FAMILIAL CEREBRAL HAEMORRHAGE AND ASSOCIATED BERRY ANEURYSM WITH NORMAL TYPE III COLLAGEN

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Introduction

Familial clustering of berry aneurysms suggests an autosomal dominant inheritance of a defect in arterial walls. In a family with five affected members in three generations we examined type III collagen synthesis from two affected family members as abnormalities of the cerebral circulation have been described in association with mutations of the COL3A1 gene.

Methods

4mm punch biopsies were taken from two affected family members. Primary fibroblast cultures were established and radiolabelled procollagens and collagens examined by Polyacrylamide Gel Electrophoresis (PAGE) using standard techniques.

Results

No abnormalities of the processing of procollagen to collagen nor of the medium and cell layer collagens were detectable in the two affected individuals as compared with normal controls. In particular collagen/procollagen alpha 1(1), alpha 2(1), alpha 1(III), alpha 1 and alpha 2(V) profiles were unchanged.

Conclusions

We have not detected any obvious abnormality of collagens I, III and V in the three generation family with familial berry aneurysm. This contrasts with Vascular EDS IV in which vascular malformations and collagen III mutations are common. The evidence in this family suggests that other candidate genes should now be examined.

CEREBROVASCULAR ACCIDENT RISK FACTORS PRESENT AT TIME OF DIAGNOSIS

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Introduction

Risk factors for cerebrovascular accidents (C.V.A.) include transient ischaemic attacks (T.I.A.), previous C.V.A., hypertension, atrial fibrillation, ischaemic heart disease, diabetes mellitus and cigarette smoking. We examined the risk factors which were present at the time of diagnosis of a cerebrovascular accident.

Methodology

Data was collected from 100 consecutive cases (52 females and 48 males) of ischaemic cerebrovascular accidents occurring in patients admitted to Care of the Elderly wards and attending the Day Hospital. All patients had infarcts proven on C.T. scan.

Results

The average age of presentation for females was 78.8 years and for males was 75.6 years. The most common risk factor [66/100 (66%)] present prior to the index C.V.A. was evidence of a previous cerebrovascular insult. Of these 66 patients, 12 had a history of T.I.A.'s, 10 had a diagnosis of a cerebrovascular accident. 20/100 (20%) and diabetes mellitus in 10/100 (10%). 4/100 (4%) had previously undergone caroid endarterectomy. 34/100 (34%) had three or more of the above risk factors, 77/100 (77%) had at least two, while 7/100 (7%) had none.

Conclusion

The most common risk factor identified was evidence of previous cerebrovascular insult. The prevalence of hypertension was equal in both males and females while ischaemic heart disease was twice as common in males.