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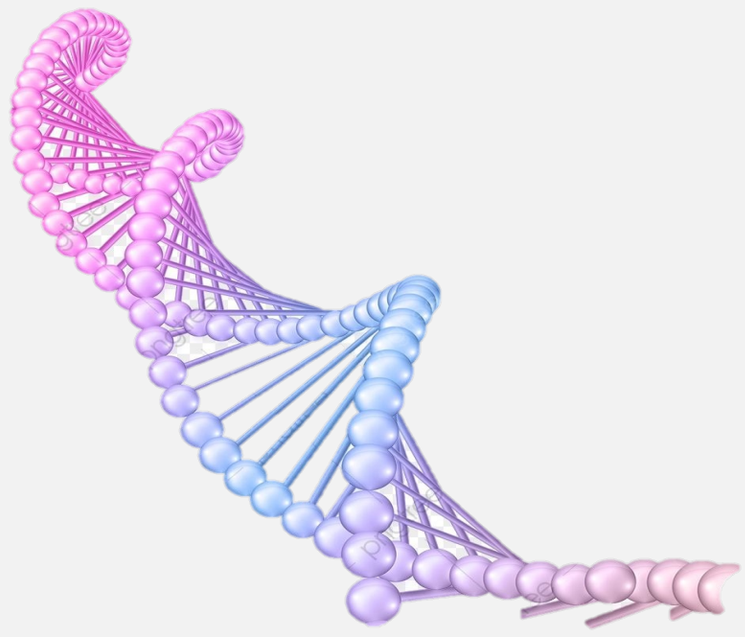
A Multi-Dimensional Challenge: A Clinical Case Presentation of an Elderly Lady with Triple Negative Breast Cancer, Metaplastic Carcinoma, Papillary Thyroid Carcinoma, and a Strong Familial Predisposition to Malignancy

Suniza Jamaris[1], YK Doon[1], LY Teoh[1], KL Ng[1]
[1] *Department of Surgery, Universiti Malaya, Kuala Lumpur, Malaysia*

INTRODUCTION

Individual with inherited cancer syndrome mutations are at a significant risk of developing tumours at a young age, as well as many tumours of the corresponding tumour spectrum developing simultaneously or metachronously. Numerous hereditary cancer disorders have already been linked to specific genetic origins. A hereditary cancer syndrome accounts for about 5% of all cancers.



CASE PRESENTATION

This clinical case presentation depicts the complex oncological journey of a 67-year-old woman with a notable medical history. Diagnosed with left breast cancer in 2007, characterised as triple negative, the patient underwent a comprehensive treatment regimen, including left breast-conserving surgery, axillary dissection, chemotherapy, and radiotherapy, leading to successful disease management. In 2006, she was also diagnosed with left shoulder liposarcoma, which was treated with wide local excision.

After 16 years of being cancer-free, the patient recently was presented with a left breast skin nodule. An excision biopsy confirmed spindle cell carcinoma, identified as metaplastic carcinoma, displaying oestrogen receptor (ER) negative, progesterone receptor (PR) positive, and HER2 negative status. However, a CT staging scan incidentally detected a suspicious left thyroid nodule, which a biopsy revealed to be papillary carcinoma. Given the rarity and distinctive features of metaplastic carcinoma, a simple mastectomy and a left hemithyroidectomy were performed in the same setting. As the medial margin was involved, a total thyroidectomy was done to address the thyroid malignancy effectively.

The clinical complexity is further heightened by the patient's substantial familial predisposition to malignancy. Her mother was diagnosed with a soft tissue malignancy at the age of 44, her brother with colon cancer at an early age of 26, and her sister with a soft tissue malignancy at age 52. Moreover, maternal relatives, including a cousin with breast cancer at age 40 and another cousin with colon cancer at age 30, accentuate the significant familial clustering of cancer.

DISCUSSION

This case underscores the significance of considering hereditary cancer syndromes in individuals with both a family history of cancer and multiple primary malignancies. A TP53 gene mutation linked to Li-Fraumeni syndrome could potentially be a factor given the diverse cancers presented, including breast cancer and sarcomas. However, it's vital to recognize that the interplay of genetic and environmental elements can drive the development of multiple primary malignancies. A thorough genetic assessment through counselling and testing is crucial to identifying specific genetic mutations. Analysing the patient's DNA can reveal known cancer-related gene mutations, aiding in assessing hereditary factors. Given the strong family history of malignancy, genetic testing becomes even more relevant. A personalised and vigilant approach to treatment and surveillance is essential in managing multiple primary malignancies, especially those with a notable familial predisposition. Understanding the genetic underpinnings can facilitate risk assessment and early cancer detection in at-risk family members, ultimately optimising patient care and outcomes.

Further research is essential to delve into the complex interplay between genetic susceptibility and the development of multiple malignancies in such intricate oncological cases.