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Session A. Breast cancer

A47 Inherited mutations in breast cancer susceptibility genes among a triple negative breast cancer cohort unselected for family history of breast cancer

B. Salvadori[1], A. Caligo[2], I. Montagnani[3], L. Spugnesi[2], A. Fontana[3], E. Ianducci[3], A. Michelotti[3], I. Ferrarini[3], I. Bertolini[3], C. De Angelis[3], S. Fancelli[3], E. Pfanner[3], A. Falcone[3]

[1]*UO Oncologia Universitaria Ospedale Santa Chiara Pisa, Pisa*
[2]*Laboratorio Genetica Medica AOUP Pisa, Pisa*
[3]*Polo Oncologico AOUP Pisa, Pisa*

TNBC defined by a lack of ER, PgR and Her 2 expression in tumor cells occurs most frequently in young or pre-menopausal women and often have a worst outcome than patients with other breast cancer subtype. Recent studies have suggested a link between BRCA mutations and TNBC. BRCA1 and BRCA2 mutations are present in 8-14% and 5% of TNBC unselected for family history of breast cancer respectively. Recent advances in DNA sequencing have led to the development of breast cancer susceptibility genes panel to germline genetic testing of patients. We assessed the frequency of mutations in predisposition gene (BRCA1 -2) and in 7 predictor genes (BARD1, PALB2, BRIP1, CDH1, PTEN, CH£CK2 and TP53) and the clinical outcome in a cohort of patients with TNBC unselected for family history of breast and ovarian cancer in order to evaluate the clinical utility of germline testing in these subset of TNBC. 36/140 frozen samples of TNBC from patients unselected for family history of BC and OC were enrolled and germline DNA was sequenced to identify mutations. Mutations were identified in 13/36 pts (36.1%), of these 7 (19.5%) in BRCA1 and BRCA2 genes. Mutations in other predisposition genes were detected in 7/36 pts, with the majority observed in genes involved in homologous recombination including PALB2 3/36 (8.3%), BARD1 2/36 5.6%), CDH! 1/36 (2.8%), BRIP1 1/36 (2.8%) associated with BRCA1 mutation, no mutations were discovered in PTEN and CHEK2. Pts with TNBC and germline mutations treated with adjuvant/neoadjuvant chemotherapy have a better outcome in term of DFS and OS than pts with TNBC without germline mutations, only 4/13 pts with mutation experienced progressive disease. Due to the restricted number of pts of our cohort is not possible to generalized our results but we can suggest that germline genetic testing BRCA1 and BRCA2 in TNBC unselected for family history of BC and OC should be considered. Although Mutations in other predisposition genes have been observed among patients with TNBC more data from large trials are needed before to translate the use of these genes in clinical practice.