Saxena et al. Genome Biology 2010, 11(Suppl 1):P34
http://genomebiology.com/2010/11/S1/P34

Genome Biology

POSTER PRESENTATION Open Access

Genomic alterations in breast cancer patients from Northeast India using 10K SNP arrays

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From Beyond the Genome: The true gene count, human evolution and disease genomics
Boston, MA, USA. 11-13 October 2010

Background

North-east India has always been a 'hotspot' for population geneticists because of its unique, strategic geographic location and the presence of linguistically, culturally and demographically diverse populations practicing various tobacco habits. This region reports a high incidence of breast cancer in females with a history of extensive exposure to tobacco. Therefore, this population is constantly exposed to a high level of genotoxic stress. Accumulation of multiple and discrete genetic events during tobacco exposure can combine to drive breast cancer pathogenesis. Direct analysis of the tumor genome can reveal the genomic events accumulated during tumor progression. Hence, we investigated genomic alterations in breast cancer patients with tobacco exposure.

Methodology

Copy number analyses of 30 breast tumors was performed using the SNP 10K array. DNA from 30 tumor samples and 10 blood samples was extracted. 250ng of genomic DNA was digested with XbaI and ligated to XbaI adaptor before subsequent PCR amplification. This was fragmented with DNaseI and visualized on a 4% Tris-borate EDTA agarose gel to confirm DNA fragmentation. Fragmented PCR products were biotin labeled, hybridized and detected with an Affymetrix Fluidics Station 450 and GeneChip Scanner 3000. Genotype information was extracted from the Mapping 10K Arrays using the Genechip DNA Analysis Software (GDAS) from Affymetrix. Genotypes derived from germline and cancer DNA was loaded into the software package dChip which was used for copy number analysis.

Results

Complex chromosomal alterations involving multiple levels of change were observed. Copy number gains were observed on Chromosome 1q31, 1q41-42,20q13, 20p11-12, 3q26-27, 6p22, 8q22-24 and 8q13. High level amplifications were observed on Chromosome 1q and 3q. These regions include genes such as *MIA3, LAMP3, ABCC5, TP63, KCNK9, IL7* and *ADCY8*. Losses were observed less frequently than gains and the minimal common regions of the most frequent losses were 11q23-q24, 17p12-p13, 18q21, 13q12-q13, 13q21, 8p21-p22 and 9p21-p23. These regions include genes such as *NCAM1, OPCML, TP53, MAPK4, CCBE1, MAST4, FBXL17* and *FER*.

Conclusion

These copy number changes might play an important role in breast carcinogenesis and the data on genetic gain and loss might provide new insight to the region specific molecular etiology of breast cancer. These data also indicate chromosomal aberrations associated with tobacco exposure.

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Published: 11 October 2010

doi:10.1186/gb-2010-11-S1-P34
Cite this article as: Saxena *et al.*: Genomic alterations in breast cancer patients from Northeast India using 10K SNP arrays. *Genome Biology* 2010 **11**(Suppl 1):P34.

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