Population-Based Biobanks and Genetics Research in Connecticut

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A new era of genomics is upon us. Recognizing the power of a population-based approach to study genetic susceptibility for disease, and simultaneously the large number of samples required for these studies, recent discussions in the U.S. Congress, NIH, CDC, and FDA have raised the possibility of a national population-based biobank. This report reviews past and current events around the world that have led to these discussions, and identifies possible actions within the Connecticut Department of Public Health (DPH) to prepare for population-based biobanks.

DEFINITION OF A POPULATION-BASED BIOBANK

A biobank is the general term for a repository of biological tissue,[1] but biobanks can be further classified by a variety of characteristics, including tissue type, purpose, ownership, volunteer group, and size (**Table I**). An example of a biobank is that created from newborn screening blood spots by state newborn screening laboratories. These biobanks may be used for population-based determinations. Tumor banks are another class of biobanks, in which tumors from cancer patients are studied for biomarkers associated with disease. A biobank of umbilical cord blood contains donated cells from a recent pregnancy for use in transplantation and stem cell research. **For the purposes of this document, a population-based biobank is defined as a large repository of donated human DNA and/or its information, collected from volunteers with and without disease, which is used to identify genes that contribute to human disease**. Statewide and national population-based biobanks in the U.S. do not currently exist. Although many *privately owned* biobanks exist across the U.S., legislatively mandated *public* biobanks are more appropriate for population-based repositories and are currently in the formative stages of development.

Table I
Human Biobank Classifications

Tissue Type
Tumor tissue, cells, blood, DNA, or DNA array results

Purpose
Research, forensics, transplantation, or diagnostics

Ownership
Private, such as academic & medical institutions, hospitals, biotechnology & pharmaceutical companies, and biobank storage companies.
Public, managed in partnership with government

Volunteer Group
Population-based, such as all newborns, adults, or pregnant women
Disease-based, including only those with a specific disease

Size
Disease group, regional, statewide, or national

AROUND THE WORLD AND WITHIN THE U.S.

There is precedence around the world for large public population-based biobanks. National population-based biobanks now exist, or are being developed, in Estonia, Canada, Iceland, Japan, Latvia, Singapore, Sweden and the United Kingdom.[1-3] Whereas some of these biobanks involve the compilation of genetic, life style and genealogical information, other biobanks are more extensive, with links to individual medical records. These large biobanks range in size, seeking from 60,000 to as many as a million volunteers. All are public population-based biobanks, managed in partnership with the national government. A few also receive partial funding from research foundations. Many lessons have been learned from these international efforts that inform important considerations for future national biobanks. For instance, a planned biobank in the Kingdom of Tonga was unsuccessful because it awarded exclusive licensing to a private company without public consultation. In contrast, the population-based biobank in Estonia has been very successful, partly because its policies have been transparent and responsive to the public. The biobank infrastructure in the United Kingdom, perhaps in response to these contrasting events, has developed policies that provide for active public involvement.

Within the U.S., a national public population-based biobank is being considered. Population-based biobanks may also be discussed in the Connecticut state legislature. Proposed bills recently introduced at the federal or state level that involve biobanks include:

1) The PREEMIE Act (U.S. Senate Bill 707),[4] which was enacted into law on December 22, 2006 with 98 co-sponsors. If appropriated, the legislation allocates $3 million annually to the CDC from 2007 through 2011. Its purpose is to link population-based biobanks with medical records, health databases, and a follow-back statewide survey called PRAMS (Pregnancy Risk Assessment Monitoring Survey). This legislation could bring funding to Connecticut to support research in preterm births and birth defects. Also, although Connecticut is currently not included in PRAMS, the legislation could support this valuable on-going survey in the state.

2) Genomics and Personalized Medicine Act (U.S. Senate Bill 3822),[5] introduced in August, 2006, which proposes development or expansion of population-based biobanks to study genetic factors that influence drug efficacy. The legislation was introduced by Senator Barak Obama, and positive discussions about a national population-based biobank have occurred at the FDA,[6] CDC,[7] and NIH.[8] The proposed legislation also allocates funds to CDC and NIH to establish a central repository of genetic information from existing biobanks around the country. Funds allocated from this legislation might be used to establish a statewide population-based biobank in Connecticut that would contribute to a larger national biobank.

3) An Act Requiring DNA Testing for Newborns (Connecticut House Bill 5743),[9] introduced in January, 2007, which proposes DNA testing at birth and storage of genetic information in the birth record. The contents of this biobank, with appropriate legislation and safeguards, could also contribute to research that seeks to understand the genes that lead to childhood diseases.

BENEFITS AND CHALLENGES FOR CONNECTICUT

The potential benefits of population-based biobanks in Connecticut are significant. Private population-based biobanks already exist at research institutions and biomedical companies across the state, and are being used to further genetics research. A public statewide repository of genetic information, compiled from existing biobanks, or independently, could facilitate research

of the genetic susceptibilities that lead to many complex diseases and adverse health events. Possible funding from 1) the PREEMIE Act to link prenatal biobanks of public health departments, 2) proposed Senate Bill U.S. S.3822 to expand existing biobanks in the state to population-based biobanks, and 3) proposed Connecticut HB. 7543 to create a newborn DNA profile, could facilitate biomedical and applied genetics research within the state that could be further translated into increased public health and awareness among state residents.

Despite the many health, research, and fiscal benefits that are possible from public population-based biobanks, these repositories have a number of ethical, legal, and social implications (ELSI).[10] Some of these very important issues include proper informed consent procedures, protection of donor confidentiality, and regular public consultation.[1,3] It has been suggested that a "Biotrust" model could address many ELSI issues associated with population-based biobanks.[11] This model is based on the legal structure of a charitable trust, with management by a non-profit organization, and with oversight by an Ethics Review Committee and a Donor Advisory Committee. In addition to issues of informed consent, confidentiality and public consultation, the Biotrust model could be used to address other important considerations such as property rights and the right to withdraw from research. Applicability of a Biotrust model for biobanks within Connecticut has not been assessed.

POTENTIAL ROLE FOR THE CONNECTICUT DPH

As discussions about population-based biobanks move forward at the national and state levels, an informed public health community is needed to play a more active role in public consultation, and in the development of legislation and infrastructure needed to support population-based biobanks. To prepare for this possibility, key DPH staff need to become knowledgeable about a number of issues related to biobanks, including informed consent models, HIPAA regulations, current trends in genetics research, past and current litigation related to biobanks, and regulatory mechanisms (**Table II**).[1] State and federal legislation should be closely monitored for introduction of future biobank legislation. In addition, staff should become knowledgeable about the usefulness of the Biotrust model for population-based biobanks. With thoughtful preparation for possible population-based biobanks, DPH will remain at the forefront, among a handful of other states, in the field of public health genomics.

Table II

Public Health Preparedness for Population-Based Biobanks in Connecticut

Extracted and modified from Swede et al[1]

- Informed consent models and opt-out policies
- Litigation about existing biobanks
- Public consultation processes about biobanks
- Assessment of the Biotrust Model
- Establishment and management of tissue banks
- HIPAA and other federal privacy regulations
- Institutional Review Boards processes
- Federal and state legislation about biobanks
- Oversight and regulatory mechanisms of biobanks
- Trends in genetics research

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