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Batch Citation Matcher
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Special Queries
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Earlier diagnosis of autosomal dominant polycystic kidney disease: importance of family history and implications for cardiovascular and renal complications.

Taylor M, Johnson AM, Tison M, Fain P, Schrier RW.

University of Colorado Health Sciences Center, Denver, CO 80262, USA.

BACKGROUND: Autosomal dominant polycystic kidney disease (ADPKD) is a common and serious cause of hereditary renal disease. The emerging possibilities to intervene early in the disease course elevate the importance of both accurate and early diagnosis of ADPKD. Family history analysis is a simple and inexpensive approach to identifying individuals at risk for ADPKD. We hypothesized that advances in knowledge of and potential interventions for ADPKD have led to increased use of family history screening. METHODS: We distributed surveys to 1,527 subjects from our ADPKD research database to determine the extent to which examination of family history was used to diagnose ADPKD, by birth cohort. RESULTS: Six hundred thirty-seven subjects with ADPKD (42%) completed and returned surveys. Family history analysis led to the initial ADPKD diagnosis in 49% of all subjects overall. In the birth-cohort analysis, ADPKD was more likely to have been diagnosed in individuals born between 1951 and 1974 because of family history (55% versus 38%; $P < 0.0002$) and patients were younger at diagnosis (27 versus 39 years; $P < 0.0001$) than individuals born before 1951. CONCLUSION: In a large cohort of subjects with ADPKD, we found increased use of family history analysis as a tool for diagnosing ADPKD and earlier age of diagnosis in the more recent birth cohort. This trend may reflect increased overall awareness of ADPKD by physicians, as

well as encouraging hypertension and proteinuria treatment outcome data that may depend on intervening early in the course of disease.

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