ACE gene I/D polymorphism and the presence of renal failure or hypertension in autosomal dominant polycystic kidney disease (ADPKD)

Sir,

Epidemiological data have suggested that interactions between multiple genetic and environmental factors are involved in the process of progressive renal damage in the course of various kidney diseases, including autosomal polycystic kidney disease [1–4].

Recently, we read with interest the results of a meta-analysis of studies examining the association between angiotensin I-converting enzyme (ACE) gene insertion/deletion (I/D) polymorphism and the presence of end-stage renal disease (ESRD) in patients with ADPKD [5]. Based on the analysis of combined data from 13 reports, Pereira et al. [5] found no proof for the involvement of ACE gene I/D marker in the development of ESRD or hypertension in ADPKD patients.

Inspired by this report, we looked into ACE I/D genotypes obtained in a small sample of 105 Caucasian ADPKD patients [58 women and 47 men, median age 43 years (25–75%: 35–54 years), of whom 42 presented renal failure (S-creatinine ≥ 130 μmol/l), with median serum creatinine of 432 μmol/l (25–75%: 192–780 μmol/l)]. Seventy patients were hypertensive (systolic blood pressure ≥ 140 mmHg and/or diastolic blood pressure ≥ 90 mmHg, or antihypertensive treatment). There were no significant differences in I/D genotype distributions between ADPKD patients, with and without renal failure. Frequencies of D/D, I/D and I/I genotypes among patients with renal failure were 33, 57, 10%, respectively, and 33, 49 and 18% in those with normal S-creatinine levels. In hypertensive patients, D/D, I/D and I/I genotype frequencies were 34, 57 and 9%, while in normotensives, they were 34, 43 and 23%, respectively. Genotype distribution obtained in 130 healthy controls was 37, 37 and 16% (D/D, I/D and I/I, respectively). We found no differences in serum ACE levels between ADPKD patients with and without renal failure, or in patients with hypertension vs normotensives. However, ACE levels were highest among carriers of the DD genotype, and lowest in the I/I group, with heterozygotes presenting intermediate levels.

In summary, our data obtained in a small sample of ADPKD patients failed to show an association between the ACE gene polymorphism and the presence of renal failure or hypertension, and can be added to further meta-analyses of ADPKD patients.

Conflict of interest statement. None declared.

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Epidemiology and investigation of acute abdominal presentations in autosomal dominant polycystic kidney disease

Sir,

Patients with autosomal dominant polycystic kidney disease (ADPKD) commonly present with a variety of inter-current abdominal symptoms that can lead to diagnostic difficulties [1,2], but there is currently no consensus regarding optimal diagnostic work-up.

We carried out a retrospective review of medical case notes of 158 ADPKD patients, mean age 50.6 ± 15 years, managed in one renal centre since January 1995. We wished to determine the epidemiology of abdominal pain and haematuria occurring in patients with established ADPKD, and also to analyse the investigative practice and clinical diagnostic accuracy in relation to these acute abdominal presentations.

There was a low incidence of acute abdominal problems with 61 episodes of abdominal pain and/or macroscopic haematuria occurring during a follow-up period that averaged 7 years (total follow-up time 1106 years); 46 (29.1%) patients experienced one or more episodes (13 patients had two episodes and one patient had three episodes), with an average of one episode occurring per 15.3 patient-years. Definitive diagnosis was determined in only 41 (65.6%) episodes. Abdominal pain was responsible for 61.7% of the episodes and a diagnosis was reached in 85% of these cases. The aetiology of macroscopic haematuria (sole presenting symptom in 31.1% of episodes) remained indeterminate in almost half the cases (47.4%). Overall, the definitive diagnoses were infection (16%), haemorrhage (18%), cyst enlargement (12%), renal calculi (10%) and unknown cause in 34.4%. The initial presumptive clinical diagnosis was correct in 72.5% of cases where a definitive diagnosis was reached by the admitting team.

Use of imaging investigations (in 90.2% of episodes) was inconsistent—33 episodes were investigated with ultrasound scan (US), 8 CT only, and in 14 patients CT was performed because US had been inconclusive. The diagnostic rate was similar with CT and US only, but it is recognized that there can be difficulties in use and interpretation of either imaging technique in patients with ADPKD. For example, in the diagnosis of renal infection, ultrasound is unable to distinguish complex septated patterns of multiple cysts from cyst abscesses [3]. Alternative forms of investigation such as nuclear scintigraphic studies have also been used...