643

# Counseling Complexities of Expanded Carrier Screening: An ATM Case Report

Amy Cronister, Fiona Wallace, Lynne Heckman, Susan Cheney, and Kristi Fissell
Integrated Genetics, Laboratory Corporation of America® Holdings

Integrated GENETICS
LabCorp Specialty Testing Group

## I. Background

Patients undergo expanded carrier screening (ECS) to learn if they are at increased risk to have a child affected with a serious disorder. Carrier screening may also reveal potential clinical consequences for patients who are found to be carriers of select conditions on the panel. In this case report, we illustrate some of the complexities of counseling when a predisposition to cancer is identified following prenatal ECS.

## II. Clinical Sequence of Events

**Patient, 34 y.o., seen by reproductive endocrinologist (RE) for infertility workup**

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**Pretest counseling for ECS provided by RE**

- Informed consent included reproductive implications of the testing
- The patient has no recollection that she was informed of potential health implications for carriers of certain conditions on the panel

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**Genetic Counselor from the laboratory contacted the patient to disclose results**

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**ECS results for the patient revealed an ATM c.8977C>T (p.Arg2993X) pathogenic variant**

- Discussed Ataxia Telangiectasia (AT) phenotype
- Reviewed clinical information from laboratory report including AT heterozygotes at increased risk for malignancies, particularly breast cancer
- Managed initial patient concern that she underwent testing to identify prenatal risks, not personal risk for cancer
- Recommended comprehensive genetic counseling
  - Two sessions scheduled; one focused on reproductive issues, the other to address the hereditary cancer implications

## III. Case Report

This case report focuses on the discussion related to the hereditary cancer implications identified for the patient. The cancer genetic counseling session included review of the genetic testing result and the implications of a pathogenic ATM variant. During the discussion, the patient restated that she had undergone testing to learn about prenatal risks, not risks related to her own health, and that she felt unprepared to learn she was at increased risk for cancer.

Pedigree analysis was performed and revealed a maternal aunt diagnosed with osteosarcoma at age 64. Common characteristics of a hereditary cancer syndrome, the concept of cancer predisposition, and the patient's risks for cancer were discussed in the context of the patient's personal and family medical history. Inheritance of an ATM mutation and the benefits of sharing the patient's test result with family members were reviewed and psychosocial issues addressed as appropriate.

A critical part of the session was discussion of available screening and surveillance including recommendations outlined in the current National Comprehensive Cancer Network (NCCN®) Clinical Practice Guidelines: Genetic/Familial High-Risk Assessment: Breast and Ovarian. (See **Table 1**.) This allowed the patient to focus on how she could use the unexpected information to make thoughtful, proactive decisions about her health care.

**Table 1. NCCN Cancer Risks and Management Discussed with Patient**

| NCCN Risk and Management Recommendations for ATM Mutation Carrier | | |
|---|---|---|
| | **Risk** | **Management** |
| **Breast Cancer** | Increased risk | • Annual mammogram starting at age 40 with consideration of tomosynthesis and breast MRI with contrast<br>• Insufficient evidence for recommendation of risk reducing mastectomy |
| **Ovarian Cancer** | Potential increase in risk | Insufficient evidence for recommendation of RRSO |
| **Other Cancers** | Unknown or insufficient evidence for pancreatic or prostate cancer | |

## IV. Discussion

The cancer genetic counseling session, allowed the patient time to:

- Separate her reproductive concerns from her own health concerns, despite her frustration with the unexpected outcome of ECS
- Adjust to the implications of the result for herself and her family members, digest the information shared regarding predisposition to cancer, and explore her perception of risk
- Identify intervention strategies designed to manage her risk

## V. Conclusions

This case illustrates how unsettling ECS results can be when they reveal secondary results that have clinical implications for the individual's health and potentially for the health of his or her close family members. This patient was effectively managed through separate genetic counseling sessions addressing both reproductive and personal/family genetic health risks.

To improve the patient experience and minimize patient alarm, ECS pretest materials and informed consent should include discussion that results may reveal potential clinical consequences for carriers including increased risk for cancer.

## VI. References

NCCN Clinical Practice Guidelines in Oncology, Genetic/Familial High Risk Assessment: Breast and Ovarian. NCCN Guidelines Version 2.2019 – July 30, 2018. Available at https://www.nccn.org/professionals/physician_gls/PDF/genetics_screening.pdf.

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