J Clin Case Rep 2017, 7:9 (Suppl)
DOI: 10.4172/2165-7920-C1-015



# Therapy related acute myeloid leukemia in a patient with heterozygous ataxia telangiectasia mutated gene mutations

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**Introduction:** Use of adjuvant chemotherapy has improved survival for patients with breast cancer. Unfortunately, agents that cause DNA damage in patients with mutations in repair genes may increase the risk of developing malignancies.

**Case:** A 36-year-old woman with heterozygous mutations in the ataxia telangiectasia mutated (ATM) gene presented with fatigue and scattered bruises. Three years ago, she was diagnosed with invasive ductal carcinoma of the left breast and had bilateral mastectomy with axillary node clearance. She received adjuvant chemotherapy that consisted of doxorubicin and cyclophosphamide followed by Paclitaxel and Trastuzumab. Lab tests revealed thrombocytopenia, leukocytosis, and anemia. Peripheral blood smear and bone marrow biopsy revealed numerous myeloblasts. FISH analysis showed rearrangement in the mixed-lineage leukemia gene (MLL). The patient was diagnosed with therapy related acute myeloid leukemia (t-AML) and treated with the "7+3" induction regimen. A bone marrow biopsy revealed residual disease and re-induction therapy was given. Complete remission was seen on a bone marrow biopsy and FISH analysis revealed no rearrangements. She received an allogenic stem cell transplant and was cured.

**Discussion:** This case highlights the risk of t-AML, which carries a poor prognosis, from certain chemotherapeutic agents for breast cancer. Induction of tumor death by chemotherapy can cause genetic arrangements that are difficult to correct. We raise the question as to whether topoisomerase II inhibitors and anthracyclines should be avoided in patients with mutations in the ATM gene. Furthermore, this case highlights why it is essential for patients and physicians to understand the risks and benefits of chemotherapy.

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