# Genetic profiling improves breast cancer prognosis and treatment

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In the late 1990s and early 2000s, improvements in molecular genetic technologies lead to the development of gene expression profiling tests. These tests provided specialists with an indication of the gene profile of individual tumours, and from there, an indication of the cancer prognosis.

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These tests were primarily developed in the United States and Western Europe and have become widely used to inform treatment recommendations for individuals where the more primitive, conventional parameters such as tumour size and involvement of lymph nodes lead are not helpful.

In developing countries, the tests are becoming more prevalent as they become more affordable and accessible.

## Study

A comprehensive breast centre in Cape Town has been using the tests for over a decade and has documented the clinical progress of over 150 patients diagnosed with breast cancer who had undergone molecular genetic profiling of breast tumours.

The tumours were selected for molecular genetic profiling as part of the normal consultative process. Patients older than 18 years with histopathologically confirmed early breast cancer, meaning that the tumour was less than 5cm across and had spread to a maximum of three axillary lymph nodes. In these patients, it is often not clear whether after the surgical part of the treatment had been done, they will benefit from the addition of chemotherapy. Oncologists are inclined to rather give chemotherapy than omit it such as not to compromise patient outcome.

Chemotherapy was recommended if the molecular genetic profile indicated a high-risk of relapse; conversely, chemotherapy was not recommended if the molecular genetic profile indicated a low risk of relapse. Each patient was consulted by team consisting of a surgical, medical and radiation oncologist team. Once the decision on further treatment was taken, established protocols were followed.

Over the last 11 years, these patients have been monitored and followed up to determine whether the individual treatment plan based on the genetic markers in the tumour resulted in a better outcome than standard treatment.

Out of over 150 patients, half were low risk based on the outcome of the molecular genetic profiling; all have survived without relapse despite not having had chemotherapy. Conversely, out of the high-risk group, despite receiving chemotherapy, three patients have relapsed indicating that the risk assessment with molecular genetic profiling was accurate. The major outcome of the study, however, was that about more than half of the patients that ordinarily would have received chemotherapy, could safely omit chemotherapy without compromising the outcome. Also, a few patients which ordinarily would not have received chemotherapy, were identified to have high risk tumours and received chemotherapy, a potentially life-saving addition to their treatment.

The outcome of this study confirmed that molecular genetic testing can reduce the number of individuals who undergo chemotherapy and also save lives due to a more aggressive treatment regime being prescribed for individuals who at first diagnosis would be seen as low risk. It gives medical professionals a new level of information from which to determine prognosis and treatment options and ultimately, allows for a more individualised approach to cancer prognosis and treatment.

## ABOUT THE AUTHOR

Dr Justus Apffelstaedt is a specialist surgeon with an interest in breast, thyroid and parathyroid health as well as soft tissue surgical oncology.