Normal Factor VIII Antigen Level in Combined Congenital Deficiency of Factor V and Factor VIII

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As a result of immunological studies, several conditions due to structural abnormalities of clotting factors have been described. Only one patient with congenital combined factor V and factor VIII deficiency has been investigated immunologically so far and found to have a normal factor VIII antigen level [1].

We have recently studied another patient with this peculiar disease [2] and were therefore able to carry out some immunological studies on whole plasma [3]. The factor VIII activity was 14% of normal, yet immunologically the factor VIII level was > 1000 u/o (fig. 1). An antiserum obtained from rabbits immunized with human factor VIII was used (Behringwerke).

Therefore, it seems that this peculiar disease is associated with normal or even elevated factor VIII antigen levels and low factor VIII activity. The discrepancy with biological factor VIII activity seems similar to that found in classical hemophilia A [1].

Some genetic considerations seem justified, namely that both sex-linked and autosomal recessive factor VIII deficiencies are associated with low factor VIII activity and normal or elevated factor VIII antigen. On the contrary, autosomal dominant conditions (von Willebrand's disease)

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Fig. 1. Electroimmunoassay (Laurell) in (1) undiluted von Willebrand's disease plasma; (2) undiluted hemophilia A plasma; (3) undiluted combined factor V and factor VIII deficiency plasma; (4)-(7) 1:8, 1:4, 1:2, and undiluted pooled normal plasma, respectively.

are associated with low factor VIII biological activity and low factor VIII antigen.

References

- Zimmerman, T. S.; Ratnoff, O. D., and Powell, A. E.: Immunological differentiation of classic hemophilia (factor VIII deficiency) and von Willebrand's disease. *J. clin. Invest.* 50: 244 (1971).
- Girolami, A.; Brunetti, A., and De Marco, L.: Congenital combined deficiency of factor V and factor VIII in a male born from a brother-sister incest. *Blut* 28: 33 (1974).