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breast cancer, early stage

176P Famosa: Evaluation of a multigene panel in patients with suspected HBOC

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Background: Objectives: Characterize 1) the frequency of mutations in patients with clinical criteria for HBOC using a 25-gene panel in a Spanish population (FAMOSA study). 2) The psychological impact of these tests and patient's counseling preferences.

Methods: Patients with breast or ovarian cancer who met the NCCN criteria for genetic testing with a) prior testing for BRCA genes with NO mutation identified; or b) recently diagnosed (<6 months) and not genetically tested, were enrolled for multiplex cancer testing (MyRisk 25-gene panel). Participants completed self-questionnaires regarding geneting counseling preferences and three psychological scales (MICRA, CWS, R-IES) at base-line, one week, three and twelve months after results disclosure.

Results: From November 14 to February 15, 210 patients were included in the FAMOSA study (109 HBOC). 61 (56%) patients were previously tested for BRCA1/2 gene mutations with conventional techniques; median age: 44y (22-77); gender: 3 males / 106 females; cancer types: breast 95 (87%); ovary 14 (13%). Overall 22 pathogenic variants were identified in 21 patients (19,3%): 10 BRCA1, 2 BRCA2, 2 PALB2, 3 MUYTH, 1 CDKN2A; 2 ATM, 1 BRAD1, 1 BRIP1. One patient had an unexpected mutation in CDKN2A gene (gluteus sarcoma age 20; bilateral breast ca; ages 45 and 50; father lung ca, age 70; brother melanoma, age 35). Three patients had a significant mutation of a recessive condition in MUYTH. Of 61 patients previously tested negative for HBOC, 1 had a pathogenic variant in BRCA1 and 17/ 19 patients with VUS were classified negative in BRCA genes with MyRisk.Patients are willing to be disclosed all available information from panel testing. Differences were observed among type of results at short and mid-term. Cancer worry was higher in moderate-penetrance carriers than high penetrance carriers. Longer follow up is ongoing.

Conclusions: Panel testing in patients with HBOC yielded a 19,3% mutation rate, increasing the yield of genetic mutations beyond BRCA. Patients are willing to be disclosed all available information from panel testing.

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