# A Novel Mutation in Child with Severe Congenital Factor X Deficiency

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## OBJECTIVES

Factor 10 (F10) is a vitamin K-dependent coagulation factor gene that plays an important role in blood clotting process encodes 10 proteins. Mutations of this gene have been associated with factor 10 deficiency (OMIM # 227 600).
F10 gene consists of 8 exons. The most frequently mutated region of the Gla domain encoded by exons 2 and 7-8. They are the catalytic domain encoded by exons.

## METHODS

Forty days old female patient was admitted to hospital with vomiting. Widespread spontaneous subdural hemorrhage was detected. He was operated on three times. Prothrombin time and activated partial thromboplastin time were prolonged. Factor 10 activity was 0%. He did not respond to vitamin K. Their parents were close relatives. There was no family history of bleeding. Giving fresh frozen plasma, operations were carried out successfully. Patients received prophylaxis for two days per week with activated prothrombin complex concentrate (FEİBA). Currently, she was two-year-old and bleeding was not detected.

## RESULTS

Genomic DNA was isolated from peripheral blood nucleated cells. Target gene of Polymerase Chain Reaction coupled with, after sequencing reaction and analyzed with the ABI 3130 XL. Performed genetic analysis covers only the coding region of the gene F10.

C.1231c identified in patients with homozygous> T mutation in the absence of the F10 is one of the most frequently reported to be mutated in exon 8 is located. His mother and father were found to be carriers.

## CONCLUSIONS

In our patient, the F10 gene located in exons 8 homozygous c.1231c> T (NM_000504.3: c.1231c> T) mutation has been reported. This mutation has not been reported in the literature and related database previously.
This mutation at position 411 in the protein (NP_000495.1: p.gln411).encoded by the F10 gene leads premature termination codon and in silico analysis suggests constructive disease mutation.

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Clotting Factor Concentrates
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