Conclusions: Anxiety and depression impacts negatively in the quality of life in Mexican patients with IBD. The Mexican version of HADS had good internal consistency and external validity, with favourable sensitivity and specificity for identifying cases of anxiety and depression in patients with IBD.

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Relationship between the concentrations of free sulphates and 5-hydroxyindoleacetic acid (5-HIAA) in urine for IBD patients

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Background: Sulphates are sparingly soluble salts of sulfuric acid, the increase of which may indicate the presence in patients inflammatory bowel disease (IBD). Recent findings demonstrate a possible role of sulphated compounds in the aetiology of IBD, where the latter is characterised by specific changes in 5-hydroxytryptamine metabolism. The aim of our study was to assess the level of sulphate in the urine in patients with IBD and to study relationship between the sulphate and 5-hydroxytryptamine metabolisms.
Methods: The study included 40 patients with IBD. Urine samples from patients with ulcerative colitis (UC) and Crohn's disease (CD) taken twice in the morning and afternoon, were used for the analysis of free sulphates and 5-hydroxyindoleacetic acid (5-HIAA) with specific detection strips and ELISA, respectively.
Results: Among 40 patients, UC was detected in 26 (65%) (10 male and 16 female), CD – in 14 (35%) (7 males and 7 females). The average age was 37.2 years. The clinical characteristics of the patients were analysed. According to the severity of disease: mild, 8 patients (20%); moderate, 17 (42.5%); severe, 15 (37.5%). Among UC patients, total colitis was observed in 15 (58%) patients, left-sided colitis in 7 (27%), proctitis in 4 (15%). Among patients with CD, colitis was observed in 6 (43%), ileocolitis in 5 (36%), terminal ileitis in 3 (21%) patients. Extraintestinal manifestations of IBD were detected in 21 (52.5%) cases, of which arthropathy in 13 (61.9%), aphthous stomatitis in 5 (23.9%), spondylitis/sacroiliitis 3 (14.2%). Complications (intestine perforation, bleeding, strictures, toxic dilatation) were identified in 7 (17.5%) patients. The average level of sulphates in patients with IBD was 746.3 ± 45.0 mg/l, while in patients with UC 690.4 ± 57.0 mg/l and in patients with CD 850.0 ± 66.9 mg/l. There were no differences between the level of sulphates in the urine of patients with UC and BK (p = 0.09). However, compared with the level of sulphate in the urine of a healthy population (<400), an increase in the sulphate content was found in both patients with UC and patients with CD. We found clear correlations (R > 0.72, p < 0.01) between the concentrations of free sulphates from the 'morning' urine and 5-HIAA from the 'afternoon' urine. CD patients demonstrated stronger (R > 0.77 vs. R > 0.72) correlation compare to UC patients in both sets of experiments.
Conclusions: Patients with IBD have a higher urine sulphate level than healthy populations, which can be useful as an indirect sign of inflammation. Our data suggest a close relationship between sulphate and 5-hydroxytryptamine metabolism.

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Diagnostic approach to monogenic inflammatory bowel disease in clinical practice: a 10-year multicentric experience

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Background: Up to 15% inflammatory bowel diseases (IBD) rising before the age of 6 years, defined as very-early-onset IBD (VEO-IBD), may have a monogenic disease. More rarely monogenic defects are found in later onset IBD. Monogenic IBD are associated with high morbidity and mortality and timely genetic diagnosis is essential for adequate treatment. Due to the wide phenotypic and genetic heterogeneity of these conditions, it is often difficult to reach a genetic diagnosis and the best diagnostic approach is still debated. Next-generation sequencing (NGS) techniques have been proposed as a screening tool especially in patients with poorly defined phenotypes. In a cohort study that included patients with VEO-IBD and early-onset IBD with severe/atypical phenotypes (EO-IBD s/a) we aimed to describe the genetic diagnoses and their therapeutic implications, define the clinical characteristics associated with monogenicity, and suggest a diagnostic approach to monogenic IBD.
Methods: Clinical information of patients with VEO-IBD and EO-IBD s/a referred to 2 Italian Centers (IRCCS Burlo Garofolo and Ospedale Bambino Gesù) for a genetic work-up over 10 years (2008–2017) were collected. From 2015 newly diagnosed patients and patients without a previous genetic diagnosis were screened using NGS, except patients with disease-specific features in whom candidate gene analysis was chosen.
Results: In total, 93 patients were collected and 14 (15%) reached a genetic diagnosis. Selective sequencing was performed in 46 patients (49%), NGS in 84 patients (90%). Causative defects were revealed by NGS in 5 patients (NOD2, TTC37, DKC1, XIAP, FERMT3) and candidate sequencing in 9 patients (3WAS, CYBA, CYBB, FOXP3, 2CD40L, XIAP). In 8 of 9 patients diagnosed with candidate sequencing, the analysis was guided by the presence of disease-specific features. One patient, with unspecific presentation, underwent sequential sequencing of multiple genes over 15 months before reaching the diagnosis (XIAP). NGS identified a new NOD2 mutation previously missed with single-gene approach. Genetic diagnosis impacted patient management in 9 patients (64%): 8 underwent bone marrow transplant (2XIAP, 3WAS, 2CD40L, FOXP3) and 1 patient introduced danazole (DKC1). Patients with monogenic IBD more frequently had thrombocytopenia (21% vs. 3%; p = 0.003),

hemophagocytosis (21% vs. 3%; $p = 0.02$), extraintestinal symptoms (100% vs. 32%; $p < 0.001$) and disease onset ≤ 1 month of life (36% vs. 1%; $p < 0.001$) when compared with the non-monogenic group.

Conclusions: We suggest using NGS in all patients presenting with non-specific clinical profiles and selective gene sequencing when clinical characteristics suggestive of specific monogenic conditions are present.

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Raised faecal calprotectin in inflammatory bowel disease (IBD) patients: 100% accurate or potential red herring?

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Background: Faecal biomarkers of gastrointestinal inflammation have appeared in the past decade, of which calprotectin, a neutrophil cytosolic protein, has been studied the most. Faecal calprotectin (FC) is increasingly being used in clinical practice as surrogate marker for intestinal inflammation. A meta-analysis of prospective studies using suspected IBD patients found the pooled FC sensitivity and specificity to be 93% and 96%, respectively. Previous studies showed that several medications, dietary supplements, sampling time, pregnancy, and body mass index have been mentioned as confounding variables affecting results. Single FC measurement may not be sufficiently accurate to evaluate gastrointestinal symptoms, and different biomarkers such as albumin and C-reactive protein, disease activity indices such as Harvey–Bradshaw index and Mayo score with or without endoscopic investigation should be used to interpret the full clinical context. The primary study aim is to assess the prevalence of this subgroup cohort and assess sensitivity and specificity of FC in our department. This subgroup identification may have clinical impact on provision of colonoscopy service if statistically significant.

Methods: This retrospective analysis study involved obtaining results of FC samples taken and correlate with colonoscopic and histological findings. The FC samples in our institution were processed in two external labs (Biomnis, Ireland, and Birmingham, UK).

Results: Our study cohort involved 80 patients (43 females, 37 males). The median age was 44. There were 38 patients with Crohn's disease, 35 with ulcerative colitis, 5 indeterminate, and 2 newly diagnosed IBD. The FC range in our external lab (Biomnis) are subdivided into 3-negative for level <50 µg/g, between 50 and 200 grey zone, and >200 is positive whilst the laboratory in Birmingham used the cut-off FC level < 60 µg/g as negative. There were 64 patients (80%) who had raised FC results. Of these, 55 (86%) had findings of colitis on histology and 9 (14%) showed negative histology ($p = 0.01$, CI 95%). There were 13 (16.3%) patients who had normal FC and had colonoscopy performed which showed colitis findings and confirmed histologically. There were 3 patients (3.7%) who had normal FC with no colitis evident endoscopically and histologically.

Conclusions: Faecal calprotectin is utilised in IBD centres as surrogate markers and initial non-invasive screening for intestinal inflammation. The FC specificity and sensitivity is variable and the possibility of confounding variables and patients' factors should be taken into account when interpreting results.

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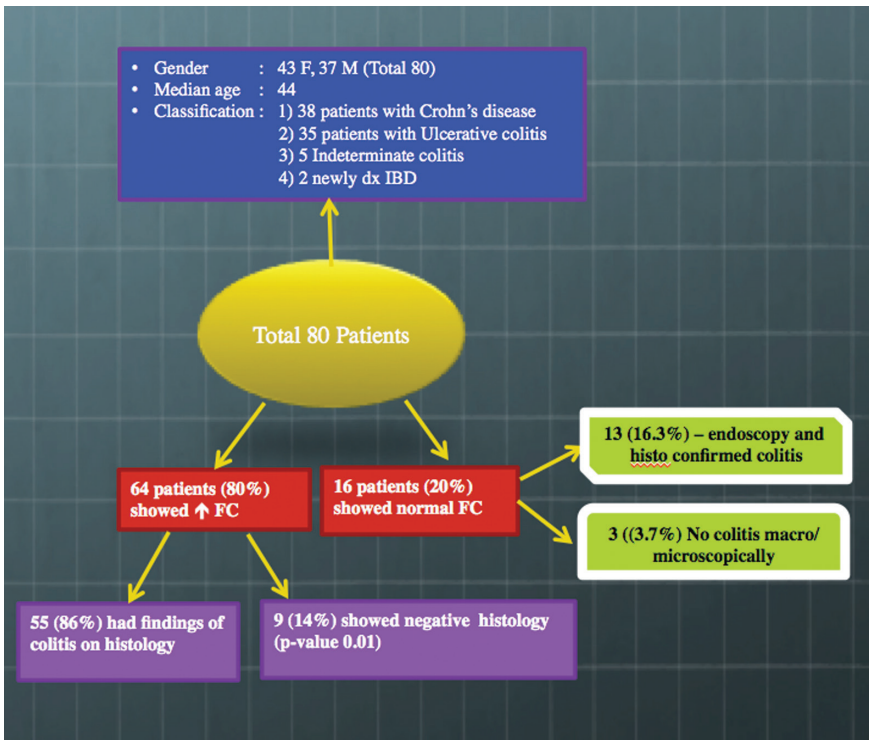

Faecal calprotectin results breakdown.