical stump and skin petechia. At two years of age, Factor X replacement therapy resulted in no serious clinical bleeding tendency. Factor X analysis of the parents of the first patient showed Factor X activity between 60-70%, indicating a heterozygous defect. Prenatal diagnosis of a subsequent pregnancy in the mother of patient 1 was done. Fetal blood sampling at 20 weeks of gestation confirmed a Factor X deficiency in the fetus and the pregnancy was terminated.

References

Contributors
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