# Primer Extension Polymerase Chain Reaction for Mutation Detection in Cancer Genes

**(VA Reference No. 12-093)**

*Novel method for the detection of mutations in cell free blood DNA which could lead to early diagnosis of multiple types of cancer*

**Technology**

Primer extension polymerase chain reaction test that could be used for the detection of mutations in cell free blood DNA

**Inventors**

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**Key Features**

- PCR method for the detection of DNA mutations in cancer genes of interest
- Could provide early detection of cancer or tumor recurrence
- Non-invasive and requires only one milliliter of blood
- Highly sensitive in detecting mutations

**Stage of Development**

Successful demonstration in lung and cancer cell lines

**Keywords**

Primer extension PCR
Cancer diagnosis
DNA mutations
Cell free blood DNA

**Patent Status**

Patent application

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***Technology***

The presence of abnormally high levels of cell free blood DNA (CFDNA) in the plasma/serum of cancer patients has been demonstrated in research. Mutations in CFDNA have been characterized in a large variety of cancer types and sites, including colorectal, pancreas, lung, bladder, head and neck and liver cancers. In addition, various types of DNA alterations have been reported in CFDNA, including point mutations, DNA hypermethylations, microsatellite instabilities, and losses of heterozygosity. Since the majority of mutation detection techniques are based on sequencing, such methods are not adequate for blood DNA samples. In addition, whole genome sequencing is not applicable either for DNA mutation detection because it requires the sequencing of hundreds of normal genomes to find the mutated genome.

The Department of Veterans Affairs (VA) has developed a primer extension polymerase chain reaction (PE-PCR) method that could be used for the detection of mutations in CFDNA. The PE-PCR method is based on the amplification of the sequences containing the mutation using primers followed by additional amplification of the DNA using nested primers PCR. The method could potentially be used for mutation detection in most of the cancer genes of clinical interest as a non-invasive research tool for early detection of tumor recurrence, and possibly for early diagnosis of cancer when surgery can be curative.

***Opportunity***

Increasing demand for technologies supporting early and accurate diagnosis of life-threatening diseases, such as cancer, is driving the overall PCR market. PCR is increasingly being applied in key research that is expected to solve numerous medical challenges and breakthrough technologies. The global PCR products market is expected to grow from nearly $5 billion in 2010 to nearly $8 billion by 2015, at an annual growth rate of nearly 10 percent.

***Competitive Advantage***

CFDNA may provide a very valuable source of genetic material as a surrogate for molecular analysis in cancer and pre-cancer patients. The novel PE-PCR method has a number of competitive advantages including the ability to test for mutations, a basic requirement to achieve the benefits of personalized cancer therapy. Compared to tissue biopsy, the novel method provides an easily accessible and non-invasive modality to determine the genetic background of tumors. Furthermore, the PE-PCR method is more sensitive in detecting mutations.

***Status***

The Department of Veterans Affairs is looking for a partner for further development and commercialization of this technology through a license and the VA inventors are available to collaborate with interested companies through a Cooperative Research and Development Agreement (CRADA).