

**Analysis of Several BRCA1 and BRCA2 Mutations in a Hospital-based Series of Unselected Breast Cancer Cases**

T. Burcoş<sup>1</sup>, D. Cimponeriu<sup>2</sup>, D.A. Ion<sup>3</sup>, S. Spandole<sup>2</sup>, P. Apostol<sup>2</sup>, M. Toma<sup>2</sup>, I. Radu<sup>2</sup>, I. Popa<sup>1</sup>, S. Stanilescu<sup>1</sup>, E. Popa<sup>1</sup>

<sup>1</sup>Department of Surgery, Coltea Hospital, Bucharest, Romania

<sup>2</sup>University of Bucharest, Department of Genetics, Bucharest, Romania

<sup>3</sup>"Carol Davila" University of Medicine and Pharmacy, Bucharest, Romania

**Abstract**

*Background:* The distribution of BRCA mutations varies significantly between populations. The spectrum of BRCA1 and BRCA2 mutations in breast cancers in the Romanian population is incompletely known. The aim of the present study is to investigate the presence of nine BRCA mutations in patients with breast cancer identified in a surgical clinic from Bucharest.

*Methods:* Unrelated women diagnosed with breast cancer from Coltea Hospital (n=114) and healthy controls (n = 150) were selected for this study. Seven mutations in BRCA1 (185delAG, 5382insC, 943ins10, E1250X, 1294del40, E1373X, R1443X) and two in BRCA2 (IVS16-2A4G and 6174delT) were tested using PCR based protocols. In addition, the presence of BRCA1 185delAG, BRCA1 5382insC, BRCA2 6174delT mutations were tested with a post amplification mutation detection system, based on the ELISA method.

*Results:* Two patients with sporadic breast cancer (2%) and one patient with family history of the disease (7.14%) have the BRCA1 5382insC mutation. No other mutation was detected in patient and control groups. The mutations were not present in the control lot.

*Conclusions:* Our results indicate that BRCA1 5382insC is a common mutation in Romanian women with breast cancer (3/114).

**Key words:** breast cancer, BRCA mutations, BRCA1 5382insC, genetic test

Corresponding author: Emil Popa, MD, PhD

Department of Surgery, Coltea Hospital

I.C. Bratianu street, no 1, 3th district, Bucharest, Romania

E-mail: emilpopa@yahoo.com