Phenotypic description of a large cohort of PSC patients in the UK
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Introduction

Primary Sclerosing cholangitis (PSC), a chronic cholestatic liver disease of unknown aetiology or pathogenesis, remains an area for active research. We describe the demographic and phenotypic characteristics of a UK cohort of 1194 patients with PSC.

Methods

All patients were recruited as part of an ongoing national collaborative effort (PSC-UK) between October 2008 and May 2011. The diagnosis of PSC was confirmed on the basis of characteristic ERCP, MRCP or histology. Patients with small-duct PSC were also included. All patients completed a descriptive phenotypic questionnaire sent at recruitment. Demographic, phenotypic data and family history were extracted from the participant questionnaires.

Results

1194 patients have returned completed questionnaires. Median age at recruitment was 59 years and 63% were male (male to female ratio = 1.7:1). 64% of patients were lifelong non-smokers and only 4.3% were smoking at recruitment. Protective effect of smoking against IBD was maintained in the PSC population (Figure 1). 63.5% reported inflammatory bowel disease, split into 87% with Ulcerative colitis and 13% with Crohn’s disease. 1.5% had a sibling with PSC and 14% had a sibling with inflammatory bowel disease. Further, 0.5% had one or more children with PSC and 4.6% had children with inflammatory bowel disease. 24% were asymptomatic, but over 50% reported itching and fatigue as the presenting symptom. Jaundice was present in 35%. The most common associated autoimmune disease was thyroid disease, present in 9% followed by Coeliac disease in 2% and type 1 diabetes mellitus in 2% (Figure 2). 16% of patients reported pan-procto or sub-total colectomy and 12.2% had undergone cholecystectomy. 2.7% of patients reported a history of colon cancer.

Conclusions

This is the largest reported demographic and phenotypic description of a single cohort of PSC patients. PSC is more common in young, non-smoking male patients. The frequency of associated inflammatory bowel disease is similar to that reported in other studies. It is plausible that siblings of patients with PSC have an increased risk of not only PSC but also inflammatory bowel disease. These data are consistent with increasing evidence pointing to a role of genetic factors in the pathogenesis of PSC.