Dear Sir,

The introduction of molecular genetics has revealed the genetic heterogeneity of autosomal dominant polycystic kidney disease (ADPKD) [1]. The human gene mapping nomenclature refers to the type of ADPKD linked to chromosome 16p as PKD1 [2]. Several studies suggest that about 90% of ADPKD in the Caucasian population is due to the PKD1 [3]. A project was initiated by the Medical and Health Research Programme of the Commission of the European Communities, Directorate Biology (COMAC-BIO) on July 1, 1989 to analyze the genetic heterogeneity of ADPKD in Europe. In order to contribute to the attempts to define the genetic diversity of ADPKD in different populations, we provided DNA samples of a well-documented Turkish family with ADPKD for genetic linkage studies in Rijks University, Leiden. The pedigree of this large family with ADPKD is given in figure 1. The genetic analysis was conducted with markers flanking the PKD1 gene. HBAP1, which is a distal marker, and 16AC2.5 and SM7, which are proximal markers for PKD1, were used in the genetic linkage analysis. It was seen that all the affected individuals shared the haplotype HBAP1 allele 2 and 16AC2.5 allele 6. According to the available DNA samples it was calculated that there was a chance of 97% in this Turkish family that the disease was caused by a mutation in chromosome 16.

The European Community consists of populations of different ethnic origins and the same heterogeneity is observed even in different parts of the same country [4]. In order to better define the underlying genetic mechanisms of this common genetic disease and provide clues to diagnosis and treatment, every effort should be undertaken to study larger groups of families with different ethnic origin.

References


Fig. 1. Pedigree of a Turkish family with ADPKD.

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