Transparent precision: explainable artificial intelligence empowered breast cancer recommendations for personalized treatment

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ABSTRACT

Breast cancer stands as a prevalent global concern, prompting extensive research into its origins and personalized treatment through artificial intelligence (AI)-driven precision medicine. However, AI's black box nature hinders result acceptance. This study delves into explainable artificial intelligence (XAI) integration for breast cancer precision medicine recommendations. Transparent AI models, fuelled by patient data, enable personalized treatment recommendations. Techniques like feature analysis and decision trees enhance transparency, fostering trust between medical practitioners and patients. This harmonizes AI's potential with the imperative for clear medical decisions, propelling breast cancer care within the precision medicine era. This research work is dedicated to leveraging clinical and genomic data from samples of metastatic breast cancer. The primary aim is to develop a machine learning (ML) model capable of predicting optimal treatment approaches, including but not limited to hormonal therapy, chemotherapy, and anti-human epidermal growth factor receptor 2 (HER2) therapy. The objective is to enhance treatment selection by harnessing advanced computational techniques and comprehensive data analysis. A decision tree model developed here for the prediction of suitable personalized treatment for breast cancer patients achieves 99.87% overall prediction accuracy. Thus, the use of XAI in healthcare will build trust in doctors as well as patients.

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1. INTRODUCTION

Breast cancer, a global health concern of significant magnitude, has spurred extensive investigations into its aetiology and the development of personalized treatment approaches, often leveraging the capabilities of artificial intelligence (AI)-driven precision medicine [1], [2]. While AI holds promise for enhancing medical decision-making, its inherent black box nature has posed challenges to the acceptance of its outcomes [3]. As the medical community strives to harness the potential of AI while ensuring transparency and accountability, this study embarks on an exploration of explainable artificial intelligence (XAI) integration within the realm of breast cancer precision medicine [4], [5]. In recent years, the convergence of AI and healthcare has opened new avenues for tailoring treatments to the individual characteristics of patients, ushering in the era of precision medicine. Breast cancer, with its diverse subtypes and complex genetic underpinnings, is an ideal candidate for such targeted therapeutic interventions. AI technologies, particularly XAI, offer a means to unravel the decision-making processes of intricate models, making them more accessible and interpretable to medical professionals and patients alike. The central aim of this study is to fill the gap between AI's predictive power and the demand for transparent and comprehensible decision-making [6]. By integrating XAI techniques, such as feature analysis and decision trees, into breast cancer precision medicine, the study seeks to empower medical practitioners and patients with clear insights into the rationale behind personalized treatment recommendations. The pivotal role of transparent AI models fuelled by patient data is highlighted, as these models pave the way for informed and collaborative medical decisions, thereby fostering a climate of trust and partnership. In the following sections, we search into the methodology and framework employed to realize XAI integration in breast cancer precision medicine. By unravelling the intricacies of AI-driven recommendations, this research endeavours to contribute to the advancement of breast cancer care within the precision medicine paradigm, ultimately culminating in improved patient outcomes. Furthermore, the study presents a specific decision tree model, showcasing its impressive 99.87% prediction accuracy in the domain of breast cancer precision medicine. Through this investigation, we aim to demonstrate the feasibility and significance of transparent AI integration in the complex landscape of breast cancer treatment. By emphasizing the harmony between AI's potential and the imperative for transparent medical decisions, we endeavour to propel the field of breast cancer care forward, bolstered by the promises of precision medicine and the elucidating power of XAI.

2. CONCEPTUALIZATION AND METHODOLOGY

2.1. Breast cancer

Breast cancer is a prevalent and complex disease which affects large number of individuals worldwide, primarily women. It is a cancer type that develops in the cells of the breast tissue, most commonly in the milk ducts or the milk-producing glands. While breast cancer can occur in men, it is far more common in women, making it the leading cancer diagnosis among females. Breast cancer is a heterogeneous and multifaceted condition, which means that it can evident in various forms, exhibit different characteristics, and require varied treatment approaches. Understanding the basics of breast cancer is vital for both patients and healthcare professionals to facilitate early detection, diagnosis, and effective management [7]. Following are the important aspects to consider:

- Risk factors: different risk factors results in the development of breast cancer, like gender, age, family history, genetic mutations (e.g., breast cancer gene 1 (BRCA1) and breast cancer gene 2 (BRCA2)), lifestyle choices, and hormonal factors such as diet and physical activity.
- Types of breast cancer: breast cancer is of different types, with the two most common being invasive and non-invasive. Invasive breast cancer spreads to other parts of the body, while non-invasive breast cancer is confined to the milk ducts or lobules.
- Early detection: early detection through regular breast self-examinations, mammograms and clinical breast exams is crucial for improving the chances of successful treatment. Recognizing the signs and symptoms of breast cancer, such as a lump or changes in breast appearance, is essential.
- Diagnosis: the diagnosis of breast cancer involves a series of tests like imaging (mammography, ultrasound, and magnetic resonance imaging (MRI)) and biopsy, to confirm the presence of cancer and determine its type and stage.
- Staging: breast cancer staging helps describe the extent of the disease and guides treatment decisions. Stages range from 0 to IV.
- Treatment options: breast cancer treatment varies depending on the stage, type, and individual patient factors. Common treatment modalities include surgery, radiation therapy, chemotherapy, targeted therapy, hormone therapy, and immunotherapy.
- Survival rates: survival rates for breast cancer have improved over the years due to advancements in early detection and treatment. However, outcomes can vary widely depending on factors such as subtype, stage, and access to healthcare.
- Support and resources: breast cancer patients and their families can benefit from a range of support services, including counselling, support, and educational resources. These services can help individuals cope with the emotional and practical challenges of breast cancer.

Breast cancer is a significant public health concern. But ongoing research, early detection efforts, and advancements in treatment options offer hope for improved outcomes and better quality of life for those affected by this disease. It is essential for individuals to stay informed about breast cancer, prioritize regular screenings, and consult with healthcare professionals for personalized guidance and care.

2.2. Precision medicine

Precision medicine, also referred as personalized medicine, is a ground breaking approach to healthcare that recognizes the uniqueness of each patient. It employs genetic, genomic, environmental, and lifestyle data to tailor medical decisions and treatments. Key components include genomics, personalized diagnostics, targeted therapies, individualized treatment plans, predictive analytics, preventive measures, and ongoing monitoring. This approach aims to provide more accurate diagnoses, customized treatments, and better healthcare outcomes. Precision medicine has already made significant strides, particularly in fields like oncology, and it has led to innovative therapies and interventions. With advancing technology and a deeper understanding of genetics and diseases, precision medicine is expected to continue growing, offering the potential for truly individualized care, better patient outcomes, and reduced healthcare costs. It represents a transformative shift in healthcare that emphasizes personalized and effective healthcare for all [8].

Precision medicine in breast cancer treatment involves tailoring medical decisions and therapies to the unique characteristics of each patient and their tumour. This approach begins with molecular subtyping, identifying specific breast cancer subtypes. Targeted therapies are then employed based on the molecular characteristics, such as hormone receptor status and human epidermal growth factor receptor 2 (HER2) expression. Genomic profiling helps identify genetic mutations and resistance mechanisms, informing treatment choices and prognosis. Personalized treatment plans consider the patient's health and tumour characteristics, optimizing therapy while minimizing side effects. Clinical trials explore innovative treatments, and post-treatment surveillance detects recurrence early. Precision medicine enhances treatment effectiveness, survival rates, and quality of life for breast cancer patients, with ongoing advancements promising even more personalized care.

2.3. Machine learning

Machine learning (ML) is a transformative field of AI that has gained immense prominence in recent years. It empowers computers and systems to learn from data, identify patterns, and make decisions or predictions without being explicitly programmed. This capability has far-reaching implications across various industries, from finance to healthcare to autonomous vehicles and natural language processing. At its core, ML revolves around the concept of enabling machines to increase their performance on a task through experience, just as humans do. This experiential learning is achieved by feeding large volumes of data into algorithms that adapt and evolve over time. As these algorithms process more data, their ability to make accurate predictions or decisions steadily improves [9].

ML can be broadly categorized into supervised learning, unsupervised learning, and reinforcement learning, each with its unique applications and techniques. In supervised learning, algorithms are trained on labelled datasets, allowing them to make predictions or classifications based on input data. Unsupervised learning involves clustering and dimensionality reduction, uncovering hidden patterns in data without explicit labels. Reinforcement learning, on the other hand, focuses on decision-making in dynamic environments, where an agent learns by interacting with its surroundings and receiving feedback.

The applications of ML are virtually limitless. It has revolutionized fields like image recognition, speech processing, recommendation systems, fraud detection, and even scientific research. Its ability to analyse vast datasets quickly and accurately makes it an indispensable tool in modern data-driven industries. As ML continues to advance, it holds the potential to transform industries, enhance automation, and drive innovation. Its real-world impact is already evident in self-driving cars, personalized healthcare, and virtual assistants. However, it also raises important ethical and societal questions, such as those related to privacy, bias, and accountability. In this era of data abundance, ML stands as a powerful tool for extracting knowledge, making predictions, and enabling automation. Its evolution promises to reshape the way we live and work, offering exciting opportunities and challenges in the ever-expanding realm of AI [10].

2.4. Explainable artificial intelligence

XAI is a pivotal concept in the AI that focuses on one of the most pressing challenges of modern AI systems: transparency and interpretability. While AI technologies, particularly ML and deep learning, have made remarkable strides in various applications, they often imagined as "black boxes," making it challenging to understand how they make specific decisions or predictions. This opacity can be a significant barrier to trust, accountability, and broader adoption. XAI seeks to bridge this gap by developing AI models and systems that can provide clear and interpretable explanations for their actions and decisions. In essence, XAI aims to make AI more transparent, understandable, and accountable to both experts and non-experts alike [11].

The need for explainability in AI is especially critical in applications where the stakes are high, such as healthcare, finance, autonomous vehicles, and legal contexts. For instance, in healthcare, an AI model that assists in diagnosing diseases should not only provide accurate predictions but also explain the reasoning behind its diagnosis to physicians and patients. XAI encompasses various techniques and approaches, including rule-based systems, feature importance analysis, saliency maps, and model-agnostic methods. These methods help to uncover the inner workings of AI models, highlight influential factors, and clarify how input data influences the output.

By enhancing transparency and interpretability, XAI not only builds trust in AI systems but also has broader societal implications. It can help identify and mitigate bias in AI, ensuring fair and ethical decision-making. Moreover, it empowers users to challenge and correct erroneous AI decisions, contributing to overall accountability. As the deployment of AI technologies becomes more pervasive in our daily lives, the importance of XAI continues to grow. Researchers, policymakers, and industry professionals are working together to develop standards, guidelines, and best practices for achieving transparency and interpretability in AI systems. The journey toward XAI represents a crucial step in harnessing the potential of AI while ensuring it aligns with human values, ethics, and understanding.

3. RELATED WORK

Zhang *et al.* [12] introduces a model called breast imaging-reporting and data system network (Bi-RADS-Net) for cancer diagnosis in breast ultrasound images. The study focuses on integrating multitask learning with XAI techniques to enhance diagnostic accuracy and transparency. The model's innovative approach aims to improve breast cancer detection while providing interpretable insights into its decision-making process, contributing to the field of AI-driven medical image analysis and its potential for enhancing cancer diagnosis [12].

Qi *et al*. [13] investigates the intricate link between sarcopenia and distant metastasis in breast cancer patients using explainable ML. Employing transparent AI techniques, the study aims to reveal underlying associations by analysing comprehensive patient data through methods like decision trees and feature importance analysis. The research holds potential to uncover predictive biomarkers and enhance understanding of the disease, offering insights that could lead to personalized treatment strategies and advances in the field of AI-driven oncology research [13].

The literature survey on explainable ML for breast cancer diagnosis underscores the increasing focus on transparent AI methods to enhance the comprehensibility and reliability of diagnostic models. Various approaches, such as decision trees, feature analysis, and rule-based systems, are explored to elucidate the intricate decision-making of ML algorithms. This trend addresses the essential challenge of rendering AI-driven diagnoses understandable, fostering collaboration between medical practitioners and AI systems. The surveyed studies highlight the pivotal role of explainability in medical contexts, aiding clinical decision-making and patient communication. The integration of explainable ML in breast cancer diagnosis offers a promising avenue for bridging the gap between advanced AI technologies and transparent medical decision-making, with the potential to significantly transform and optimize clinical practices [14].

The literature review highlights the emerging trend of integrating XAI into breast cancer prediction, focusing on the innovative approach presented by Maouche *et al.* [15]. In this study, XAI is employed to predict breast cancer metastasis using clinicopathological data, exemplifying the fusion of advanced AI methods with clinical relevance. By unveiling the factors influencing predictions, XAI enhances diagnostic transparency and empowers clinicians to make informed decisions. The review underscores the transformative potential of XAI in revolutionizing medical practices, while acknowledging the broader research landscape exploring the application of XAI in cancer prediction and healthcare, ultimately reshaping the future of breast cancer care and precision medicine [15].

Kontham *et al*. [16] introduces a notable advancement in breast cancer prediction through an end-to-end XAI system. By enhancing the interpretability of predictive models, the study addresses the crucial need for transparent and comprehensible decision-making in medical AI. The integration of XAI techniques enables a deeper understanding of the factors influencing predictions, bridging the gap between complex algorithms and clinical insights. Situated at the convergence of AI, healthcare, and IoT, the research showcases the multidisciplinary nature of modern medical innovation. The authors' work underscores the transformative potential of transparent AI in improving breast cancer prediction models, emphasizing its significance for advancing diagnostic accuracy, patient trust, and overall healthcare quality [16].

Lamy *et al.* [17] contributes to the evolving landscape of breast cancer diagnosis by introducing a novel visual case-based reasoning methodology within the realm of XAI. This research addresses the pressing need for transparent and interpretable AI-driven diagnostic systems. Through visual representations of cases, the study pioneers an approach that aims to demystify complex AI decision-making processes, making them more accessible to medical practitioners. By seamlessly integrating XAI principles and visual reasoning techniques, the research enhances the comprehensibility of AI-derived breast cancer diagnoses, potentially fostering trust and facilitating informed clinical decision-making. This work aligns with the broader context of employing XAI to bridge the gap between advanced AI models and the imperative for transparent and actionable medical insights, underscoring its significance for advancing breast cancer care [17].

Massafra *et al.* [18] focuses on classifying invasive disease events in breast cancer using XAI. Their research not only achieves accurate classification but also emphasizes transparency through XAI techniques, enhancing the understanding of the AI model's decision-making. This work illustrates the growing importance of interpretable AI outcomes in healthcare, especially oncology, by bridging the gap between complex algorithms and actionable clinical insights. The study holds promise for advancing breast cancer care and precision medicine, highlighting the transformative potential of transparent AI in developing the future of medical diagnostics [18].

Amoroso *et al.* [19] presents a forward-looking roadmap for harnessing XAI to strengthen breast cancer therapies. The study offers a comprehensive overview of the potential applications and benefits of XAI in advancing the field of breast cancer treatment. By including transparent AI methodologies, the authors propose a promising approach to not only enhance the accuracy of therapies but also to elucidate the rationale behind AI-driven decisions. This roadmap underscores the pressing need to bridge the gap between medical practitioners and advanced AI algorithms, fostering trust and confidence in AI-powered treatments. The authors' wide review signifies the emergence of XAI as a transformative tool in oncology, paving the way for more personalized and effective breast cancer therapies. The publication of this study in a reputable scientific journal shows its significance within the academic community and its potential influence on shaping future research directions in the pursuit of improved breast cancer care [19].

Chakraborty *et al.* [20] presents a notable contribution to the field of breast cancer prognosis through the innovative application of XAI. Their study delves into uncovering novel insights into the tumour microenvironment conditions that correlate with improved prognosis for breast cancer patients. By harnessing XAI techniques, the authors offer a transparent and interpretable approach to unravel the intricate interplay between tumour microenvironment factors and patient outcomes. This work exemplifies the growing role of XAI in elucidating complex medical relationships, shedding light on previously unrecognized aspects of breast cancer prognosis. The study's findings not only emphasize the significance of the tumour microenvironment but also underscore the transformative potential of XAI in revolutionizing our understanding of cancer progression [20].

Chen *et al*. [21] provides a significant contribution to the domain of cancer diagnosis and treatment by harnessing the potential of AI in the era of precision medicine. The study focuses on the transformative role of AI in guiding clinical decision-making for cancer patients. By coupling the latest advancements in AI technologies, the authors showcase its capacity to enhance prognostic predictions, diagnostic accuracy, and treatment selection. The study underscores AI's pivotal role in unravelling the complexities of individualized patient care through precision medicine approaches. The integration of AI algorithms and predictive models empowers medical professionals with efficient tools for identifying optimal treatments, thus improving patient outcomes. Moreover, the authors emphasize the synergy between AI and precision medicine, highlighting its potential to revolutionize cancer diagnosis and treatment paradigms. The publication's placement in a reputable journal underscores its recognition within the scientific community, affirming its relevance in shaping the future of cancer care by amalgamating AI's capabilities with the principles of precision medicine [21], [22].

4. METHOD

4.1. Dataset preparation and machine learning model building

This study utilizes a genomic dataset sourced from breast cancer tumour samples [23]–[25]. This dataset consists of a total of 1198 samples and encompasses a rich array of information, including clinical data, mutated gene details, and treatment outcomes associated with breast cancer tumour samples. To smooth the data, disparate records were amalgamated into a unified file, enhancing usability. A meticulous cleaning process was undertaken, involving the removal of columns featuring a substantial number of missing values. Additionally, the tumour samples were meticulously categorized into distinct breast cancer subtypes, namely Luminal A, Luminal B, HER2+, and triple negative. This labelling scheme played a pivotal role in predicting appropriate treatments according to breast cancer subtype, culminating in more precise treatment prognostications.

The primary challenge at hand involves the prediction of optimal treatment strategies, an endeavour characterized as a multiclass classification problem. Consequently, a decision tree model has been judiciously selected to address this issue. The decision tree model is meticulously trained utilizing a consolidated dataset, where the target class pertains to specific treatments, encompassing chemotherapy, and hormone therapy.

4.2. Hyperparameter tuning

Within the context of the decision tree, the hyper parameter "max-depth" holds significance. To determine the optimal depth value for the decision tree, a process of cross-validation is executed on the dataset. As illustrated in Figure 1, a peak accuracy of 99.87% is achieved at a depth of 24.

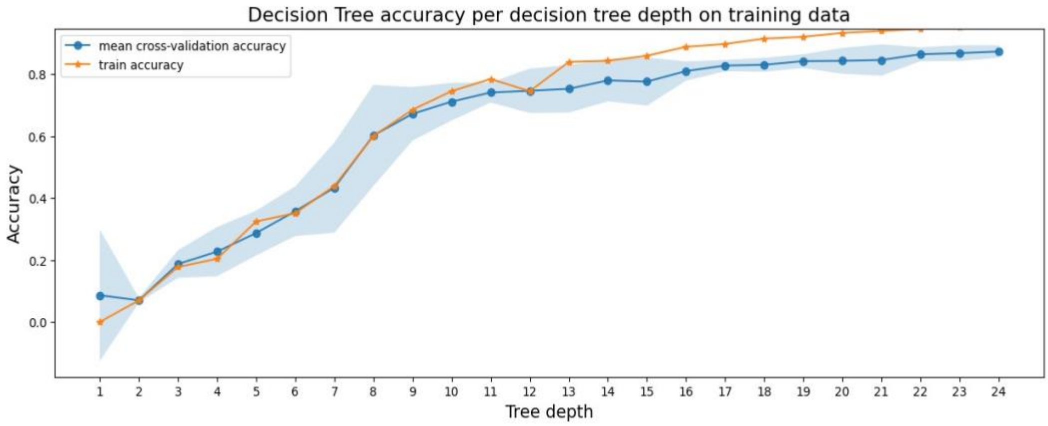

Figure 1. Line plot depicting cross-validation accuracy of the decision tree model

4.3. Decision tree model performance

A decision tree model is formulated with the objective of predicting appropriate treatments for patients with breast cancer metastases. The classification report, as depicted in Figure 2, provides insights into the performance of the developed decision tree model. Notably, the model demonstrates an impressive training accuracy of 99.87%, while achieving a commendable testing accuracy of 89.29%. Specifically, the model showcases precise predictions for various treatments: chemotherapy with a precision of 90%, hormone single therapy with a precision of 90%, anti-HER2 therapy with a precision of 97%, and hormone cyclin-dependent kinase 4/6 inhibitors (CDK46i) therapy with a precision of 80%. The classification report distinctly demonstrates the decision tree model's effective classification across all four treatment classes.

	precision	recall	f1-score	support
class Chemotherapy	0.90	0.99	0.94	342
class Hormonesingle	0.90	0.67	0.77	332
class Anti_HER2	0.97	1.00	0.99	351
class HormoneCDK46i	0.80	0.90	0.85	330
micro avg	0.89	0.89	0.89	1355
macro avg	0.89	0.89	0.89	1355
weighted avg	0.90	0.89	0.89	1355
samples avg	0.89	0.89	0.89	1355

Training Accuracy: 0.9987345776652958
Testing Accuracy: 0.8929889298892989

Figure 2. Classification report illustrating performance of the decision tree model

4.4. Explaining predictions of decision tree model

A significant challenge when employing ML models for healthcare predictions is instilling user trust. ML models often function as opaque black boxes, leaving users without insight into the rationale behind specific predictions. To address this, XAI technology has emerged, fostering interpretable models that establish transparency and foster user confidence. The local interpretable model agnostic explanation (LIME) technique is an XAI approach developed to illuminate predictions generated by decision tree models. An illustrative explanation produced by the decision tree model for the prediction of hormone single therapy is showcased in Figure 3.

LIME simplifies explanations into easily understandable bar plots, incorporating two distinct colour bars: red and green. This method leverages feature importance to craft explanations. Features that bolster the prediction—such as denovo metaplastic breast carcinoma (MBC) breast cancer type, prior_local_recurrence, overall tumour grade, Luminal A and Luminal B breast cancer subtypes, and mutated genes FOXA1, CDH1, NF1—are depicted in green, with their importance values presented on the far-left side of the plot. Conversely, other features counteract the prediction.

Consequently, it can be inferred that hormone single therapy could be recommended for metastatic breast cancer patients under specific conditions: when prior local recurrence is evident, the overall tumour grade is relevant, and the patient exhibits Luminal A or Luminal B breast cancer subtypes. Moreover, mutations in genes FOXA1, CDH1, and NF1 further strengthen this recommendation. This comprehensive approach enhances the trustworthiness of treatment recommendations, aiding healthcare practitioners in making informed decisions. Same explanation as given in the bar plot in Figure 3, can be given in notebook form as shown in Figure 4.

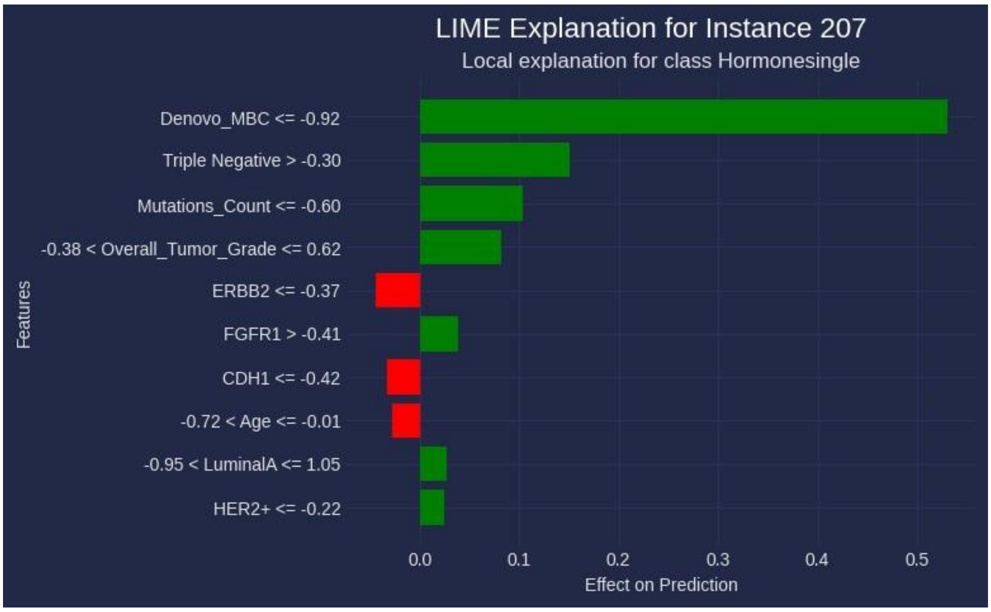

Figure 3. Bar plot explanation by LIME model

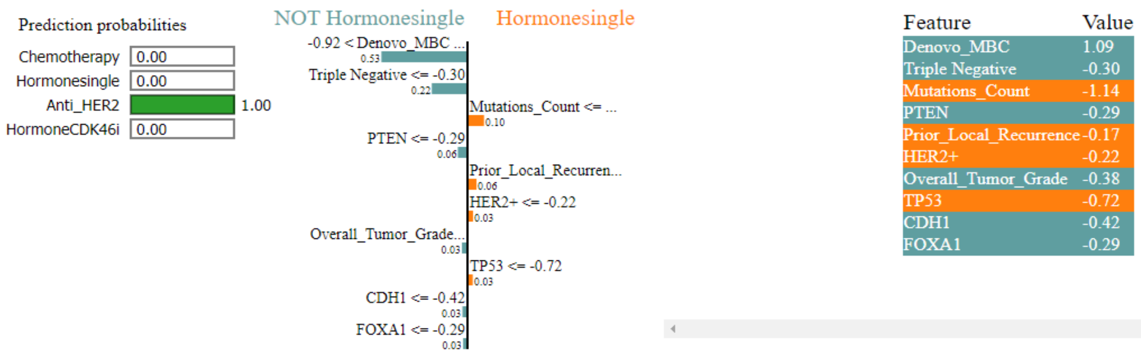

Figure 4. LIME explanation in notebook form

5. CONCLUSION

Breast cancer remains a prominent global health concern, spurring significant research efforts towards understanding its etiology and advancing personalized treatment strategies through the application of AI-driven precision medicine. Nonetheless, the inherent opacity of AI models has posed challenges in gaining widespread acceptance of their results. This study has delved into the realm of XAI, strategically integrating it into the landscape of breast cancer precision medicine recommendations. By embracing transparent AI models that draw insights from patient data, this research has successfully enabled the formulation of highly personalized treatment recommendations. Techniques such as feature analysis and decision trees have played a pivotal role in enhancing the transparency of AI-generated predictions. The establishment of this transparency bridges the crucial gap of trust between medical practitioners and patients, thereby harmonizing the immense potential of AI with the indispensable need for lucid and informed medical decisions. The focus on leveraging clinical and genomic data from metastatic breast cancer samples has culminated in the development of a robust ML model, primarily a decision tree, for the prediction of optimal personalized treatment. Remarkably, this model achieves an outstanding overall prediction accuracy of 99.87%, further affirming the efficacy of employing XAI principles in healthcare contexts. In the broader context of healthcare, the integration of XAI holds promise for fostering trust not only among medical professionals but also among patients. As the medical community advances towards the precision medicine era, the synergy between XAI and breast cancer care epitomizes a significant stride towards accessible, comprehensible, and effective treatment recommendations. Ultimately, this research demonstrates that the convergence of AI and XAI has the potential to reshape the landscape of medical decision-making, engendering a new era of informed and confident choices for patients and practitioners alike. However, the study lags testing the predicted results on actual patient's samples in real time. The research work shall be carried out in collaboration with the cancer hospitals or research centers.

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