“Do I have cancer?” was the terrifying thought running through my mother’s mind after a follow-up phone call regarding her routine mammogram screen. It took two different types of invasive biopsies and a further scan over a 10-week period, before her specialist was finally able to give her the all-clear. This is a familiar tale for many Kiwis of today, however an alarming rate of us are on the receiving end of the terrifying positive diagnosis “I’m afraid you have cancer.”

Cancer is a disease that has been prevalent throughout history. Ancient Egypt had the oldest recorded breast cancer case and tumours have even been discovered within dinosaur fossils. Nevertheless, there is still no magic pill available for the cure of cancer despite the significant advancement in scientific knowledge. What is well established is that cancer arises from changes called mutations within the cell, primarily in the DNA. Understanding these changes in the DNA within the tumour will thereby allow identification of the processes causing tumour growth, treatment resistance and cancer spread to other parts of the body.

Acquiring tumour DNA via traditional means usually involves invasive measures such as surgical biopsies. More often, tumours can grow in places that are difficult and potentially dangerous for surgical removal. Furthermore, certain individuals may only present a detectable tumour via radiological imaging techniques at a very late stage of their disease, thereby limiting their treatment options.

Another more readily available source of tumour DNA is the blood. This is in the form of circulating tumour DNA (ctDNA), which consists of short fragments of DNA expelled from tumour cells into the circulatory system. ctDNA contains the same genetic alterations found within the tumour and can be readily acquired by a routine blood draw. Thus, ctDNA can potentially provide clinicians with a non-invasive means to accurately measure tumour burden, monitor patients’ response to treatment and even identify cancer spread throughout the body. Essentially, this provides a polaroid picture of the tumour’s molecular changes in a much shorter time frame than current clinical practices.
Although this concept has been proven successful by many different research groups over the last couple of years in a laboratory setting, its application in the clinical setting has been met with reservation. This is mostly due to the lack of validation and standardization of analytic techniques used for ctDNA analysis.

The purpose of my research is to develop a standardized protocol for ctDNA analysis as a diagnostic and surveillance clinical tool for cancer patients in New Zealand. This involves rigorously testing the methodology using synthetic ctDNA samples with known mutations to identify sources of variability for specific techniques such as droplet digital PCR and Next Generation Sequencing. Upon validation, this protocol will then be applied to cancer patient samples such as breast, colorectal, rectal and child-hood cancers.

Monitoring of mutations present in the blood plasma of patients serially obtained prior to chemotherapy will provide important information such as treatment resistance and tumour burden and more importantly specific genetic characteristics which leads to cancer spread. Ultimately, the application of ctDNA into a clinical setting will provide specialists with real-time molecular information in which clinical decisions such as treatment changes can be actioned quickly.

What is even more exciting is that, due to its simplistic acquisition method, cancer surveillance can be shifted away from secondary hospital care into primary care such as a General Practitioner’s office or health care centre. This will greatly benefit patients who may have difficulty accessing specialist care available in major hospitals such as those who live in rural parts of New Zealand. My study is part of the Healthier Lives National Science Challenge, governed by the Ministry of Health NZ. The aim of the challenge is to reduce the burden of non-communicable diseases such as cancer on our society by funding research to “deliver the right preventions to the right populations and the right treatments to the right patients”. One of the aims of the current New Zealand health strategy is to provide access to diagnosis and treatment of diseases such as cancer in a primary care setting. We are a little closer to achieving this world renowned innovative shift in health care service, as my research shows that blood will tell!