DOI: 10.29289/259453942018V28S1018

ANALYSIS OF A SYNONYMOUS SINGLE NUCLEOTIDE VARIANT IN PLATELET-DERIVED GROWTH FACTOR RECEPTOR ALPHA GENE IN TRIPLE NEGATIVE BREAST CANCER

Joana D. R. Santos[1], Nancy C. F. L. Ferreira[1,3], Darley L. F. Filho[3], Cintia R. C. Rocha[1,2], Carlos H. M. Castelletti[1], José L. L. Filho[1,2], Danyelly B. G. Martins[1,2]

[1]Molecular Prospecting Sector, Laboratório de Imunopatologia Keizo Asami, Universidade Federal de Pernambuco (UFPE) – Recife (PE), Brazil.
[2]Department of Biochemistry, Centro de Biociências, UFPE – Recife (PE), Brazil.
[3]Hospital Barão de Lucena – Recife (PE), Brazil.
Financial support: CAPES, FACEPE, CNPq.

Objective: This research had the purpose of ascertain if mutations in PDGFRA gene (platelet-derived growth factor receptor alpha) are associated with triple negative breast cancer, and compare this group with luminal subtype. Clinical, and staging data were considered. **Methods:** Genomic DNA was extracted of 12 tissue samples in which 9 from luminal subtype and 3 TNBC by *DNeasy® Blood & Tissue Kit* (Qiagen). PDGFRA gene were amplified by PCR (polymerase chain reaction) with specific primers and *GoTaq® Green* Master Mix (Promega), in the thermal cycler *Veriti* (Applied Biosystems). The PCR products were purified by *ExoSAP- IT PCR Product Cleanup* (Thermo Fisher Scientific). PCR fragments were then sequenced using *BigDye Terminator* (Applied Biosystems) on a *MegaBACE 1000 Sequencing System* (GE Healthcare). **Results:** The results were used to performance sequence alignment in *CLC Genomics Workbench* according to PDGFRA gene sequence (NG_009250).The age of women were from 32 to 70 years. All TNBCs patients in this study were recidivant in breast cancer. Upon analysis of the PDGFRA genome, a mutation was found in all TNBC samples, and in none of the luminal subtype. The mutation consists of an A> G exchange at position 50792 of the DNA strand. The amino acid encoded by this region, proline, remains preserved. Thus it is considered a sSNV (Synonymous Single Nucleotide Variant), classified how a type of silent mutation. Despite being classified as silente, is now known that sSNVs may have multiple consequences for RNA maturation and stability as as well as protein translation. In addition, tissue-specific and tumor-specific changes in tRNA (transfer ribonucleic acid) expression combined with symmetric tRNA abundance may play a role. Like many receptor tyrosine kinases (RTKs), PDGFRA is involved in the progression of a variety of cancers either by overexpression or by increased activity. **Conclusion:** Systematic reporting of sSNVs will be essential to achieve positive progress in our understanding of the full spectrum of functional effects associated with genomic variants in the population as well as in each patient. Knowing this genetic information is essential for precision medicine. In the future it will provide diagnosis and treatment specific to each individual.