Nile University

Core Requirement

BMD427: Applied Bioinformatics"SP23

Interpretation for the analysis

[Literature review]

Exploring Genomic Variants and Mutations in PRJNA188274 Dataset: Insights into Breast Cancer Development and Progression

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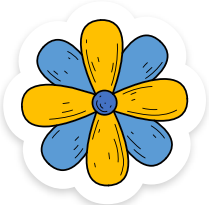

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Interpretation for the analysis

Breast cancer affects a lot of people, and it has significant rates of morbidity and death all around the world. The breast tissue experiences aberrant cell proliferation, resulting in tumors that have the ability to spread to distant organs and infiltrate surrounding tissues. Breast cancer is an important global health concern and, in terms of frequency range, is the second most common disease among women globally, behind skin cancer. Genetic changes or DNA damage are the primary causes of this illness, which results in aberrant cellular proliferation and the creation of tumors inside the breast tissue.

Breast cancer is a complicated disease because of its heterogeneity, which includes a wide range of molecular subtypes and inconsistent clinical results. Unravelling the precise variations or mutations present in the genome is essential to gaining a thorough understanding of breast cancer and developing efficient techniques for its prevention, detection, and treatment.

Breast cancer susceptibility, disease development, and treatment response are all significantly influenced by genetic differences. The underlying genetic pathways that promote the onset and progression of breast cancer can be better understood by finding these variations. Additionally, research into the genetic profile of breast cancer can help in the creation of focused treatments and personalized medical strategies.

Finding certain mutations or variations linked to breast cancer can aid in choosing the best course of treatment for each patient, enhancing clinical results and minimizing harmful adverse effects. The examination of the complete genome and the detection of genetic changes on an unprecedented scale have been made possible by improvements in high-throughput sequencing methods and bioinformatics applications.

Scientists may now examine the patient's genome for significant mutations and genetic abnormalities that lead to breast cancer disease. It's essential to comprehend the genetics of breast cancer in order to create efficient preventive, early diagnosis, and treatment plans. By enabling the exact detection of variations and mutations within the genome, cutting-edge genomic sequencing technologies have completely changed the way breast cancer is studied. Single nucleotide variants, insertions, deletions, and structural changes are just a few of the genomic modifications that may be easily detected thanks to these cutting-edge methods.

These discoveries into the molecular processes that underlie the pathogenesis of breast cancer have been made as a result of extensive genomic research. These researches have revealed the complexity of breast cancer, exposing many subtypes with distinctive genetic profiles and clinical characteristics.

The development of breast cancer is known to be significantly influenced by genetic alterations and DNA damage. Specific genetic changes linked to the onset and development of breast cancer have been found in several investigations. These mutations can affect tumor suppressor genes, which govern cell cycle regulation and inhibit aberrant cell development, or oncogenes, which increase cell growth and division.

Additionally, genetic changes that raise the risk of breast cancer can be brought on by environmental factors that damage DNA, such as radiation exposure or exposure to certain chemicals. Modern genomic sequencing methods have been used to identify the precise variations or mutations found in the genomes of breast cancer patients.

With the use of these technologies, it is possible to identify genomic modifications including single nucleotide variants, insertions, deletions, and structural variations, which sheds important light on the molecular pathways behind the pathogenesis of breast cancer. The complexity of breast

cancer, with several subtypes distinguished by particular genetic mutations and clinical behaviors, has been uncovered by thorough genomic research.

Our study focuses to determine the variants present in the genomic single data PRJNA188274, which serves as a valuable resource for researchers studying breast cancer genetics and presents a comprehensive workflow for variant identification and analysis in the PRJNA188274 dataset. **Our work compares several genomic variants and Mutations approaches**, to provide a complete comparative understanding of the genetic basis of breast cancer as follows:

Table 1. Literature review

Authors	Problem Definition	Dataset	Methods	Results
Neil M. Iyengar, MD; Clifford A. Hudis, MD; and Andrew J. Dannenberg, MD	The paper reviews the current understanding of the relationship between obesity, inflammation, and breast cancer development and progression.	The paper is a review article.	The paper is a narrative review that summarizes and synthesizes existing literature on the relationship between obesity, inflammation, and breast cancer development and progression.	The paper identifies several mechanisms by which obesity and inflammation promote breast cancer development and progression, including alterations in hormone metabolism, immune function, and the tumor microenvironment.
Véronique Chabottaux and Agnès Noël	The paper addresses the current understanding of the role of matrix metalloproteinases (MMPs) in breast cancer progression and metastasis.	The paper is a review article.	The paper is a narrative review that summarizes and synthesizes existing literature on the role of MMPs in breast cancer progression and metastasis	The findings inform the development of targeted treatment strategies for breast cancer that aim to inhibit MMP activity or expression.

Duppala, S.K.; Kour, B.; Shukla, N.; Dhakane, M.A.; Mishra, A.K.; Digumarti, R.; Pawar, S.C.; Suravajhala, P.N.; Vuree, S.	The paper identifies genetic mutations in the Indian phenotype of cervical cancer using exome sequencing.	The study included a total of 50 patients with cervical cancer who were recruited from a single center in India. Exome sequencing was performed on tumor tissue and matched normal tissue from each patient.	Exome sequencing was performed on tumor tissue and matched normal tissue from each patient. The sequencing data were analyzed to identify genetic mutations in the tumor tissue that were not present in the matched normal tissue.	The study provides new insights into the genetic mutations that contribute to the development and progression of cervical cancer in the Indian population.
Bo Yu, Jia Su, Qian Shi, Xiaoyu Zhang, Yong Chen, Gang Wu, and Jian Huang.	The paper investigates the role of KMT5A-methylated SNIP1 in promoting metastasis in triple-negative breast cancer (TNBC) and to elucidate the underlying molecular mechanisms	The study included TNBC patient samples, TNBC cell lines, and mouse models of TNBC metastasis.	The study used various molecular biology techniques, including chromatin immunoprecipitation (ChIP), luciferase reporter assays, in vitro and in vivo metastasis assays, and RNA sequencing (RNA-seq), to investigate the role of KMT5A-methylated SNIP1 in TNBC metastasis and to elucidate the underlying molecular mechanisms.	the results shows new insights into the molecular mechanisms underlying TNBC metastasis and identifies KMT5A-methylated SNIP1 as a potential therapeutic target in TNBC. The findings can inform the development of targeted prevention and treatment strategies for TNBC metastasis.
Andrew J. Gooding, Bin Zhang, Farid K. Jahanbani, Jiguang Wang, Ling Li, Lixin Liu, and Qihong Huang	The paper addresses the role of the long non-coding RNA (lncRNA) BORG in breast cancer.	The study used breast cancer patient samples, and breast cancer cell lines.	The study used various molecular biology techniques, including RNA sequencing (RNA-seq), in vitro and in vivo metastasis.	The results targeted treatment strategies for breast cancer metastasis and disease recurrence.

Conclusion & Analysis

The literature papers investigated the molecular mechanisms underlying the development and progression of different types of cancer. While the studies provide important insights into the molecular mechanisms underlying cancer development and progression, there are some limitations to consider. As sample size was relatively small, this limits the generalizability of the findings to other populations and highlights the need for replication studies in larger sample sizes. Additionally, although the findings may not fully translate to humans, and further clinical studies are needed to validate the findings in patients.

While, in our study, the identified variants (mutations) in the genomic single data PRJNA188274 in chromosome 22 in Homo sapiens, provided us with valuable insights into the genetic causes of breast cancer. The identified variants (mutations) were distributed across different chromosomes, indicating the heterogeneity of breast cancer at the genetic level. Furthermore, our study contributes to a better understanding of the genetic basis of breast cancer in homo sapiens.