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00 0440 HEPARIN-AFFINITY OF ANTITHROMBIN III IN A FAMILY WITH CONGENITAL ANTITHROMBIN III DEFICIENCY

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We found a new thrombophilic family with antithrombin III / AT-III / deficiency. In the members of this family both immunologic and functional methods revealed similarly decreased levels of AT-III. Gelfiltration displayed identical size of AT-III molecule of patient and normal alike. On the basis of these findings we assumed that in this family normal AT-III is produced but only in diminished quantity. Experiments on the heparin-affinity of AT-III did not support this assumption. The AT-III of the proposita migrated slower than that of normal person in the heparinized agarose gel. In the course of the heparin-affinity chromato - graphy the AT-III of the proposita could be eluted at lower salt concentrations than normal AT-III.

Thus we conclude that even in the case of "true" AT-III deficiency the molecule might have some qualitative deviation from normal.

.15 0441 THREE TYPES OF HEREDITARY ANTITHROMBIN III DEFICIENCY

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The authors detected in the last seven years 15 patients with hereditary antithrombin III /AT III/ abnormality. All of them had typical clinical signs of recurrent arterious and venous thromboembolie. The abnormality inherited as an autosomal trait. Three types of the abnormality could be observed. In Type I both quantity and function of AT III were extremely decreased. In Type II AT III is normal in quantity but abnormal in function. In Type III AT III is quantitatively normal and also its function seems normal as far as its basic activity is concerned /activity measured in absence of heparin/, but its abnormality becomes manifest in the presence of heparin in vitro/and also in vivo/. 5 of the patients belonged to Type I, 4 to Type II and 6 to Type III. In 60 examined family members of the 15 patients an abnormal AT III could be observed in 44, clinical signs in 23.
The examination of AT III activity in the presence of a given amount of heparin is of great importance in recognition of the different types of antithrombin III abnormalities.

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