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Brief Communication

Novel duplication in the *F12* gene in a patient with recurrent angioedema

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Highlights

- â€¢ A novel mutation was identified in a case of recurrent angioedema with normal C1-INH.
- â€¢ The mutation located in *F12* gene (c.892_909dup) is missing in control population.
- â€¢ It is causing the repeated presence of 6 amino acids in factor XII.
- â€¢ It shows partial co-segregation with angioedema symptoms in the family of the patient.

Abstract

Edema formation is mediated by histamine or bradykinin release and may have several hereditary and acquired causes. In hereditary forms of bradykinin-mediated angioedemas, mutations in the genes encoding C1-inhibitor (*SERPING1*) as well as coagulation factor XII (*F12*) have been described. We present a novel *F12* gene mutation, a duplication of 18 base pairs (c.892_909dup) in a 37-year-old woman with recurrent angioedema and normal C1-inhibitor level. A single episode of facial edema in the family of the patient showed co-segregation with the mutation. This duplication is causing the repeated presence of 6 amino acids (p.298â€“303) in the same region of factor XII, as those three mutations described previously in cases of hereditary angioedema with normal C1-INH function. These results may confirm the importance of the proline-rich region of factor XII protein in edema formation.

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Keywords

Angioedema; Factor XII; Mutation

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