binding protein 1, ABP1; EC 1.4.3.6). ABP1 enzyme activity in bowel mucosa of patients with IBD is 50% lower than in healthy individuals. The reason for this decrease remains unknown, but it may be due in part to polymorphisms in the ABP1 gene. The present study investigated whether a common single nucleotide polymorphism (SNP, refSNP ID: rs1049793), which is located in exon 3 (2029G) and cause amino acid substitutions (His645Asp) in the ABP1 enzyme, is related to CD.

Aims: To analyse the association between the presence of a non-synonymous single nucleotide polymorphism at diamine oxidase gene and the risk of developing Crohn's disease (CD) and to analyse its influence on the clinical course of these patients.

Methods: In this prospective, case-control study, 210 unrelated Caucasian consecutive CD patients were recruited at the Inflammatory Bowel Disease Unit of a single tertiary centre (Hospital Clinico San Carlos) in Madrid, Spain. All patients were phenotyped and followed up for a median time of 8.7 years (range 4.1–14.5 years). A total of 261 healthy volunteers from the same geographic area were also recruited and matched with patients. Both cases and controls were analysed for the presence of His645Asp amino acid substitutions in the diamine oxidase enzyme, using amplification-restriction procedures. The protocol was approved by the Ethics Committee of the Hospital and all patients and controls gave informed consent before inclusion in the study.

Results: No significant differences were found in the distribution of ABP1 alleles between CD patients and healthy volunteers [for variant alleles = OR 1.15 (95% CI 0.86–1.55)]. The distribution of ABP1 genotypes did not differ when patients were subdivided according to gender, mean age at diagnosis, mean duration of the disease, family history of IBD, smoking habit, previous appendectomy or tonsillectomy, Montreal classification, perianal CD, extraintestinal manifestations of CD (cutaneous, articular, ocular and hepatic) and severity, i.e. need for immunosuppressive therapies, biological treatments and/or surgery.

Conclusion: Our results suggest that the His645Asp polymorphism of the histamine metabolising enzyme ABP1 may not be related to the risk of developing CD. Moreover, this gene does not seem to play a role in disease activity.

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MEFV mutations modify the disease severity of inflammatory bowel disease

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Background: Genetic susceptibility plays an important role in the etiology of inflammatory bowel disease (IBD). Familial Mediterranean Fever (FMF), the prototype of a group of inherited inflammatory disorders was found to be associated with some mutations in the FMF gene (MEFV). In this present study, we examined the frequencies of three MEFV mutations (M694V, V726A and E148Q) in our patients with IBD and their effects on disease behavior.

Patients and Method: Eighty two UC patients, 95 CD patients and 186 normal individuals were analyzed. The tested individuals were screened for the most common five MEFV mutations (M694V, V726A and E148Q) by molecular genetic studies using polymerase chain reaction with the ARMS (amplification refractory mutation system) and restriction fragment length polymorphism method.

Results: The most common mutation was E148Q in both UC and CD groups (17% and 10.5% respectively). This was followed by M694V mutation (4.8% in CD, 6.3% in UC) and V726A (2.5% in CD and 1% in UC). One CD patient (1.2%) and two UC patients (2.1%) were found to be homozygous for M694V mutation. Moreover, one UC patient (1%) was homozygous for the E148Q mutation. The number of homozygous patients for these mutations did not differ between the groups. In the control group, there were no homozygous MEFV mutations. In all groups, no individual was compound heterozygous for any combinations of the mutations. None of the MEFV mutations frequencies was significantly different between the groups.

Conclusion: Our findings suggest that the presence of MEFV mutations modifies disease severity rather than being a candidate gene for IBD.

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Knowledge of the investigation and management of patients who have undergone pouch surgery is lacking in gastroenterology trainees


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Introduction: Managing patients with ulcerative colitis forms a significant part of the workload for gastroenterologists, one-third will eventually require surgery. Restorative proctocolectomy (RPC) is the operation of choice. Unfortunately many patients continue to suffer from IBD in the form of pouchitis and cuffitis and other problems following surgery. Gastroenterologists are increasingly involved in the care of these patients.

Method: We aimed to assess the knowledge and confidence of the management of these patients in gastroenterology trainees. 56 of 63 gastroenterology Specialist registrars (SpRs) from three regions completed anonymous questionnaires relating to their perceived knowledge and confidence in the management of RPC patients. We excluded those trainees without national training numbers.

Results: Of the SpRs participating eleven were year 1, nine were year 2, five were year 3, sixteen were year 4, nine were year 5, in two the year was not given. 5% felt confident to discuss the advantages and disadvantages of RPC with patients. 57% had been involved in the care of RPC patients. 40% had performed and 7% observed a flexible pouchoscopy. 9% felt confident to investigate RPC patients with pouch dysfunction. 74% had never attended a teaching session on RPC, 88% however would like to do so.

Conclusion: A good understanding of the investigation and management of RPC patients is essential to all gastroenterologists involved in the care of IBD patients. This survey demonstrates that current knowledge of the investigation and management of RPC patients in gastroenterology trainees is suboptimal, further training is warranted and would be welcomed by this group. This may improve patient care.