Hekmet Bugrein, J Cancer Sci Ther 2014, 7:10
http://dx.doi.org/10.4172/1948-5956.C1.061

OMICS International
conferenceseries.com

8th Euro Global Summit on
Cancer Therapy
November 03-05, 2015 Valencia, Spain

Genotype and phenotype correlation of breast cancer in BRCA carriers and non-carriers

Hekmet Bugrein
Nara Medical University School of Medicine, Japan

Introduction: Breast cancer is both genetically and histopathologically heterogeneous disease, the biological basis for this heterogeneity is unknown, although there are some distinct phenotype-genotype correlations. Approximately 5% to 10% of breast cancer is hereditary and *BRCA1* and *BRCA2* genes are responsible for the majority of hereditary breast cancers. *BRCA1* and *BRCA2* mutant cells have defects in the DNA repair and chromosomal instability with marked defects in cell division. In the literatures *BRCA1* and *BRCA2* associated breast cancers have different clinical, histological, immune-phenotypic features. To validate the effect of *BRCA1* or *BRCA2* germ line mutation on breast cancer aggressiveness and on the clinical features, we analyzed, correlate the genotype and compared histological and molecular status and clinical variables of 23 breast cancer patients with BRCA gene mutation carriers and histopathological and molecular characters of breast cancer in 50 patients affected in the same age group but with no pathogenic mutations or variants of unknown significant (VUS) in either *BRCA1* or *BRCA2* genes.

Methods: A retrospective review of medical records was conducted to determine the clinical characteristics, the molecular results of BRCA testing and the tumor characteristics from the histopathology reports in addition to a new review of the tumor blocks. All of the cases were evaluated at the high risk hereditary breast and ovarian cancer clinic at the national center of cancer care and research (NCCCR) in state of Qatar from 2013 – 2015.

Results: 75 patients with breast cancers were diagnosed at ages 50 year and below, (50) patients were BRCA negative and (25) patients were BRCA positive, (17) were *BRCA1* mutation carriers ,(6) patients were *BRCA2* mutation carriers and (2) carried mutations in both *BRCA1* and *BRCA2* genes. On MLPA the most detected mutation in BRCA gene is small frame shift deletion or insertion in one or more exons and protein truncation. In this study age group carriers and non-carriers are young (50 and below) but BRCA mutation detected with higher incidence in younger ages, histopathology of invasive ductal carcinoma (IDC) was detected in (95%) of BRCA gene mutation carriers with (52%) high nuclear grade 3 and a higher proliferative rate were detected, in non-carriers control group nuclear grade 3 in 36% of cases and high proliferative rate in 31%. Triple negative (56.5%) ,triple positive (13%), ER PR positive Her2 negative (26%) and ER PR negative Her2 positive (4.3%) in BRCA carriers in non-carriers TN in 17%, triple positive 13%, ER PR positive Her 2 negative 44%, ER PR negative Her 2 positive 13%.

Conclusion: These results suggest that tumors associated with BRCA mutations are more likely to be basal sub type, has more aggressive behavior (invasive, Grade 3, Triple negative) affect younger age group, patient presented in more advanced stage further analysis needed to determine clinical outcomes in BRCA positive compare to control in regards to local and systemic relapse, overall survival . However will continue to build up our data base for better characterization of our hereditary breast cancer cases at clinical and molecular level and use this information for future development of targeted anti-cancer agents.

Biography

Lyne Racette is an Assistant Professor of Ophthalmology at the Eugene and Marilyn Glick Eye Institute at Indiana University's School of Medicine (Indianapolis, IN), and an Adjunct Professor at Indiana University's School of Optometry (Bloomington, IN). After receiving her PhD from Carleton University (Ottawa, Canada), she joined the University of California at San Diego (San Diego, CA) where she completed a Postdoctoral Fellowship at the Hamilton Glaucoma Center.

h.bugrein@yahoo.com

Notes: