

## Development of a genetic database resource for monitoring of breast cancer patients at risk of physical and psychological complications

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**Background.** Although recent advances in treatment for breast cancer have improved survival rates, psychological morbidity remains a significant concern. Depression and anxiety are common complications and are exacerbated by the prospect of chemotherapy and the potential for tumour recurrence. A newly developed MammaPrint Prescreen Algorithm (MPA) to identify patients eligible for the 70-gene profile has allowed ~60% of early-stage breast cancer patients to forego chemotherapy due to greater confidence in long-term survival. The need to monitor health outcomes in breast cancer patients and obtain prospective data has been identified as an important priority.

**Methods.** In response to a needs assessment survey amongst healthcare professionals in South Africa, a pathology supported genetic testing (PSGT) service was established for breast cancer patients and linked to a health outcomes research project. The family history, clinical information, genetic and pathology test results of 150 breast cancer patients were captured using a secure online database. The genetic component included results from (i) the 70-gene MPA profile, (ii) BRCA1/2 mutation detection and (iii) a comprehensive wellness screen including CYP2D6, COMT and MTHFR genotyping.

**Results.** Analysis of patient data permitted comprehensive risk profiling for targeted health monitoring over time. A representative case, who underwent all 3 types of genetic tests due to a strong family history and bilateral tumours, was designated low-risk MPA, BRCA negative, with a functional CYP2D6 gene supporting the use of standard tamoxifen doses. However, genetic risk factors associated with a dysfunction of the methylation pathway and oestrogen metabolism were identified in both the index case and her daughter with breast cancer.

**Conclusion.** MPA may allow up to 60% of patients to forego chemotherapy, but consideration of additional information as part of a wellness screen may facilitate early identification of patients at increased risk of physical and psychological complications. PSGT using an integrated database for collating clinical information relevant to the laboratory tests performed *(i)* adds confidence in conservative treatment decisions, *(ii)* combines these results with genetic data for more effective health monitoring, and *(iii)* offers an accelerated translational research opportunity that may benefit breast cancer patients and their family members.