screened the allelic frequencies of $(GT)_n$ repeats in the HO-1 gene promoter in these obese children. NAFLD was determined through liver ultrasonography. Because the distribution of numbers of $(GT)_n$ repeats was bimodal, we divided the alleles into 2 subclasses: class S included shorter ($_n$ repeats in HO-1 gene promoter on paediatric NAFLD.

Results Of 101 obese subjects, 27 (26.7%) had NAFLD. The alanine aminotransferase (ALT) level was higher in patients carrying L alleles (L/L and L/S) than patients with S alleles (S/S) [46.2 ± 49.3 IU/L versus 30.2 ± 20.1 IU/L]. The significant risk factors for paediatric NAFLD were patients carrying L alleles (L/L and L/S) (11.613; 95% CI, 1.2 to 112.43; p = 0.034), and HOMAR-IR (1.37; 95% CI, 1.05 to 1.79; p = 0.019).

Conclusions In this hospital-based study, the obese children with larger GT repeats and insulin resistance were susceptibility to have NAFLD.

PS-075 GENETIC COUNSELING OF WILSON DISEASE IN EGYPTIAN CHILDREN: EVALUATION OF ATP7B GENE MUTATION AS A USEFUL TOOL

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Backgrounds and aims Wilson disease (WD) is an autosomal recessive disorder of copper transport, ATP7B, the gene mutated in WD, consists of 21 exons and encodes a 1,465 amino acid protein representing a copper transporting P-type adenosine triphosphatase (ATPase). Our aim was to evaluate His1069Gln mutation in exon 14 of ATP7B gene as a useful tool in genetic counseling.

Methods This study was conducted on 20 index cases with WD (group I) and their siblings (group II, n = 27), 47 apparently healthy children as control group (group III). Diagnosis of WD was made if the patient had hepatic and/ or neurologic disease in addition to at least two of the following six criteria: Positive family history of WD, Low ceruloplasmin level <20 mg/dl., Presence of kyser- Fleisher ring, Liver biopsy suggestive of WD, Elevated 24 hour urinary copper excretion and finally Coomb's negative hemolytic anemia. Molecular testing for ATP7B (His1069Gln) done by PCR based on restricted fragment length polymorphism assay.

Results It was found that the frequency of the His1069Gln gene mutation was 25% among index cases group, 14.8% among siblings of the index group. As regard genotype- phenotype association there was no statistically significant difference between different types of disease phenotype regarding the mean age of onset, gender and presence of ATP7B gene mutation.

Conclusions The His1069Gln of ATP7B gene mutation could be used as a detection tool for WD in high risk population.

PS-076 NOVEL MUTATIONS UNDERLYING RARE FAMILIAL ENTEROPATHIES

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Background Microvillous Inclusion Disease (MVID) and Congenital Tufting Enteropathy (CTE) are congenital disorders of the intestinal epithelial cells that cause an intractable diarrhoea since birth. MVID and CTE are very rare disorders and inherited as autosomal recessive traits. Recently, mutations of *MYO5B* and *EpCAM* were identified as the underlying lesion resulting in MVID and CTE, respectively.

Method Four Saudi families were investigated, three with children affected by MVID and one with a child affected by CTE. Five patients and available unaffected individuals were subjected to genome-wide homozygosity scans using the Affymetrix 250K SNP array. Analysis with the copy number tool CNAG identified shared homozygous regions unique to the affected subjects.

Results Of the three families with MVID, homozygosity was observed in two families at a locus on chromosome 18 which included *MYO5B*. Sequencing of *MYO5B* in individuals from these families identified two novel nonsense mutations in exons 24 and 36 (Q1047X and E1589X). In the other family homozygosity was absent at the *MYO5B* locus. However, a locus on chromosome 2 which included *EpCAM* was found to be homozygous in this family. Sequencing of *EpCAM* identified a 1 bp insertion (c.499insC) in exon 5 resulting in premature truncation of the mature protein. This was consistent with this family being classified as having CTE rather than MVID. The fourth family studied was referred with a diagnosis of CTE and was found to have the same 1 bp insertion consistent with a common founder.

Conclusion We have identified novel nonsense mutations in *MYO5B* and *EpCAM* associated with congenital enteropathies in Saudi families. Our findings expand the limited spectrum of *MYO5B* and *EpCAM* mutations associated with gastrointestinal genetic disorders and provide an opportunity to investigate phenotype/ genotype correlations.

PS-077 THE IMPLICATION OF IL-6 572 C/G GENE POLYMORPHISMS IN CHILDREN'S MALNUTRITION

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Malnutrition is a disorder caused by reducing the energetical and/or protein input, with adverse effects on tissue and body form and function and different clinical consequences.

Aim

The aim of this study was to establish the correlations between the polymorphisms of the IL-6 572 gene in malnourished children.

Methods We assessed 283 hospitalised children divided into two groups: group I (control) included 110 patients with normal nutritional status, and group II consisted of 173 malnourished patients.

The two groups underwent IL-6 572 C/G polymorphism testing, measurement of anthropometric parameters (mid-upper arm circumference, MUAC and tricipital skinfold thickness, TST), and paraclinical evaluation (protein, albumin).

Results We observed that the differences between the anthropometric parameters (MUAC and TST) corresponding to the three types of genotypes (CC, CG, GG) were statistically significant lower in malnourished children with all the three genotype of IL–6 572 (p = 0.0001). Same correlations were noticed with low serum albumin levels.

Conclusion In children with malnutrition BMI, MUAC, TST and low serum albumin levels correlated with genotype GG and CG of the *IL-6* 572 gene.

PS-078 CLINICAL RELEVANCE OF GAMMA-GLUTAMYL TRANSPEPTIDASE IN CHILDHOOD OBESITY

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Background and aims Metabolic risk leads to severe comorbidities in obesity. We evaluate the relationship between the values of gamma-glutamyl trans peptidase (GGT), a marker of hepatic involvement, and cardio metabolic risk factors in obese children.
Methods A prospective cross-sectional study of 147 children (aged 7 to 16 years) was carried out. Ninety-five children were obese with a body mass index standard deviation score (SDS-BMI) >2 and 52 children were normal weight. Patients with endocrine disease or syndromic obesity were excluded. We have analysed clinical parameters of adiposity (fat mass by bioelectrical impedance, waist and hip circumference), blood pressure, and classical biochemical parameters indicative of metabolic risk (lipid profile, glucose and insulin). Additionally, novel parameters related to metabolic risk such as uric acid, retinol binding protein (RBP4), cystatinC, homocysteine, thyrotropin, ultrasensitive C-reactive protein (CRP) and GGTwere also determined. Statistical analysis was made ANCOVA test and Pearson partial correlation adjusting for gender, age, Tanner stage, and BMI.
Results GGT was higher in the children with SDS-BMI >4 with respect children with SDS-BMI between 2 and 4(16.3 ± 5.8 *vs* 18.4 ± 8.8 IU/L, p = 0.025). Both groups were statistically significant with respect normal weight (12.2 ± 2.9 IU/L, p < 0.0001 and p < 0.001 respectively). GGT was correlated with SDS-BMI (p < 0.0001), waist circumference (p < 0.001), percentage of fat mass (p < 0.01), SDS of systolic blood pressure (p < 0.010), total cholesterol (p < 0.0001), LDL cholesterol (p < 0.0001), triglycerides (p < 0.0001), RBP4 (p < 0.047), thyrotropin (p < 0.019) and CRP (p < 0.044).
Conclusion GGT is a marker associated with several metabolic risk factors, which highlights the importance of considering hepatic impairment as a component of this syndrome.

PS-079 WITHDRAWN

PS-080 PREVALENCE OF OVERWEIGHT IN PAEDIATRIC INFLAMMATORY BOWEL DISEASE IN SAUDI ARABIA

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Background and aim Excess weight in inflammatory bowel disease (IBD) represents an additional morbidity, and yet the prevalence has been rarely reported. The aim of this report is to establish the prevalence of overweight in children with IBD in the Kingdom of Saudi Arabia (KSA).
Methods Data from a cohort of children in the KSA diagnosed with IBD were analysed retrospectively. Growth parameters were recorded at diagnosis and body mass index (BMI) was calculated using the formula (weight/height^2). The KSA charts were used as reference. Excess weight categories were defined as overweight (BMI-for age ≥85th to <95th), obesity ≥95th to <97th), and severe obesity ≥97th percentile. Chi-square test was used and p-value of <0.05 was considered significant.
Results There were 417 children from birth to 18 years of age, including 133 ulcerative colitis (UC) (32%), and 284 Crohn disease (CD) (68%). The prevalence of excess weight was 12/133 (9%) in UC and 23/284 (8.1%) in CD (p = 0.063) much lower than in Western reports. However, the more common prevalence of excess weight in UC than CD, although not significant (p = 0.063), was similar to patterns from other population. The commonest form of excess weight was overweight 20/35 (57%), followed by obesity 9/35 (26%), and severe obesity 6/35 (17%).
Conclusion The pattern of excess weight in KSA children with IBD is similar to Western literature. However, a much lower prevalence is demonstrated. Identification of factors associated with the low prevalence of overweight and obesity is needed.

PS-081 LACTOBICILLUS ACIDOPHILUS ATTENUATED SALMONELLA-INDUCED INTESTINAL INFLAMMATION VIA TGF-BETA/SMADS SIGNALLING

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Aims To investigate whether probiotics and/or prebiotics attenuate *Salmonella typhimurium* induced NF-κB activation via Smad7 and IκBα expression in the human colorectal epithelial $CaCO_2$ cells; to determine the molecular mechanisms of preventive effects of probiotics on intestinal infection.
Material and methods $CaCO_2$ cells were administered probiotic (*Lactobicillus acidophilus*) and/or prebiotic (inulin supplemented with oligofructose). Subsequently, the cells were infected with *S. typhimurium*. The culture supernatants and cell lysates were collected for cytokine determination and western blot analysis. The $CaCO_2$ cells were also transfected with plasmids containing Smads or NF-κB responsive reporter luciferase. After transfection, supernatants from cells were collected for luciferase assay. Involvement of miR-21 (Smad7 silencer) from supernatants of infected cells in the presence or absence of probiotics was determined.
Results The probiotics significantly suppressed NF-κB activation elevated by *S. typhimurium*. IL-8 mRNA was significantly lower in probiotics pretreated $CaCO_2$ cells compared with the cells infected with *S. typhimurium* alone. Synbiotics showed strongly suppressed effects on IL-8 and TNF-α gene transcriptions elevated by *S. typhimurium*. Pretreatment of probiotics increased IκBα expression level. Consistent with IκBα expression, pretreatment of probiotics increased 7 folds of Smad3/4 activity. The protein expressions of TGF-β and Smad7 in *S. typhimurium* infected cells with or without probiotics were determined by immunoblotting. Compared to *S. typhimurium* infection alone, pretreatment with probiotics and synbiotics induced 20 and 4 folds of miR 21 expressions, respectively.