

# Study of Karyotypes in Patients of Breast Carcinoma in Gujarat State

Dr. Lalit C. Ratanpara<sup>1</sup>, Dr. Paras S. Srimankar<sup>2\*</sup>

<sup>1</sup>Tutor, Department of Anatomy, PDU Govt. Medical College, Rajkot, Gujarat, India

<sup>2</sup>Associate Professor, Department of Anatomy, B. J. Medical College, Ahmedabad, Gujarat, India

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\*Corresponding author: Dr. Paras S. Shrimankar

## Abstract

**Objectives:** Carcinoma of the breast has commanded the interest of all the disciplines of medicine by magnitude and serious consequences of the lesion. In India the incidence of the disease is low in comparison to western countries. Cancer of the breast is the commonest cancer - affecting woman worldwide. The objective of this study is to determine the relative frequency of genetic abnormality in cases of Breast carcinoma. **Methods:** The present study includes observations of 25 cases diagnosed as breast carcinoma from Gujarat cancer and research institute and Civil Hospital, Ahmedabad, during the year of 2014-15. About 30 metaphase plates were observed in each case and finally, a photograph was obtained from a good quality metaphase slide. Correlation of chromosomal finding was done with similar studies done in past. **Results:** The findings in present study suggest no structural and/or numerical chromosomal abnormality was detected in peripheral blood lymphocytes of any patients. **Conclusion:** The Karyotypic study helps to find out some numerical as well as structural chromosomal abnormalities and also useful in implementing a cost effective management of detecting early breast cancer in family members and to provide them with necessary preventive measures and treatment.

**Keywords:** Karyotypes, Breast Carcinoma, cancer, treatment.

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## INTRODUCTION

The female breast is an insignia of femininity and a symbol of womanhood. Hence its disease evoke a fear of mutilation and loss of femininity [15].

Breast cancer is the world's most common invasive cancer among females<sup>6</sup>. It is a type of cancer originating from breast tissue, most commonly from the inner lining of milk ducts or the lobules that supply the ducts with milk [2].

Breast cancer has been known to mankind since ancient times. It has been mentioned in almost every period of recorded history. Earlier, however, it was a matter of taboo and embarrassment that meant detection and diagnosis was rare. In the 1990's the symbol of breast cancer - the pink ribbon - brought out a revolution against this cancer [5].

Breast as an organ has a high rate of malignancy. Hence the dictum 'Any tumor in the breast is considered malignant unless proved otherwise [15].' Carcinoma breast constitutes one of the most common cancer in women [6].

As per WHO, Incidence of Breast carcinoma is 27% of all cancers in females. In 2013, an estimated 2,32,340 new cases of invasive breast cancer has been diagnosed among women and approximately 39,620 women had died from breast cancer worldwide [1].

In India, it is 2<sup>nd</sup> to carcinoma cervix with annual incidence of 17 per 1 lakh. As per the ICMR-PBCR data, breast cancer is the commonest cancer among women in urban registries of Delhi, Mumbai, Ahmedabad, Calcutta and Trivandrum where it constitutes more than 30% of all cancers in females [13]. In the rural areas, breast cancer is the second most common cancer in women after cervical cancer [4, 18].

Breast cancer is a tumor that starts from cells of the breast tissue, either in cells that line the ducts that carry milk to the nipples (ductal cancer) and/or in cells that line the lobules, which are glands involved in milk production. Breast tumors can be benign or malignant, the former are not life-threatening, can usually be removed, do not invade adjacent tissues or spread to other parts of the body and can include fibrocystic tissue, fibroadenomas and benign breast disease. Malignant breast tumors are cancerous and can invade

surrounding tissues or metastasize to other parts of the body via the lymphatic system (lymphatic vessels and lymph nodes), such as the liver and bone [13].

More than 90% of the tumors are said to arise from the epithelium of the duct system, Sarcoma comprises a separate group due to its origin from the stromal tissue. Carcinosarcoma on the other hand derives origin from both the epithelial and the stromal tissue [13].

Genomic instability is a prerequisite for the onset of any cancer. It is estimated that 5% to 10% of breast cancer cases result from inherited mutations. These mutations are present in less than 1% of the general population, but occur more often in certain ethnic groups such as those of Ashkenazi (Eastern European) Jewish descent. There are two major susceptibility genes that were identified till now are BRCA 1 and BRCA 2 [1]. These two breast cancer susceptibility genes together account for most of inherited breast cancer. However, it is likely that other numerical and structural chromosomal abnormalities may also play a role in the development of sporadic breast cancers [16].

Finding of structural and/or numerical chromosomal aberration in the breast cancer help surgeons to identify patients whose family members and relatives are highly susceptible to develop breast cancer. It is also useful in implementing a cost effective management by detecting early breast cancer in family member and take necessary preventive measures and treatment [7]. It is also helpful to determine molecular mechanism of mammary tumorigenesis. Though, Mammography is the single most effective method in early detection of Breast cancer but due to some limitation of mammography [1], the emphasis is given on initiating karyotyping as a screening recommendation for detecting early age of onset in hereditary breast cancer patients. Molecular analysis can be used to detect the defect at the gene level but it is a tedious and very costly technique [7].

In present study, Karyotyping was done in 25 cases of Breast carcinoma to find out underlying chromosomal abnormalities.

## MATERIAL AND METHOD

The present study includes observations of 25 cases diagnosed as breast carcinoma from Gujarat cancer and research institute and Civil Hospital, Ahmedabad, during the year of 2014-15.

Personal data including name, age, sex, onset and type of illness, relevant personal history, past history and family history were noted. In addition, vital statistics, brief clinical assessment, all routine and specific investigations regarding Breast carcinoma were also noted. Karyotyping was done in all 25 cases.

## METHOD OF KARYOTYPING

Blood samples of the patients were obtained in a heparinized container. The cultivation was done under all aseptic precautions on the same day of the aspiration. After an incubation period of 69.5 hours at 37°C, the cell division was arrested at metaphase stage by adding colchicine and after total 72 hours of incubation period the lymphocytes were harvested [9].

The sample was centrifuged to remove supernatant fluid at 3000 rpm for 10 minute. The hypotonic solution (KCl 0.075 M) was added at 37°C for 20 min to swell the cells, and the sample was treated thrice with chilled fixative (3:1 ratio of methanol: acetic acid). Finally, the slides were prepared for Karyotype study. All the slides were scanned for metaphase detection. The readings were then taken from each slide and noted. Thereafter, those slides showing metaphase with good morphology of chromosomes were selected and kept under non-humid dry wooden boxes for aging process [9].

Approximately after 7 days of harvesting, Giemsa banding procedure was done using freshly prepared Trypsin-EDTA solution and Giemsa stain. The procedure protocols were followed according to the guidelines from the book Rooney and Czepulkowski, Human Cytogenetics: A practical approach [9].

A band is defined as the part of chromosome, which is clearly distinguishable from its adjacent segments by appearing darker or lighter. The first banding method was quinacrine fluorescence method; developed in 1970, which was followed by various methods of Giemsa staining. A Giemsa dye mixture reveals identical patterns of dark and light bands along the chromosomes. This kind of technique is called G-Banding method and resulting bands are G bands where the dark bands contain mainly A-T rich DNA and the light bands are G-C rich DNA [10].

About 30 metaphase plates were observed in each case and finally, a photograph was obtained from a good quality metaphase slide. The chromosomal findings were described according to the international system of Human Cytogenetic Nomenclatures and finally, Karyotypes were prepared using Automatic Karyotyping software. Correlation of chromosomal finding was done with similar studies done in past.

## RESULTS

**Table-1: Showing distribution of cases according to cytogenetics findings**

Metaphase findings	Carcinoma patients	
	No. of patients	Percentage (%)
Numerical abnormality	0	0
Structural abnormality	0	0
Normal Karyotype	25	100

Above table shows that no structural and/or numerical chromosomal abnormality was detected in peripheral blood lymphocytes of any patients.

## DISCUSSION

Breast Carcinoma is the commonest invasive cancer all over the world. Because of its high frequency in India also, early diagnostic procedures, genetic counselling and population screening are very important. As a part of this, in present study 25 clinically diagnosed cases of Breast carcinoma were selected. Relevant and significant clinical parameters were noted. Karyotype study of all 25 cases was done and an attempt was made to find out chromosomal abnormalities seen in cases of Breast carcinoma. The data of present study were compared with others reported in literature.

Genetic instability is a definitive feature of human cancer. Breast cancer patients also had structural and/or numerical abnormalities in their cultured lymphocyte.

According to study of Bieche I *et al.*, [3], 22% cases of breast carcinoma showed loss of chromosome 8p in their cultured lymphocytes.

According to study of Pandis N *et al.*, [11], out of 97 breast carcinoma patients 25 cases observed with loss of chromosome 8p by conventional G-banding.

According to study of Teixeira MR *et al.*, [17], out of 20 patients of study group 5 cases found with breaks on chromosome 3p, whereas 2 cases detected with del(1)(q42).

According to study of Pathmanathan *et al.*, [12], chromosome 8p deletion found in 18 patients and trisomy 17 found in 3 patients out of 200 cases of their study group.

According to study of Santhi Latha Pandrangi [14], Karyotype analysis of breast carcinoma patients showed aneuploidy, deletions and multiple rearrangements in chromosomes 7, 9, 11, and X.

According to study of Hinglaj Saha [8], Cytogenetic studies on 10 breast carcinoma patients shows variety of chromosomal aberrations including aneuploidy, polyploidy, cluster of cells, acrocentric associations, chromosomal breaks and gaps in their peripheral blood leucocytes.

As shown in Table-1 in present study, no structural and/or numerical chromosomal abnormality found in peripheral blood lymphocytes of any patients. Like other studies genetic association in breast carcinoma could not be found. It may be due to small sample size. It is possible that the patients may have genetic abnormalities at the level of gene instead

involving whole chromosome or large portion of it and so could only be detected by molecular study.

## CONCLUSION

Breast carcinoma is the world's most common invasive cancer among females. Its incidence in India is about 17 per 1 lakh women. The present work "Study of karyotypes in patients of breast carcinoma in gujarat state" was carried out on 25 patients from Gujarat cancer research institute and Civil hospital, Ahmedabad. Following conclusion was made from the results of present study.

None of the patients showed structural and/or numerical chromosomal abnormality in their karyotypic study of peripheral blood lymphocytes it may be due to small sample size. The patients may have genetic abnormalities at the level of gene instead involving whole chromosome or large portion of it and so could only be detected by molecular studies like Fluorescence in situ Hybridization (FISH) or Comparative Genomic Hybridization (CGH). Even though, The Karyotypic study helps to find out some numerical as well as structural chromosomal abnormalities and also useful in implementing a cost effective management of detecting early breast cancer in family members and to provide them with necessary preventive measures and treatment.

The results of the present study may be used as reference guide for future studies about Breast carcinoma as well as for screening procedure and to map out preventive strategies against breast carcinoma.

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