Humaid O Al-Shamsi et al., J Cancer Sci Ther 2017, 9:1(Suppl)
http://dx.doi.org/10.4172/1948-5956-C1-096

Molecular spectra, frequencies, and distribution patterns of somatic mutations using next generation sequencing in Arab women with breast cancer

Humaid O Al-Shamsi[1,2], **Aydah Al-Awadhi**[1], **Banu Arun**[1] and **Nuhad K Ibrahim**[1]
[1]The University of Texas MD Anderson Cancer Center, USA
[2]Khalifa Bin Zayed Al Nahyan Foundation, UAE

Purpose: Breast cancer in Arab world has unique clinicopatholigical features including early onset, higher grade and higher *HER2* amplification, the aim of this retrospective study was to assess the molecular spectra, frequencies, and distribution patterns of somatic mutations using next generation sequencing (NGS) in Arab women with breast cancer.

Subjects & Methods: 78 consecutive Arab women with breast cancer whose tumors had been evaluated using NGS were identified and retrospectively reviewed. We recorded patient characteristics, tumor pathological features, the rate of somatic mutations found on the NGS.

Results: The median age at diagnosis was 52.3 years (range: 37-82 years). 30 (38.5%) of the 78 patients were 50 years of age or younger. A familial history of breast cancer was documented in 30 (38.5%) patients. NGS revealed the following somatic mutation rates: *TP53*, 23%; *ATM*, 2.5%; *IDH1*, 2.5%; *IDH2*, 3.8%; *PTEN*, 7.7%; *PIK3CA*, 15.4%; *APC*, 7.7%; *NPMA1*, 1.3%; *MPL*, 1.3%; *JAK2*, 2.5%; *KIT*, 7.7%; *KRAS*, 3.8%; *NRAS*, 3.8%. *DH1* and *IDH2* were 2.5% and 3.8% respectively. Two patients (2.5%) had *JAK2* mutations and both had an advanced triple-negative disease. Compared with Western population, Arab women have higher rates of *APC, PTEN, KIT, KRAS, NRAS* and *DH1* somatic mutations and lower rates of *TP53* and *PIK3CA* somatic mutations compared to Western women. *ATM* mutation rate was similar. Two novel somatic mutations were identified NPM1 and *MPL* with undefined role in breast cancer pathogenesis.

Conclusions: Our results revealed differences in the genetic profiles and mutation hotspots in Arab women with breast cancer compared to the reported genetic profiles of Western women with breast cancer. These results may have clinical implications in some of the actionable mutations and their targeted therapies once the roles of these somatic mutations in breast cancer tumorigenesis are more defined.

Biography

Humaid O Al-Shamsi is currently working as an Assistant Professor, University of Texas MD Anderson Cancer Center and he is also positioned as an Assistant Clinical Professor (Part Time) in the Department of Oncology at McMaster University. He has been a recipient of many award and grants. His research experience includes various programs, contributions and participation at different countries for diverse fields of study. His research interests reflect in his wide range of publications in various national and international journals. His research interests include Oncology, Radialogy, Hepatology, Clinical Oncology, etc.

nibrahim@mdanderson.org

Notes: