

# comunicazioni orali

## SESSIONE COMUNALE AMCLI-SIBioC

### Il laboratorio nella diagnostica cardiologica

Venerdì 17 Ottobre 2003, 10.00-13.00 Sala Michelangelo,  
Palazzo dei Congressi, piano -I

#### CO6.1

##### THYROID PROFILE EVALUATION AND POSTOPERATIVE ATRIAL FIBRILLATION

**Storti S., Cerillo A.G., Mariani M., Giannelli I., Fontani G., Fusani L., Parri M.S., Kallushi E., Clerico A., Biagini A.**

Istituto di Fisiologia Clinica del CNR -  
Ospedale "G. Pasquinucci", via Aurelia Sud, 54100 Massa

**Objective.** Increasing evidences suggest that thyroid metabolism has a major role on the prognosis of cardiac patients. Cardiac surgery is a well recognized cause of thyroid dysfunction. This study evaluates the role of thyroid profile evaluation in the preoperative assessment of cardiac surgery patients.

**Methods.** Free triiodothyronine (fT3), free thyroxine (fT4) and thyroid stimulating hormone (TSH) were assayed in 107 consecutive patients undergoing coronary artery bypass grafting (CABG) by the AxSYM® (Abbott Laboratories, Diagnostic Division, Abbott Park, USA) Microparticle Enzyme Immunoassay (MEIA); serum samples, collected in serum separator tubes, were immediately centrifuged and stored at -20°C till the analysis was performed. All the samples from the same patient were analyzed together in the same session in order to minimize the inter-assays variations.

**Results.** A strong correlation was found between low fT3 concentration at admission and postoperative atrial fibrillation (AF), that occurred in 33 (30.8%) patients. An older age ( $p=0.03$ ), no therapy with  $\beta$ -blockers ( $p=0.08$ ), chronic obstructive pulmonary disease ( $p=0.08$ ), lower left ventricle ejection fraction ( $p=0.09$ ), and lower fT3 concentration ( $p=0.001$ ) were all univariate predictors of postoperative AF. On multivariate analysis, low fT3 resulted to be the most important independent predictor of postoperative AF

(OR 4.425; 95% CI, 1.745-11.235;  $p=0.001$ ).

**Conclusions.** Our data show the existence of a strong, previously unrecognized, relationship between low fT3 at admission and AF after CABG. These finding remarks the importance of thyroid profile laboratory evaluation in patients undergoing CABG, in order to reduce postoperative AF risk.

**Reference.** Lein I, Ojamaa K. Mechanism of disease: thyroid hormone and the cardiovascular system. *N Engl J Med* 2001; 344-501-509

#### CO6.2

##### PREVALENCE AND CLINICAL PROFILE OF CARDIAC MYOSIN-BINDING PROTEIN C GENE MUTATIONS ASSOCIATED WITH HYPERTROPHIC CARDIOMYOPATHY IN TUSCANY.

**Girolami F°; Passerini I°; Mariottini A°; Olivetto J.\*; Cecchi F\*.; Vargiu D\*.; Torricelli F°.**

°Cytogenetics and Genetics Unit, Azienda Ospedaliera Careggi, Florence, Italy.

\*Myocardial Diseases, Azienda Ospedaliera Careggi, Florence, Italy;

**Objectives:** We studied the clinical and genetic features of hypertrophic cardiomyopathy (HCM) caused by mutations in MYBPC3 in 57 consecutive patients from Italy.

**Background:** HCM is an autosomal dominant disorder characterized by unexplained left ventricular hypertrophy, usually asymmetric and involving the interventricular septum. More than 130 mutations in nine different genes encoding sarcomere proteins have been

identified; studies on Japanese and French families have shown that HCM caused by MYBPC3 mutations may have a delayed onset and is usually benign. However, genotype-phenotype relationship in other populations remains unclear.

Methods: All 35 exons of MYBPC3 were screened by DHPLC followed by automatic sequencing in 57 consecutive patients with HCM. The diagnosis was based on two-dimensional echocardiographic identification of a hypertrophied, non dilated left ventricle, in the absence of another cardiac or systematic disease capable of producing the magnitude of wall thickening evident.

Results: Overall, we identified 8 MYBPC3 mutations in 7 patients (a 12% prevalence). Six mutations were novel (IVS18+1C>T, insT753, A522T, V771M, E165D, R1241L), and not detectable in 100 normal controls, thus excluding the possibility of polymorphisms; two (A522T and V771M) were found in heterozygosity in the same patient. The remaining two mutations have been previously reported (Q969X, IVS23+1A >G). Average age at diagnosis in the 7 patients was  $40.1 \pm 13.6$ ; echocardiographic features and outcome were variable. Six of the eight patients had a benign outcome and family history; one (E165D, male, age 42), is currently in NYHA functional class III due to end stage progression of the disease; and one (IVS18+1C>T, female, age 54) had severe functional limitation.

Conclusions: In a regional HCM population from Italy, MYBPC3 mutations were common and were associated with a broad spectrum of clinical and echocardiographic manifestations. Most of the mutations had not been previously described, including 2 occurring in the same patient.