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Combined Congenital Deficiency of Factor V and Factor VIII

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The combined congenital deficiency of factors V and VIII is an interesting and rare syndrome which, to our knowledge, has never been described in childhood. We would like to report a newborn infant, born to consanguineous parents, with this combined defect.

Case Report

Z.G. was born spontaneously at the 38th week of gestation. After delivery he presented serious bleeding manifestations and even after having received several transfusions of fresh plasma he did not improve. Development of hydrocephalus, following purulent meningitis, required shunt procedure. He died at 2 months because of respiratory complications. The laboratory findings revealed very low levels of both factors V and VIII (factor V, 4%; factor VIII, 100/o), prolonged prothrombin and partial thromboplastin times; factor VIII R:Ag was 100%. Routine laboratory tests were normal, split fibrin products absent. The coagulation study of the patient's relatives showed in the

< LJ
ΓJ O Studied, normal
HI φ Not studied
[+] 0 Deceased
ψ\ c) Factor V asymptomatic deficiency
@ Hemophilia carrier
B Propositus, factor V and factor VII! deficiency
Φ CD CD O © Φ ΘΦffl E Q u
UÏ%Γ% ı-S-S ω %6 ı ó 5 6 ı ı
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Eiöö r%ı!% fñl

Fig. 1. Family pedigree.

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mother a factor VIII/factor VIII R:Ag ratio compatible with a carrier of hemophilia A (0.48) and a mild asymptomatic reduction of factor V in 1 paternal and in 3 maternal relatives. The first brother had likewise been affected by bleeding manifestations and died a few days after birth.

Discussion

The combined deficiency of factors V and VIII is a heterogeneous condition [2]. Analysis of the pedigree in our case (fig. 1) showed that factor V deficiency could have been inherited with autosomal recessive mode. Moreover, as the factor VIII/ factor VIII R:Ag ratio in the mother was within the range of hemophilia A, a casual concomitant association between classical hemophilia A and autosomal recessive deficiency of factor V is possible.

This condition could have caused a very marked reduction of both factors, accompanied by serious bleeding manifestations even during the neonatal period. Pathogenesis of the defect is uncertain. By means of immunological studies [1], a common precursor deficiency of factors V and VIII procoagulant has been suggested. Recently Graham [3] hypothesized that it could be coded for an enzyme which is not able to activate the two factors completely in deficient individuals.

However, the biochemical basis of this rare disorder is not yet clear and further contributions will be necessary to clarify it.

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