Familial intracranial aneurysms


The familial aggregation of intracranial aneurysms is well known but has been difficult to investigate because of the invasive nature of screening tests. These authors set out to estimate the prevalence of intracranial aneurysms in symptom-free first degree relatives of affected patients, and to find out if familial aggregation of cases could be accounted for by pedigrees with adult polycystic renal disease. Records of 1445 patients with cerebrovascular malformations were used to identify 1150 with intracranial aneurysms. Telephone enquiry and further records were studied to identify relatives who were also affected and symptomatic. Magnetic resonance angiography was offered to 698 asymptomatic first degree relatives over 30 years of age, with digital subtraction angiography considered where abnormalities were detected. Ultrasonography was used to screen for renal disease. A total of 438 relatives were screened of whom 36 were shown to have intracranial aneurysms. The incidence in first degree relatives of patients is 8.7%. No new pedigrees with polycystic renal disease were identified by the screening. The authors point out that it is not possible to know how many screened people were given false negative results in this investigation. One relative with a normal screening result has subsequently had a subarachnoid haemorrhage. Despite this problem the study has suggested a two to fourfold increase in risk to first degree relatives. A further concern is the high cost of magnetic resonance angiography and its lack of universal availability. Detection of asymptomatic aneurysms would allow surgery to be carried out much more safely than in the acute presentation of a ruptured aneurysm. Seven percent of families with familial intracranial aneurysms were affected with polycystic renal disease. The authors suggest that there is a distinct condition of familial intracranial aneurysms and highlight the need for further studies to document the best genetic model for this condition.

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Incidence of insulin-dependent diabetes mellitus among Sardinia-heritage children born in Lazio region, Italy


The interaction between genetic and environmental factors in the aetiology of diabetes continues to puzzle. Muntoni and colleagues have studied the incidence of insulin dependant diabetes in children born in the Lazio region of Italy (which has a low incidence of diabetes of 6.5 per million) who are of Sardinian ancestry (which has a high incidence of 30.2). The Sardinian emigration took place from 1950 to 1980, and those living in Lazio are said to be representative of the general Italian population. Children born of one or two parents of Sardinian ancestry born in Lazio were identified from census data. Two independent methods were used to identify diabetic children and the same 17 children were identified with both methods; three had two Sardinian parents and seven each had a Sardinian father or mother. There did not seem to be high exposure to Sardinian environmental factors, as none returned to the island frequently or used special foods from Sardinia. The children of two Sardinian parents have an incidence of diabetes similar to Sardinians living on Sardinia (but because of small numbers the figure has wide confidence limits). The children who had one Sardinian parent have an incidence of diabetes half that of Sardinians but double that of those living in Lazio. Drawing conclusions from the data is still puzzling. The results suggest a strong genetic influence in the aetiology of insulin dependent diabetes. Sardinians are apparently a homogenous population who might carry certain genes at high frequency. However, the retention of the high risk after migration appears to be at odds with some earlier studies in other locations. Furthermore incidence of diabetes on Sardinia has increased steeply over the past 25 years or so and it seems unlikely that this change is genetically mediated. The possibility of environmental factors which are unidentifed but which have been taken with migrants is not yet excluded. The authors emphasise the contribution of both genetic and environmental factors to diabetes.

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