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28606 – ASSOCIATION BETWEEN NEUROFIBROMATOSIS TYPE 1 AND BREAST CANCER: A CASE REPORT AND LITERATURE REVIEW

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Introduction: Neurofibromatosis type 1 (NF1), also known as von Recklinghausen's disease, is an autosomal dominant disorder characterized by the development of benign tumors of the peripheral nerves, known as neurofibromas. NF1 is caused by mutations in the NF1 gene, which encodes a protein called neurofibromin that regulates cell growth and proliferation. It is also known that individuals with NF1 have an increased risk of developing certain types of cancer, including breast cancer. **Methodology:** A systematic review of the literature was conducted using electronic databases, including the United States National Library of Medicine (PubMed), Scopus, and Embase, to identify relevant studies. The search terms included "Neurofibromatosis type 1," "breast cancer associated with genetic mutations," and "genetic neoplasms." Studies were included if they reported data on the association between NF1 and breast cancer or other neoplasms. The quality of the included studies was assessed using predefined criteria, and relevant data were extracted and analyzed. The studies encompassed a diverse range of populations and methodologies, including meta-analyses, literature review articles, case-control studies, cohort studies, and population-based cancer registry data. A total of 55 articles were selected. **Conclusion:** In conclusion, the association between neurofibromatosis type 1 and breast cancer represents a significant clinical challenge that warrants attention from both healthcare professionals and medical specialists. The link between NF1 and breast cancer requires further investigation to better understand the underlying mechanisms and potential implications for treatment outcomes. This clinical case illustrates the association between NF1 and an increased risk of breast cancer, as well as other neoplasms. This literature review, featuring important studies and authors and illustrated by this report, aims to clarify the connection between these two conditions, highlighting the complex interaction of genetic, molecular, and environmental factors. Genetic mutations in the NF1 gene, dysregulation of cellular pathways, hormonal factors, family history of cancer, and the presence of neurofibromas all play a role in increasing the risk of breast cancer in individuals with NF1. Understanding the underlying mechanisms of this association is crucial for developing effective prevention and treatment strategies for NF1 patients at elevated risk of breast cancer. Furthermore, breast cancer treatment in this population remains poorly defined. Patients with neurofibromatosis type 1 pose not only diagnostic challenges but also a unique surgical challenge for both the patient and the surgeon. Although breast-conserving therapy is an option for treating breast cancer, systemic radiotherapy may theoretically induce secondary radiation-related cancers in NF1 patients. However, given the scarcity of data in this patient population, randomized clinical trials are necessary to better elucidate optimal management strategies and the safety of surgical treatments in this group. Deepening our understanding of this association, improving outcomes for affected patients, and developing targeted interventions through a multidisciplinary approach involving mastologists, oncologists, and geneticists aim to reduce the risk of breast cancer in this population. Despite these findings and the classification of NF1 patients as high-risk individuals, current guidelines do not provide specific recommendations regarding screening programs for this category of patients.