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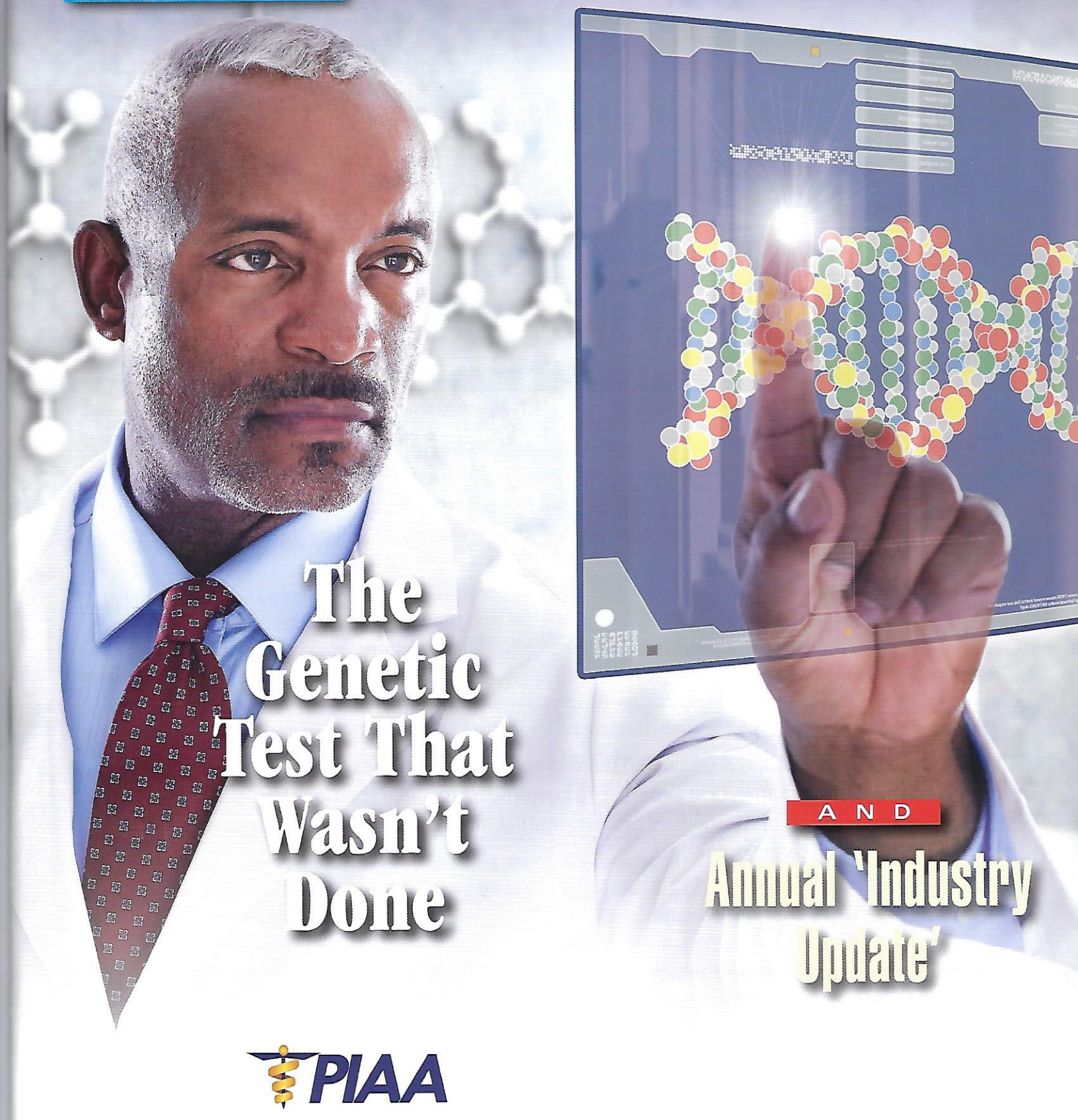

The Genetic Test That Wasn't Done

AND

Annual 'Industry Update'

PIAA

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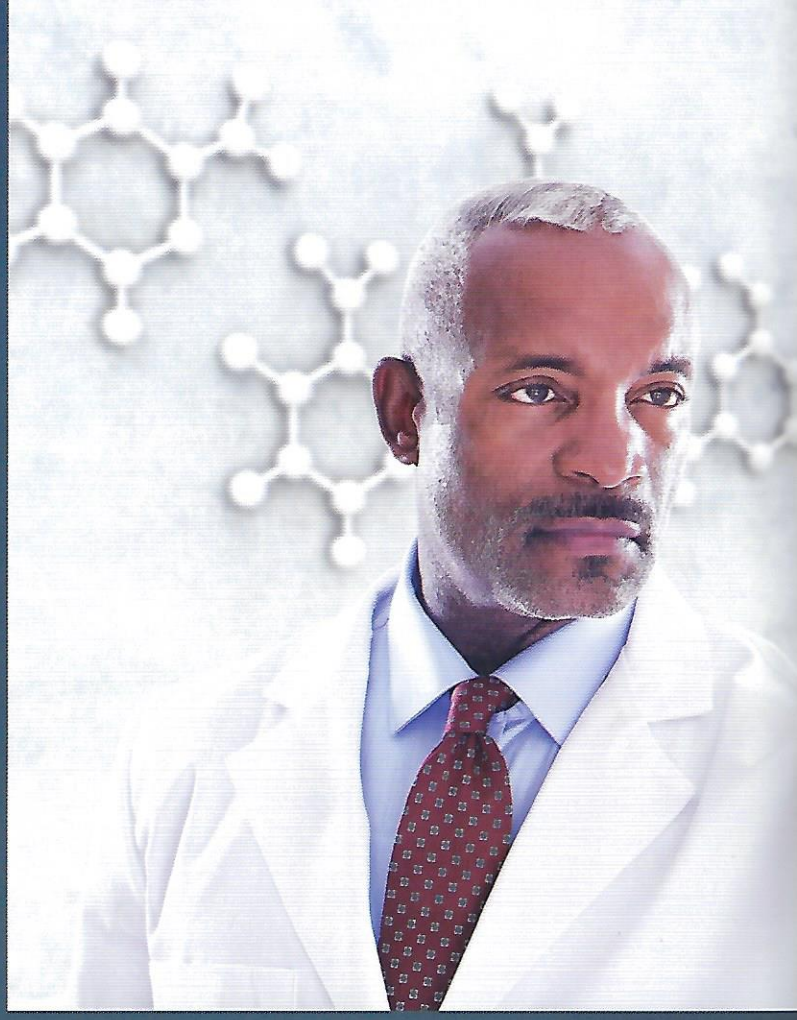
Failure to Recommend Genetic Testing:

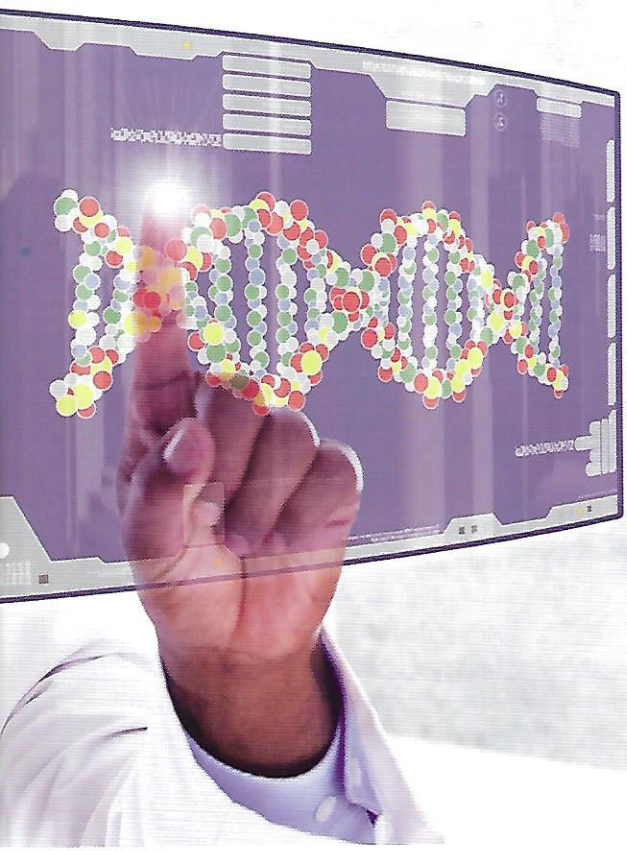
The Next Wave of Medical Professional Liability Lawsuits?



A 44-year-old woman was under the care of an obstetrician-gynecologist. She had no active medical problems and three children, ages 13, 11, and 9. Her maternal grandmother had died of breast cancer at age 64, and her mother had been treated for ovarian cancer.

Based on her family history, she had undergone yearly mammography for the past four years. The mammograms showed increased breast density, but were interpreted as negative for disease. Then, approximately nine months after her most recent mammogram, she discovered a lump in her left breast. She was diagnosed as having breast cancer; she underwent surgery, radiation therapy, and chemotherapy, but subsequently died of her disease, three years after diagnosis.

Her spouse filed a medical professional liability (MPL) lawsuit against the patient's obstetrician-gynecologist and the radiologist who had interpreted the mammograms, alleging a delay in diagnosis. At trial, the plaintiff's expert witness pointed to an anomaly on the patient's most recent mammogram that was located in the area where the cancer arose and testified that the mammogram had been misinterpreted. He also testified that the patient should have undergone genetic testing to determine if she had a *BRCA* mutation. The jury found against both physicians and awarded the family $4 million.



Delay in diagnosis of breast cancer is one of the most common, and most expensive, types of MPL lawsuits. According to PIAA data, although most patients who develop breast cancer are over the age of 50, most breast cancer-related lawsuits are filed by women who are under age 50.[1] This apparent paradox is explained by two factors. First, the disease is often more aggressive (and therefore more likely to be fatal) in younger patients. Second, plaintiff's attorneys prefer cases in which juries are likely to award large sums of money, as commonly happens when a case involves young children who lose a parent. Because the outcome of these lawsuits often hinges on the *retrospective* analysis of a mammogram, they can be very difficult to defend.

Although mammography is a useful screening tool, it is far from perfect. Mammographic sensitivity (the capacity of mammography to visualize cancer) is approximately 80% among women with predominately fatty breasts, but only 40% in women who have extremely dense breasts.[2] Because dense breast tissue is more common in younger women, mammography is least reliable in the patients who pose the greatest potential risk for an MPL claim.

To address this shortcoming, mammography is sometimes supplemented with sonography or MRI. Although these measures can help in detecting additional cancers, they may also reveal areas of concern that are not cancer but that necessitate additional imaging studies or biopsy, and thereby expose healthy patients to additional risks.

A potential solution is to change the nature of our approach in a fundamental way. Rather than waiting for a cancer to develop and then trying to detect it with sufficient time to successfully treat it, a much more effective approach would be to identify those patients who are likely to develop breast cancer and intervene before they do so. This is the promise of genetic testing.

Although most breast cancers are not related to the *BRCA* genes, patients who possess a pathogenic *BRCA* mutation have a 50% risk of developing breast cancer by age 50 and an approximate 85% risk of developing breast cancer in their lifetimes.[3] In terms of improving patient care and also reducing MPL exposure, it is imperative that these patients be identified.

To facilitate this, genetic testing for *BRCA* mutations is currently recommended by numerous entities, including the American College of Obstetricians and Gynecologists and the National Comprehensive Cancer Network. Because *BRCA* mutations are relatively rare (affecting approximately 1 out of 400 people in the general population), testing should be limited to patients who have positive family histories.[4] The basic criteria are summarized in Table 1.

Table 1. National Comprehensive Cancer Network: Basic Criteria for *BRCA* Analysis

- One first- or second-degree relative diagnosed with breast cancer at or under age 45
- One first- or second-degree relative diagnosed with ovarian cancer
- Two breast cancers on the same side of the family, one diagnosed in an individual under age 50
- Three breast cancers on the same side of the family, diagnosed in persons of any age
- One first- or second-degree relative diagnosed with triple-negative breast cancer at or under age 60
- Three relatives on the same side of the family with any combination of breast, ovarian, pancreatic, or prostate cancer
- Known *BRCA* mutation within the family.

Discrepancies in genetic tests

Although genetic testing holds great promise, it also raises several MPL concerns. First, it has been estimated that up to 14 million women in the U.S. meet the criteria listed in Table 1, of which only 1 million have been tested for *BRCA*.[5] This is problematic, because many of these women are *BRCA* positive, at high risk of developing cancer, and do not know it. Should they remain untested and untreated, develop cancer, and then file an MPL lawsuit, their cases will be nearly indefensible.

This was the situation in a Connecticut case from 2012; the case summary at the beginning of this article is based on it .[6] Given the risks to both patient and physician, it is imperative that patients who

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meet the criteria in Table 1 be tested.

The second concern derives from the fact that genetic testing is not regulated by the FDA. Although the labs that provide these tests are required to comply with the Clinical Laboratory Improvement Act (CLIA), each facility is permitted to perform the testing according to its own methodology, and there is no requirement that the results be validated against an external standard. Thus, the patient's result is somewhat dependent on the particular lab that performs the test. As an example, one study found that two labs interpreted 17 of 32 variants differently.[7]

Prior to 2013, the risks posed by this variation had been largely contained, because Myriad Genetics owned patents on the *BRCA* genes and did almost all of the testing. However, in 2013, the Supreme Court invalidated the patents and thereby opened the door for "generic" competition from other labs. Unfortunately, without the FDA guarantee of equivalency that usually accompanies generic versions, the stage was set for medical and legal complications.

Table 2. Factors to Consider in Evaluating Laboratory Quality

- Percentage of the gene evaluated
- Depth of the intron sequenced
- Database and algorithms used to interpret variants
- Analytical sensitivity and specificity
- Operating history, supporting data, and quality control measures
- Commitment to variant reclassification when new information is discovered
- Communication of both the initial result and any follow-up results

In an Ohio case, a 48-year-old woman underwent genetic testing at a local lab, and the result was positive for a *BRCA* mutation. Believing that she was at high risk of cancer, the patient had her breasts, uterus, and ovaries removed. When the patient's parents were tested to determine which side of the family carried the mutation, they both tested negative. The patient then underwent repeat testing, and an outside laboratory determined that she did not have a *BRCA* mutation, meaning that her surgeries had been unnecessary. She filed a lawsuit, and the case was settled for $2 million.[8]

Due to the significant variation in test results, and consequent potential for patient harm, the FDA is contemplating a regulatory structure that would standardize genetic testing. In the interim, the burden is (unfortunately) on clinicians to ensure that the laboratories they use are capable of producing accurate results. Factors to consider in making this determination are listed in Table 2.

The final concern of import for MPL is that the patient's health insurance company may not cover *BRCA* testing unless the specimen is sent to a lab of the insurer's choosing—typically selected because that lab is less expensive. As long as the physician is satisfied that the insurer's lab is of high quality, this is neither a medical nor a legal problem. However, physicians should not fall into the trap of presuming that they can simply follow the insurer's directive to use an inferior lab and that the insurer is the party that will be held liable for any harm that results. While there are certainly situations where a health insurer could be liable, a physician is never relieved of his duty to advocate on behalf of the patient.

As a result, when faced with a managed-care restriction that compromises patient care, physicians should weigh the degree of risk and assess the alternatives—and then make a reasonable effort to overcome the restriction, documenting their efforts. In some situations, the risk will be so minimal that no effort is required, while in others it will be great, and a significant effort required.

With respect to *BRCA* testing, an incorrect result is likely to cause devastating consequences (a false-positive, leading to unnecessary surgery, and a false-negative, leading to incorrect assurances that the patient is not at increased risk of cancer). Given the degree of risk for both patient and healthcare professional, it would be imprudent for a physician to simply follow a managed care company's directive and use an inferior lab.

Instead, physicians should either request permission to use another lab, find out whether the patient can cover the cost herself, or delay testing until the patient can make alternate arrangements. If none of these are viable, then using the insurer's lab may be the only choice. Although this would increase the risk of an incorrect result that compromises patient care and quite possibly leads to litigation, the physician would be in a highly defensible position, provided he has documented the efforts that were made.

Conclusion

"Failure to diagnose" genetic mutations that predispose patients to developing cancer are poised to become the next wave of MPL lawsuits. In order to limit the financial risk posed by the millions of women who meet the criteria for such testing but have not been tested, MPL/HPL organizations can educate their insureds about the relevant guidelines, the importance of maintaining a current family history of cancers, and the risks posed by both managed care restrictions and the lack of FDA oversight. This education should target both obstetrician-gynecologists and primary care physicians, as the latter provide some 20% to 30% of women's healthcare. PIAA

Law and Medicine has created a complimentary online CME activity, available at www.lawandmed.com.

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