P015 A dedicated screening for early detection of Prostate cancer (Pca) in men with germline mutations in DNA-Repair Genes (DRG)

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Introduction & Objectives: The aim is to investigate feasibility of a dedicated screening for male with high genetic risk (HR) of Pca as carriers of germline DRG mutations and its sensitivity on early diagnosis. The secondary outcome is participation whether mutated and compliance at screening.

Materials & Methods: DRG mutated healthy men came from families of women with breast/ovarian cancer identified reviewing the genealogical trees of patients with BRCA1/2 mutation or from HR Pca patients aged 35-75 years old(yo) with Gleason Score(GS)≥4+3 or <50yo with GS≥3+3. If positive, a targeted DRGs mutational analysis was offered to all son and I degree male relatives between 35-69yo and those mutated were enrolled in the screening for early detection PCa.Mutated healthy men were screened by the Prostate Health Index (PHI) and digital rectal examination (DRE) every year. In case of positive DRE, patients underwent an MRI and, if PIRADS≥3, a target fusion biopsy (TB) + systematic biopsy (SB) was performed, or only SB if PIRADS1-2. In case of negative DRE, patients were stratified according to PHI.If PHI≥40 patients underwent an MRI with SB or SB+TB if PIRADS≥3. In case of 20≤PHI<40 patients underwent an MRI with TB+SB when PIRADS≥3, or they were screened annually by PHI and DRE if PIRADS1-2. If PHI<20, patients were screened annually.

Results: In 12 months we enrolled 62 mutated men, all patients' relatives of breast/ovarian cancer women with a DRG mutation, who all accepted to follow the screening. Mean age was 52 yo. 37(59.7%) were BRCA2 mutation carriers, 17(27.4%) BRCA1, 3(4.8%) PMS2, 2(3.2%) MSH2, 1(1.6%) MLH1, 1(1.6%) BRIP1, 1(1.6%) ATM. 49(79%) were born in Northern Italy, 9(14.5%) in Southern Italy, 4(6.5%) in central Italy. 47(75.8%) had at least one child.Only 3(4.8%) had no child and weren't married. 48(77.4%) had a level of education equal or higher than diploma, 18(29%) were graduates. Only 17(27.4%) had Pca history. All 62 men had negative DRE. Median PSA was 0.79 ng/ml, median PHI 15.0. 44 men had PHI<20; 16 had PHI 20≤PHI<40 and 2 PHI≥40. As a result 18 men underwent an MRI because PHI≥20, obtaining 10 PIRADS1, 7 PIRADS2 and 1 PIRADS4 (negative biopsy), the latter will be screened annually. The 2 patients with PHI≥40 had negative MRI, nevertheless they underwent biopsies based on their high PHI (negative biopsy). The 44 with PHI<20 will continue to be screened annually.

Conclusions: All those mutated for DRG accepted to follow the screening. An accurate evaluation of the genealogical trees of breast/ovarian cancer BRCA1-2 mutated patients and relatives of men with PCa and DRG mutation allows the detection of men potentially carriers of DRG mutations and consequently enroll them in a dedicated screening, currently absent. As a result, we set out to design a screening based on PHI and MRI aiming to improve the capacity of early diagnosis as well as creating a potentially personalized approach.