Concept Clearance for RFA (SBIR/STTR)

Development of Genomic Technologies for Non-invasive Sample Collection Methods

National Advisory Council for Human Genome Research, February 2012

Purpose

The National Human Genome Research Institute (NHGRI) proposes an RFA to explore the development of technologies for cost-effective, comprehensive nucleic acids-based analysis of human samples in settings that benefit from facile, large-scale, non-invasive sample collection and cost-effective sample storage.

Background

Genomic technologies have advanced dramatically over the past decade to the point where the prospect of incorporating individuals' whole genome sequence information into their medical care is under serious discussion and careful study. Over the next several years, genome sequencing of large numbers of individuals in the context of clinical studies and ongoing medical care is expected to increase substantially the clinical impact of genomic sequence information. At the same time, the costs of collecting and interpreting genomic data are falling below the costs to conduct individual genetic tests.

Recognizing these trends, NHGRI and the Eunice Kennedy Shriver National Institute of Child Health and Human Development held a workshop in December 2010 to identify elements of a trans-NIH research agenda that could inform the possible application of new genomic technologies to newborn screening and child health. To address opportunities for using genomic sequence information to broaden understanding of diseases relevant to the newborn period, a U19 program, "High Throughput Genomic Analysis in Children with Newborn Screening Disorders" will be initiated concurrent to this RFA. It will focus on 3 coordinated areas: large-scale data collection and analysis, clinical research, and ethical, legal and social implications research. Recognizing the importance of technology development in this area and the potential for it to be conducted through small business mechanisms, NHGRI is proposing this parallel SBIR/STTR solicitation. Developing new, sophisticated and increasingly cost-effective techniques for DNA-based sequencing and analysis from non-invasive samples was specifically recognized at the workshop as a critical need for this field.

Importantly, other fields are in need of such technologies as well. Phlebotomy specimens, the typical source of genomic DNA for sequencing, have extensive and stringent requirements for collection, aliquoting, shipment and storage, typically at temperatures of -20 C or lower. Samples such as blood spots or saliva, however, can be stored at room temperature, are readily collected and shipped, and provide easy alternatives to whole blood collection. Currently available for monitoring of drug levels, renal function, diabetes, and lipid levels, blood spots are also of particular value in remote or under-resourced research settings where low-temperature transportation and storage are challenging. Technology permitting large-scale DNA sequencing

of such samples could be of considerable value in clinical and research settings that benefit from facile, non-invasive sample collection and cost-effective sample storage.

Research Scope and Objectives

This RFA would focus on development of novel, or adaptation and integration of existing, technologies for cost-effective, comprehensive nucleic acids-based analysis of samples from a variety of clinical and research settings. For all studies, maintaining or improving cost effectiveness, sensitivity, and quality at least equivalent to that which can be obtained from whole blood will be an important goal. This might include refining existing technologies to obtain DNA sequence, epigenomic, and/or transcriptomic data from samples including, but not limited to, saliva or blood spots. Other options may include developing sample collection methods (e.g., improved chemistry of the blood card/filter systems) that provide better sample quality for a wider variety of tests.

Awardees under this SBIR/STTR program will be expected to coordinate as needed with the related U19 “High Throughput Genomic Analysis in Children with Newborn Screening Disorders” and other NHGRI sequencing programs.

Mechanism of Support

This initiative would use the NIH SBIR/STTR (Small Business Innovation Research/ Small Business Technology Transfer) award mechanism. Three to five SBIR/STTR awards for phase I and/or phase II studies would be made.

Funds Available

NHGRI will commit roughly $1.5M per year for 3 years for awards using the SBIR and STTR mechanisms.