High Blood Press Cardiovasc Prev 2007; 14 (3): 145-196
1120-9879/07/0003-0145/$44.95/0

Genetics and Pharmacogenomics

3.7 A Black Cardiac Paraganglioma in a Patient Carrier of SDHD Mutation

C. Letizia,[1] *D. Cotesta,*[1] *L. Petramala,*[1] *A. Verrienti,*[1] *S. Filetti,*[1] *A. Barletta,*[2] *S. Volpe,*[2] *F. Miraldi*[2]

(1) Department of Clinical Sciences, University of Rome "La Sapienza", Ro Rome; (2) Department of Heart Surgery, University of Rome "La Sapienza", Rome, It Italy

We report a case of a 69 year old woman admitted to our hospital with a worsening dyspnoea in the last three months. In the past she had surgical removal of mediastinic paraganglioma and a removal of paraganglioma of left carotid glomous; arterial hypertension since 15 years before with a satisfactory control of blood pressure with anti-hypertensive drugs.

A transthoracic echocardiography showed a mass (size 31 x 38 mm) adherent to back wall of right atrium, a MRI of heart confirmed the presence of this mass. Then, a surgery treatment was performed but the atrial mass have been incompletely removed because involved the sectal lembus of tricuspidal valve and superior wall of coronary sinus as far as atrio-ventricular junction; during the operation there was a transitory increase of arterial pressure (PA 180/100 mmHg, FC 92 bpm).The histopathological examination of the removed mass showed the feature of a black paraganglioma.

Because of personal history, the site and the histopathological features of the removed lesion paraganglioma/pheochromocytoma syndrome was suspected.

The genetic analysis revealed a an insertion into 4th exon of SDHD gene (444-445 ins ATCT) on etherozygosis, this alteration has not never been reported before. The diagnosis of paraganglioma/pheochromocytoma syndrome type 1 (PGL 1) (OMIM 168000) was made.