Background: Breast cancer (BC) is the most common form of malignant tumor in women worldwide. 60–70% of BC are hormone receptor-positive (HR+), HER2-negative (HER2–). The purpose of this analysis was to enhance understanding on the epidemiology for women with PIK3CA-mutant HR+/HER2– metastatic breast cancer (mBC).

Methods: PIK3CA mutations were tested from tumor biopsy (N = 1617) and circulating tumor DNA (ctDNA) (N = 1466) from patients enrolled into BOLERO-2, BELLE-2 and BELLE-3, which are three randomized Phase III studies in HR+/HER2– mBC. Various PIK3CA mutation testing methods were applied, including Next-Generation Sequencing (NGS) and Polymerase Chain Reaction (PCR) for tumor biopsies, as well as BEAMing and droplet digital PCR for ctDNA samples.

Results: Prevalence of the PIK3CA mutations among tissue biopsies ranged from 34.1% to 41.1%, while prevalence of the PIK3CA mutations among liquid biopsies ranged from 27.5% to 43.3%. Besides gene-level analysis, the PIK3CA prevalence by hot spots and by exons was examined as well. Further, subgroup analysis of PIK3CA prevalence had been conducted based on patient cohort (2L vs 3L), mutation testing hot spots and by exons was examined as well. Further, subgroup analysis of PIK3CA ranged from 27.5% to 43.3%. Besides gene-level analysis, the PIK3CA prevalence by exons was examined as well. Further, subgroup analysis of PIK3CA ranged from 27.5% to 43.3%.

Conclusions: PIK3CA mutations (specifically hotspots H1047R, E545K and E542K) frequently occur in HR+/HER2– mBC. The prevalence of PIK3CA mutations are in a relatively narrow range across the three randomized Phase III studies in HR+/HER2– mBC regardless of tissue types and testing methods.

Legal entity responsible for the study: Novartis Pharma.

Funding: Novartis Pharma.