



# **Detection of Somatic Mutations**

Exposure to environmental mutagens, a number of which are carcinogenic, is a major health problem in today's surroundings. These deleterious agents elevate the likelihood of genetic damage that increases the probability of being affected by complex diseases such as cancer. This is especially true for people who are involved in professions that lead to regular exposure to occupational hazards such as asbestos, smoke, X-rays or radioactive radiation. Early detection of somatic mutations caused by these environmental harmful factors is essential for predicting the probability of cancer. Researchers at NSU have developed a diagnostic technology for detecting cancer by using frequency of gene mutations in Glycophorin A. This invention will offer health professionals a quicker, more accurate and affordable diagnostic tool. It is well established that early detection and therapeutic intervention of complex diseases such as cancer results in significantly better outcome. Therefore, this highly specific diagnostic assay will offer the opportunity of more efficient therapeutic intervention for these diseases.

#### Technology

Drs. Latimer and Grant have invented a novel method for detection of mutations that can be utilized as biomarkers for complex conditions such as cancer. This novel technology utilizes labeled specific antibodies to detect and quantify M and N alleles of Glycophorin present on the cell surface of red blood cells. Labelled antibodies targeting these M and N alleles are used on blood samples and are quantified using flow cytometry. This test is designed to determine mutation frequency of Glycophorin A alleles, detecting cells with somatic allele-loss phenotypes and quantifying cells with somatic mutations. It will serve as a rapid indicator based on physiological conditions that will allow the probability or existence or extent of a multifactorial complex condition such as cancer to be determined. This assay is not only efficient but also simple, accurate and has a relatively short turnover time. The diagnosis of genetic abnormalities will enable health professionals to predict the possibility of cancer and suggest appropriate preventive measures even before the onset of the disease.

## Application

- This diagnostic assay can be used to detect mutations that can be indicative of onset of cancer.
- Professionals working in environments that expose them regularly to mutation-inducing environmental factors can be checked frequently for mutation using this method.
- This immunoassay can be utilized to monitor effect of cancer therapies. It will be ideal for monitoring chemotherapeutic agents whose primary mode of action is DNA directed genotoxicity.



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#### **Advantages/Benefits**

- It requires a small volume (20-40 microliter) of blood sample from the subject and the result can be
  obtained within a day. HPRT gene mutation test that offers similar results require over 50 ml of blood
  and 2 weeks of time for analysis
- It is simple and affordable compared to complex molecular diagnostic assays that are used to detect mutations.
- This assay can be administered through a portable flow cytometer and does not require any large complex equipment, making it highly accessible.

#### **Status of Development**

The efficacy of this method for detecting somatic mutations has been tested on multiple subjects and the data has been published in peer-reviewed publications.

#### **Patent Status**

US Patent Application filed on 20 November 2015.

## Information on Inventors



• Dr. Jean J. Latimer is currently an Associate Professor at the College of Pharmacy and serves as the Director of the NSU AutoNation Breast and Solid Tumor Cancer Institute.



• Dr. Stephen Grant is an Associate Professor of Public Health at NSU's College of Osteopathic Medicine.

Both Dr. Grant and Dr. Latimer have published several peer-reviewed research articles and they have funded projects in areas of environmental public health, breast cancer and gene mutation.

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