Mitochondrial mutations in tumor and in nipple aspirate fluid

Breast cancer detection has been a concern of women for many years. Women are encouraged to manually check for masses on a regular basis and have an annual mammogram. This is an important practice due to the fact that early detection increases the likeliness of effective treatment. Unfortunately, breast cancer in its earliest stages is difficult to detect with the previously mentioned techniques. In addition, results of these tests are confirmed with invasive tissue biopsies. This procedure is painful and can have unexpected complications. New methods of non-invasive detection are currently being analyzed. One such technique is the analysis of nipple aspirate fluid (NAF). NAF is collected painlessly from the nipple using a modified breast pump. The fluid contains a mix of cells along with extracellular proteins found within the breast duct. The cell concentration of the NAF is unfortunately low making nuclear DNA collection difficult. One alternative is the analysis of the mitochondrial genome of the cells. This is a much more feasible approach due to the numerous mitochondria found within the cells each with its own circular segment of DNA. Mitochondrial DNA has also been shown to be an effective detection technique in other cancers. Three samples were collected from fifteen individuals diagnosed with breast cancer. For each patient, a skin tissue or blood sample was used for the collection of DNA from non-cancerous cells. In addition, portions of excised tissue known to contain cancer cells were used to collect cancerous DNA. The mitochondrial genome of both samples was then sequenced and analyzed for changes between the normal and tumor samples. This project found several undocumented polymorphisms along with a few areas of interest possibly useful for the detection of the presence of cancerous cells. These areas are currently being analyzed to determine if similar results may be recorded using the NAF samples.