The most described and important cancer in woman is breast cancer. Occupying a staggering second place of cancers with high incidences. The disease can be found more in women than in men, covering a 100 to 1 ratio (1,2). Historically breast cancer always was rated the second most common cancer in North America in comparison with lung cancer that has been rated as number one (3,4,5). According to Society AC. et al breast cancer outdistances lung cancer to be the most prominent cancer (6). Meanwhile in South-Africa studies reported that the lifetime breast cancer risk for women is 1 in 28, with 0,7% of all deaths caused by breast cancer.

Previous reports suggested that the African-American population has an increased unequal weight of aggressive early-onset breast cancer in comparison to various population subgroups (8,9). Moreover, Sundquist M et al 2002 reported that 10-20% of the early-onset group (under 40 years) were carriers of a BCRA1/BCRA2 mutation (10). Additionally, multiple research groups on breast cancer susceptibility have focussed on variants in high penetrance genes such as BCRA1, BCRA2 and TP53. However, in this report the centre of attention shifted to {ATM, CHEK2, BRIP1, PALB2, RAD50, NBN and RB1} which are medium penetrance. The other low penetrance genes were PTEN, RAD51C, BARD1, STK11 and CDH1(11). These genes were selected based on study of genes that play major roles / most involved in cancer susceptibility. Neuhausen SL et al 2002. reported that different population groups can be influenced by altering variants (12). Although, most of the available databases illustrated data generally from European populations and the Asian population, a few studies have reported data of African-American populations. However, because of extensively fluctuating levels of ethnic groups, the data can be difficult to clarify/decipher. Moreover, cancer-related variant data/statistics is extremely rare (13). A lot of previously reported South-African studies on black females, have concentrates mainly on BRCA1 and BCRA2 variants and other single genes (15-17). Now-a-days doing research on unique variants in certain populations with minimal genomic data has gained significant increased interest (18-23).

During the last 10 years, racial subgroups distinguished research on pathogenesis has seen an increase. For the simple reason that the quality of different techniques such as third generation sequencing (TGS), whole genome (WGS), exome sequencing (ES) has enhanced. However, WGS is still rather costly, ES and directed panel sequencing (DPS) are becoming cheaper. Especially DPS has gained interest, this due to the ability of commercial pre-designing panels with the pathogenesis that researchers are interested in. Now, various businesses are manufacturing carcinoma sequencing panels which will be accessible for both germ line and somatic studies.

In this study on breast carcinoma susceptibility genes in black South African females the Illumina TruSight Cancer Panel was utilized which is a capture-based panel. The panel targets 94 carcinoma-related genes and 284 SNPs, that were earlier correlated with a tendency towards carcinomas. 166 black [South] African females were investigated with ages varying between 18 and 54 years.

A screenshot of a cell phone

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