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Myoclonic Epilepsy with Ragged Red Fibers syndrome and its potential link to breast cancer development: biological and clinical implications

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Objectives: To understand the possible link between Myoclonic Epilepsy with Ragged Red Fibers (MERRF) syndrome and breast cancer development. **Methods:** This study consists of a literature review, gathering pertinent articles from the last 15 years. The bibliographic research included articles written in English, selected according to their relevance. Exclusion criteria involved articles that presented patients with other significant comorbidities and those receiving active chemotherapy or radiation therapy. **Results:** MERRF is a rare mitochondrial disease with predominant progressive myoclonus. It is commonly present alongside myoclonic epilepsy, cerebellar ataxia, sensorineural deafness, short stature, cutaneous lipomas, and myopathy. Currently, there are no studies that defend a direct correlation between MERRF syndrome and breast cancer, but there are studies that suggest that mitochondrial defects may result in breast cancer. Breast cancer incidence and mortality have been consistently high among women, and recently, the role of mitochondria in breast cancer has received increasing attention. Mitochondria, as the powerhouse of the cell, are central to both the pathophysiology of MERRF and the development of cancer, including breast cancer. In MERRF, mutations in mitochondrial deoxyribonucleic acid (DNA) lead to energy deficits, oxidative stress, and disrupted cellular processes, which affect tissues with high metabolic demands, such as muscles and neurons. This mitochondrial dysfunction can contribute to the accumulation of abnormal cells and altered signaling pathways, processes that are also implicated in cancer initiation and progression. While MERRF itself is not directly linked to breast cancer, the defective mitochondrial function observed in MERRF patients might predispose them to cellular instabilities, potentially increasing the risk of malignancies like breast cancer. **Conclusion:** The intricate relationship between mitochondrial dysfunction, oxidative stress, and cellular transformation suggests that MERRF may share underlying mechanisms that also drive tumorigenesis, providing an intriguing area for further exploration in cancer research, since there are no studies that link these conditions.

Keywords: neoplasms; mitochondria; MERRF syndrome.