Identification of the recently described plasminogen gene mutation p.Lys330Glu in a family from Northern Germany with hereditary angioedema

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Hereditary angioedema (HAE) is a life-threatening disease characterized by recurrent episodes of subcutaneous and mucosal swellings and abdominal cramping. Corticosteroids and antihistamines, which are usually beneficial in histamine-induced acquired angioedema, are not effective in HAE. Therefore, diagnosing HAE correctly is crucial for affected patients. We report a family from Northern Germany with six individuals suffering from recurrent swellings, indicating HAE. Laboratory tests and genetic diagnostics of the genes; SERPING1; , encoding C1 esterase inhibitor (C1-INH), and; F12,; encoding coagulation factor XII, were unremarkable. In three affected and one yet unaffected member of the family, we were then able to identify the c.988A >(also termed c.1100A >) mutation in the; plasminogen; (; PLG); gene, which has recently been described in several families with HAE. This mutation leads to a missense mutation with an amino acid exchange p.Lys330Glu in the kringle 3 domain of plasminogen. There was no direct relationship between the earlier described cases with this mutation and the family we report here. In all affected members of the family, the symptoms manifested in adulthood, with swellings of the face, tongue and larynx, including a fatal case of a 19ăyear-old female individual. The frequency of the attacks was variable, ranging between once per year to once a month. In one individual, we also found decreased serum levels of plasminogen as well as coagulation factor XII. As previously reported in patients with PLG defects, icatibant proved to be very effective in controlling acute attacks, indicating an involvement of bradykinin in the pathogenesis.
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