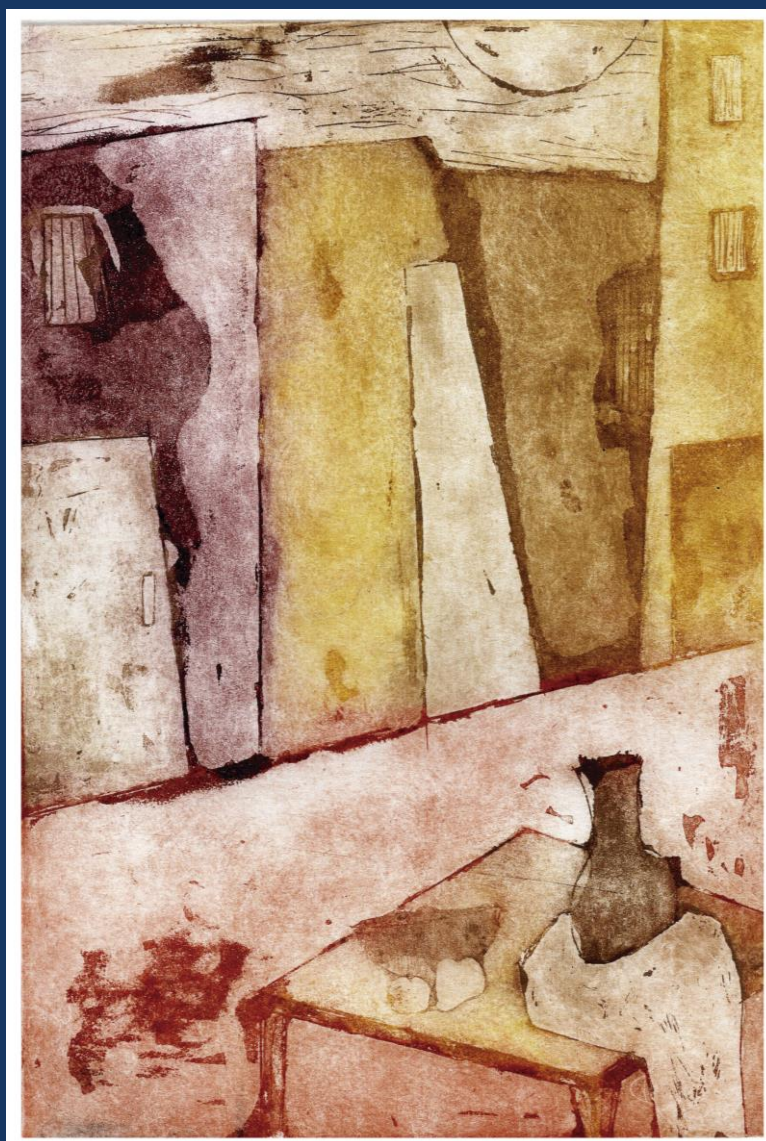


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# medicina

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La Tapa (Ver pág. 4)  
**Atardecer en la tarde**  
Antonella Ricagni

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**Abstract/Resumen:** Previous studies in a group of adults who belong to the Project for Protection of Vulnerable Population against Chronic Non-communicable Diseases (PROTEGER) of Ministry of Health, demonstrated that 65% of participants presented three or more associated risk factors, such as glycemia, cholesterol, triglycerides, blood pressure, fat mass and salt intake; being fat mass increased in 86 %. In this work, the aim was to assess less frequently evaluated risk factors in a group of older adults such as fat mass and salt intake. A descriptive study was conducted in 14 women (W) (75.3 ± 9.9 y) and 10 men (M) (77.1 ± 8.8 y) who had signed an informed consent. These subjects had either attended the Primary Health Care Center Pueblo Nuevo of Basavilbaso (Entre Ríos province) or reside at nursing homes for elderly in Buenos Aires province. Body weight (BW, kg) and height (H, m) were determined to calculate body mass index (BMI= BW/H<sup>2</sup>, kg/m<sup>2</sup>). Fat-free mass (FFM, kg) was evaluated by deuterium dilution technique and fat mass (FM, kg) was obtained as FM= BW-FFM. Sodium, potassium and creatinine excretion were determined from spot urine and the 24 h urinary sodium excretion was estimated using the INTERSALT equation. The results showed that 93% of W and 50% of M were overweight or obese. Moreover, FM % was increased in 78.6 % of W (38.1 ± 9.3) and 60 % of M (24.1 ± 10.8). In addition, 58.3 % of participants presented salt intake higher than the WHO recommendation of 5 grams/day (6.4 ± 2.9) with a decreased K/Na ratio (0.8 ± 0.7) in 79.2 %. An increase in fat mass and salt intake was observed in this group of older adults and findings showed that both risk factors were presented in 50 % of the cases. The assessment of these less frequently evaluated risk factors would contribute as a tool for better diagnosis of the chronic non-communicable diseases.

### **0356 - NOVEL FMO3 MUTATIONS INVOLVED IN TRIMETHYLAMINURIA DISORDER**

**Sofia STUPNIKI** (1) | Leonardo DIONISIO(1) | Eugenio AZTIRIA(1) | Maximiliano ALDA(2) | Makiko SHIMIZU(3) | Hiroshi YAMAZAKI(3) | Guillermo SPITZMAUL(1)

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**Abstract/Resumen:** Trimethylaminuria is a human autosomal recessive disorder due to mutations in the gene of the enzyme flavin-containing monooxygenase 3 (FMO3). In this condition, odorous trimethylamine (TMA) cannot be converted into its non-odorous N-oxide. Consequently, TMA accumulates and is excreted through urine and sweat being responsible of the characteristic malodor of the patients. Clinical diagnosis is based on the quantification of TMA in urine samples and is confirmed by genetic analysis which additionally contributes to risk stratification. None of them are performed in Argentina, where diagnosis is based on the body odor. The aim of this case study was to correlate the clinical diagnosis with genetic variants of FMO3. The index patient is a female (12 y.o.) with fishy malodor since she was a toddler and with an exacerbation of the condition since puberty. Symptoms are only controlled by strict diet restriction. The patient and unaffected direct relatives were analyzed to build up and evaluate the genetic pedigree. gDNA was obtained from jaw swabs. We used specific primers designed for the coding exons (2 to 9) to amplify them by PCR followed by sequencing. The mutant enzyme activity was tested using

heterologous expression. The index case carried 2 SNPs of the FMO3 gene: c.472G>A (E158K) and c.923A>G (E308G). She also carried two novel mutations resulting in amino acid substitutions: P73L and F140S. All of them were in heterozygosity. The mother carried the P73L/E158K/E308G allele and the father the F140S allele. Functional analyses showed a 50 and 90 % decrease of the catalytic capacity for the mother and the father alleles, respectively (n= 3). Based on the family pedigree, we identified a compound heterozygous patient for the two novel point mutations. Functional analysis demonstrated a drastic reduction in enzyme activity for each allele, which combined with the already known changes promote the severe condition exhibited by the patient.

### **0428 - DECREASED HDL-MEDIATED CHOLESTEROL EFFLUX AND ANTIOXIDANT ACTIVITY IN OBESE CHILDREN AND ADOLESCENTS**

**Maximiliano MARTIN** (1) | Laura GAETE(2) | Viviana OSTA(2) | Walter Francisco TETZLAFF(1) | Eliana Elizabeth BOTTA(1) | Florencia FERRARO(1) | Ezequiel LOZANO CHIAPPE(1) | Laura BOERO(1) | Liliana TRIFONE(2) | Fernando BRITES(1)

**UNIVERSIDAD DE BUENOS AIRES. FFYB. DEPTO DE BIOQUÍMICA CLÍNICA. LAB. DE LÍPIDOS Y ATROSCLEROSIS. (1); HOSPITAL GUTIERREZ (2)**

**Abstract/Resumen:** The presence of obesity during infancy is associated to the development of related complications such as dyslipidemia, a well-known risk factor for cardiovascular disease. High density lipoproteins (HDL) represent the only antiatherogenic fraction and are responsible for the promotion of cholesterol efflux from macrophages and the inhibition of low density lipoprotein (LDL) oxidation. The aim of present work was to evaluate HDL capacity to promote cellular cholesterol efflux and the activity of two HDL-associated proteins, cholesteryl ester transfer protein (CETP) and paraoxonase 1 (PON1), in 25 obese children and adolescents and 20 healthy controls. There were no differences in age or sex between the groups. Triglycerides (TG), total cholesterol (TC), HDL-C, LDL-C, apolipoproteins (apo) A-I and B, glucose and insulin were measured by standardized methods. HOMA-IR was calculated. The human monocyte line THP-1 was employed to determine cellular cholesterol efflux. CETP was analysed by a radiometric method and PON1 employing two substrates: paraoxon (PON activity) and phenylacetate (ARE activity). The obese children and adolescents showed higher triglycerides, LDL-C and HOMA-IR (p<0.01), in addition to lower HDL-C and apo A-I levels. Both cholesterol efflux (6.0 ± 1.6 vs. 7.6 ± 2.1 %; p<0.01) and ARE activity (94 ± 12 vs. 103 ± 19 µmol/ml.min; p<0.05) were significantly lower in the obese group compared to healthy controls. Cholesterol efflux correlated with BMI-z (r= -0.43; p<0.05) and TG (r= -0.28; p<0.05). ARE correlated with HDL-C (r= 0.38; p<0.05), apo A-I (r= 0.37; p<0.05) and PON (r= 0.48; p<0.05). Obese children and adolescents presented insulin resistance and a more atherogenic lipid profile in addition to lower capacity to promote cellular cholesterol efflux and ARE activity, a reflex of HDL antioxidant capacity. These findings would be indicative of functionally altered HDL particles with decreased atheroprotective potential.

### **0434 - ABNORMAL HDL QUALITY AND CAPACITY TO ACQUIRE PHOSPHOLIPIDS IN PATIENTS WITH RHEUMATOID ARTHRITIS: EFFECT OF TOFACITINIB AND TOCILIZUMAB.**

**Eliana Elizabeth BOTTA** (1) | Florencia PIERINI(2) | Maximiliano MARTIN(1) | Walter Francisco TETZLAFF(1) | María Soledad SAEZ(2) | Osvaldo CERDA(3) | Ignacio GANDINO(2) | Gustavo CITERA(3) | Laura BOERO(1) | Javier ROSA(2) | Patricia SORROCHE(2) | Tomás MEROÑO(1) | Anatol KONTUSH(4) | Enrique SORIANO(2) | Fernando BRITES(1)

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