Faster genetic testing method will likely transform care for patients with breast cancer

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Faster and cheaper DNA sequencing techniques will likely improve care for patients with breast cancer but also create challenges for clinicians as they counsel patients on their treatment options. Those are among the conclusions of a study published recently in the *BJS* (*British Journal of Surgery*). The findings provide insights into how genetic advances will soon be affecting patient care.

When a woman is diagnosed with breast cancer, it's important to know as much about her tumour as possible to determine the best treatment. Most cases of breast cancer are sporadic, but a minority are hereditary and caused by one or more mutations in genes such as BRCA1 or BRCA2. To find such genetic mutations in newly diagnosed patients, researchers must sequence the woman's DNA, which is generally a relatively slow process that generates results weeks or months after patients have started treatment. Next generation sequencing (NGS) is a newer method of sequencing DNA that processes large amounts of data. It's faster and more expensive than conventional sequencing, but in recent years it has become cheaper and more widely accessible by rapid advances in computing power. With the use of NGS, which will soon become the mainstay of clinical genetics, breast cancer units will likely be able to get the results of genetic testing before patients begin their breast cancer treatment.

In a collaboration between breast surgeons and medical geneticists at the

Norfolk & Norwich University Hospital and Addenbrooke's Hospital in Cambridge, UK, Simon Pilgrim, MD, and his colleagues searched the medical literature to identify relevant studies relating to breast cancer genetics and then looked to see what impact NGS will have on breast cancer units as well as what opportunities will arise for improving treatment for patients.

The researchers found that because NGS will allow breast cancer clinicians to know whether patients carry high-risk mutations (which might increase their risk of developing another breast cancer in the same or other breast in the future) before the start of treatment, more women might opt for mastectomy instead of breast-conserving surgery or for double rather than single mastectomy. Dr Pilgrim noted that some of the mutations that are detected might also confer increased risks of developing other cancers, which would indicate the need to monitor at-risk patients closely for these cancers as well. "The converse is also true," he said. "NGS can be used to find genetic mutations in people with other hereditary cancers or conditions. For example, a woman with ovarian cancer might be found to have a BRCA1 mutation, and hence breast units must be prepared to handle questions about the management of breast cancer risk in patients referred by other specialties." Dr Pilgrim added that NGS will have implications for the relatives of people found to have mutations linked to breast or other cancers, and therefore family members may also wish to undergo testing after appropriate counseling.

In the longer term, being able to identify patients with breast cancer who carry certain gene mutations before they start treatment will help researchers conduct clinical trials to establish which treatments are best for carriers of particular mutations. Dr Pilgrim noted that research already suggests that BRCA1-associated breast cancer is more responsive to certain chemotherapy drugs than sporadic breast cancers are.

More information: "Opportunities and challenges of next-generation DNA sequencing for breast units." S. M. Pilgrim, S. Pain, and M. Tischkowitz. BJS; Published Online: March 27th, 2014 DOI: 10.1002/bjs.9458