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LIII REUNIÓN ANUAL DE LA
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XI REUNIÓN ANUAL DE LA
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doscopy to the Hospital de Gastroenterología "Dr. Carlos Bonorino Udaondo". Histopathology and *H. pylori* diagnosis were evaluated from gastric biopsies. Serum ghrelin concentration, PGI and PGII were measured by ELISA. Kruskal-Wallis and Mann Whitney tests were applied. Thirty-five individuals were included to date, 77.1% female, with an age of 40.2±13.0y. *H. pylori* prevalence was 71.4% (CI95% 54.9-83.7%). Median ghrelin levels were 396.0 pg/ml (IQR 315.0-501.0); PGI, 45.1 ng/ml (IQR 29.5-55.7); PGII, 3.3 ng/ml (IQR 2.0-6.0); PGI/PGII 11.0 (8.0-16.7). PGI and PGII were significantly higher for *H. pylori* infected compared to non-infected patients (*P*=0.014 and *P*=0.0001, respectively); however, PGI/PGII ratio did not differ between both groups (*P*=0.14). Serum PGII differed significantly between normal mucosa, chronic inactive and active gastritis of the antrum and corpus (*P*=0.003 and *P*=0.049, respectively), being higher in the presence of gastric pathology. These preliminary results suggest that the measured biochemical variables might be useful as potential biomarkers of gastric pathology; further analysis after inclusion of the total sample size for this protocol is needed to confirm these findings.

121. (582) **BENEFICIAL PROPERTIES OF PASSIFLORA CAERULEA ON INTESTINE IN STRESS MODEL INDUCED BY IMMOBILIZATION IN RATS**

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Irritable bowel syndrome (IBS) is a prevalent gastrointestinal disorder that has been related with psychological factors. There is growing evidence that IBS or IBS-like symptoms could be a prodrome before diagnosis of Inflammatory Bowel Disease or can be observed in IBD patients in remission. Its treatment is challenging and use of alternative medicines, especially herbal therapies is increasing. In this sense, *Passiflora caerulea* (PC), a native plant that previously showed to possess beneficial properties in preclinical models related to Inflammatory Bowel Disease was studied. Male Sprague Dawley rats weighing 210 –230 g were used. Four groups of animals (n=6) were treated with PC (250 and 500 mg/Kg), clonazepam (Clo) 1 mg/Kg or water during four days (S). In the fifth day these groups were restrained using a plastic rodent restrainer. Control group (C) were not subjected to stress. Immediately after 6 hours, blood was collected and colon and ileon were removed for posterior analysis. Exposure to acute immobilization produced an increase in plasmatic corticosterone level (S: 487.4±193.8 ng/mL, C: 118.2±28.9) and a slight increase in blood glucose. These effects were reduced by treatment with PC (250 mg/Kg: 195.5±26.5, 500 mg/Kg: 74.25±14.4) and reference drug (Clo: 244.0±20.1). Redox status expressed as GSH:GSSG ratio was increased in ileon and colon in S groups, meanwhile the extract significantly reduced this effect only in ileon. Histopalogical analysis showed that goblet cells were depleted by immobilization in ileon and colon and both doses of extract in ileon and only higher dose in colon ameliorated these effects. Western blot analysis showed that P53 and caspase-3 proteins were increased in ileon and colon of stressed animals, meanwhile PC reduced both parameters in both portions of intestine. These observations suggested that PC could represent a promising treatment to improve gastrointestinal health in stressful conditions.

GENÉTICA

122. (037) **GJB2 AND GJB6 GENETIC VARIANT CURATION IN A NON-SYNDROMIC HEARING LOSS COHORT FROM ARGENTINA**

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Hereditary hearing impairment affects 1-500 newborn children. It is characterized by the large number of genes involved (more than 100) and its phenotype heterogeneity. Despite the wide genetic variety of hearing impairment, the most commonly mutated genes in severe to profound autosomal recessive non-syndromic hearing loss are *GJB2* and *GJB6*, accounting for nearly 50% of the cases in most populations around the Mediterranean Sea. Molecular diagnosis enables proper genetic counseling and medical prognosis to patients. Therefore, correct interpretation of the phenotypic consequences of genetic variants is crucial in genetic diagnosis, since discrepancies in sequence variant interpretation and classification has been reported to lead to serious impact in patient health maintenance.In this study we aimed to identify the genetic causes of hearing loss and performed a manual genetic variant curation following the American College of Medical Genetics and Genomics/Association for Molecular Pathology ACMG/AMP standards and hearing-loss-gene-specific criteria of the ClinGen Hearing Loss Expert Panel.A total of 600 patients were studied for genetic variants in GJB2 and *GJB6* genes by Sanger Sequencing technique and Multiplex Gap-PCR, respectively.Overall, 48 different sequence variants were detected in our cohort of patients, being the c.35delG the most common causative variant identified. Besides, more than 50% of sequence variants were reclassified from their previous categorization in ClinVar after careful manual analysis. These results provide an accurately analysed and interpreted set of variants to be taken into account by clinicians and the scientific community, and hence, aid the precise genetic counseling to patients.

123. (184) **ANALYSIS OF TP53 ABERRATIONS IN PATIENTS WITH CHRONIC LYMPHOCYTIC LEUKEMIA**

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Chronic lymphocytic leukemia (CLL) is the most common form of adult leukemia in the Western world. *TP53* (*Tumor protein P53*) gene deletions and mutations constitute important prognostic and predictive factors associated to poor outcome in CLL, with relevance in clinical decisions. We evaluated *TP53* alterations in 218 CLL patients (131 men; mean age: 64.9 years, range: 35-87 years; RAI clinical stages: 0: 36%; I-II: 47.1%; III-IV: 16.9%), to analyze their frequency, distribution and association with prognostic factors of the disease. PCR and bidirectional Sanger sequencing to evaluate IGHV (*immunoglobulin heavy variable region*) and *TP53* mutational status were used. Cytogenetic and FISH (*Fluorescence in situ hybridization*) analysis were performed. The study was approved by the Local Ethics Committee. All individuals gave their informed consent. Twenty-nine patients (13.3%) had *TP53* aberrations. Deletion 17p13 [del(17p)] was observed in 26 cases (12%); 3 patients showed only *TP53* mutations (*TP53*-M), and 10 cases had *TP53* deletion and mutation. Two deletions and two insertions (3 frameshifts) (26.7%), and 11 point mutations (73.4%) (1 at the splicing site and 10 missense), were found; 86.6% variants were located in the DNA-binding domain (exons 4-8). Cases with *TP53*-M showed significantly higher mean percentages of leukemic cells with del(17p) (33.5%) than those with unmutated *TP53* (12%) (p=0.016). *TP53*-M patients showed association with unmutated IGHV (87.5% cases) and chromosome alterations (88.9%), including complex karyotypes. Similar clinical behavior between cases with del(17p)