DNA Specimen Provenance Assignment (DSPA) Test Confirms Identity of Transposed Specimens

**BACKGROUND**

After undergoing an ultrasound-guided breast biopsy at her physician’s office, a 54-year-old female patient (Patient A) was diagnosed with invasive ductal carcinoma in her right breast. Less than two months after the diagnosis was rendered a simple/partial mastectomy was performed to treat the cancer. However, when the extracted breast tissue was examined by a pathologist following the surgery, there were no cancer cells detected. These findings were inconsistent with the original biopsy and radiology results, which lead the pathologist to contact Strand to investigate the situation and help determine whether Patient A really had cancer.

**MATERIALS AND METHODS:**

Upon receiving notification of this discrepancy, Strand sent a Specimen Source Verification (SSV) kit to the pathologist so a DNA reference sample (buccal swab) and tissue samples could be collected for the patient in question (Patient A). A DNA Specimen Provenance Assignment (DSPA) test indicated that the four slides used to render the original diagnosis and the buccal swab findings were a complete DNA non-match.

Following these results, three additional SSV kits were sent out so Patient A could be re-swabbed for additional verification, along with the other two patients who were biopsied at the physician’s office the same day (Patient B and Patient C). Buccal swabs for these other two patients were tested against the original four malignant samples that were deemed a non-match for Patient A. In addition, samples of the extracted mastectomy tissue were also analyzed in this next round of testing.

**RESULTS:**

DNA Specimen Provenance Assignment (DSPA) testing confirmed that the original four tissue samples that led to a cancer diagnosis did not belong to Patient A (the benign mastectomy tissue, however, was a match). Results for Patient B also indicated a DNA non-match, and it was determined that the tissues in question actually belonged to Patient C.

It is unclear how this specimen transposition occurred, but regardless of the cause, this situation resulted in misdiagnosis and an unnecessary surgery for Patient A, as well as delayed treatment for Patient C who actually had cancer.

**CONCLUSIONS:**

A DSPA test was able to verify the provenance of the samples in question and enabled the cancer diagnosis to be assigned to the correct patient. Had testing been performed on a prospective basis using the patented know error® system, the error could have been identified prior to the unnecessary treatment of Patient A. The hospital involved in the case is now using the know error® system with all breast biopsies procedures to complete the diagnostic testing cycle and protect its patients from adverse outcomes.