ABSTRACT

Genetic Risk in Primary care project: can a standardized family history questionnaire (FHQ) identify genetic risk in general practice?

Nadeem Qureshi, University of Nottingham, Queens Medical Centre

Objectives: To assess the feasibility of using a self-administered Family History Questionnaire (FHQ), given to patients when they register with a family doctor, to identify genetic risk.

Methods: A cross-sectional survey, with patients completing a brief paper based self - administered FHQ at the time they came to register with the doctor's office, was undertaken in 13 British family doctors' offices. The FHQ systematically enquired about family history of genetic conditions in first degree relatives, grandparents and significant other relatives. This was followed by a standard pedigree recording interview, adopting the approach used by genetic counselors in clinical practice. Two clinical geneticists scored each instrument (FHQ and pedigree interview) for familial risk of both chronic multifactorial diseases and single gene disorders. Through consensus meetings, the geneticist reached agreement on the scores given to each instrument. The genetic risks scores identified by both instruments were compared.

Results: 326 new registrants participated, with 121 completing both the FHQ and genetic interview. A score 'higher than population risk' was obtained with 24% of FHQs and 36 % of the genetic interview. There was a 77% agreement in the scores obtained with the 2 instruments, with a moderate kappa of 0.52. (95% CI 0.40 -0.64). There was 90% agreement in the scores for a family history of premature coronary heart disease (Kappa 0.67; 95% CI 0.49 to 0.85).

Conclusions: Compared to the pedigree interview, the FHQ identified most informants scored at normal population risk, those "warranting specialist referral", and informants with a family history of premature coronary heart disease. It was less effective in identifying those with a possible single gene disorder for whom "more information was required". Genetic advances, and the establishment of evidence-based interventions, will lead to an increasing number of occasions when enquiring about the family history will benefit the health of patients and their families.