Position Statement: Genomic Biobanking for Research

Background

Genomic biobanking for research is the storage of data generated from donated human biospecimens with linked clinical information, health or family history, or information generated from genetic analysis within structured resources for use in future genomic or genetic research potentially conducted by outside researchers (Annaratone et al., 2021; Coppola et al., 2019; Paskal et al., 2018). Analysis of large-scale genetic data combined with corresponding clinical data offers the potential for increased knowledge of disease processes that affect human health (e.g., Taliun et al., 2021). Genomic biobanks promote efficiency through multidisciplinary and multi-institutional efforts in the collection, management, and distribution of genomic and genetic data for future research use (Coppola et al., 2019). Complex biobank repositories have enabled swift evolution of scientific knowledge, necessitating careful ethical navigation of issues such as confidentiality and informed consent for unclear future use of genomic data and data sharing (National Cancer Institute [NCI], 2016; Paskal et al., 2018; Sotelo et al., 2021). Nurses across various settings will encounter individuals faced with decisions regarding genomic biobanking. Genomic biobanks are also rich data sources for nurse researchers. Therefore, nurses need to understand the benefits, risks, and ethical issues associated with applications of this technology to effectively educate, advocate for, and support individuals, families, and populations.

Benefits of genomic biobanking include the potential to gain knowledge through future genomic and genetic research on disorders that require large numbers of biospecimens and corresponding clinical data. Given the time between biospecimen banking and the subsequent analysis of aggregate data, future findings are more likely to benefit the greater society and future clinical populations than research participants who contributed data. Precision medicine, which incorporates the use of the human genome to predict disease susceptibility, disease prognosis, or response to treatment, is advanced through identification of genomic variation and highly powered association testing with large diverse biobanks (Carress et al., 2021). For biobanked data to advantage high quality health care that is inclusive, fair, and equitable across populations, these data must be representative of all races, ethnicities, and subpopulations, with rigorous methods for collection and annotation (Annaratone et al., 2021; Carress et al., 2021).

This position statement focuses on the ethical issues arising from genomic biobanking for use in future and ongoing genomic and genetic research and the responsibilities of nurses in the application of this technology.

Ethical Issues

Confidentiality Concerns. Potentially identifiable genomic and genetic information and ever evolving technologic capabilities challenge maintenance of participant confidentiality. Consequently, breach of participants' confidentiality is one of the major potential harms associated with genomic biobanking research (Bledsoe, 2017; Gymrek et al., 2013). This was the greatest concern identified by participants in a survey investigating public attitudes toward issues in biobanking research with 13,000 respondents from 11 United States (US) health care systems (Sanderson et al., 2017). These concerns apply to genetically distinct populations, as well (Algee-Hewitt et al., 2016). For these reasons, guidelines to promote the ethical conduct of research involving genomic biobanking for research and to protect the confidentiality of participants should be in place, despite variation in international legislation (Kasperbauer et al., 2018; NCI, 2016; Sotelo et al., 2021).

Informed Consent. The informed consent process documents the participants' indications of understanding study aims and their voluntary nature of participation. There are several key considerations for genomic research. The informed consent process for genomic research should provide potential participants with information about the scope of anticipated genetic research activities such as future data sharing, potential benefits and risks, whether future results will be disclosed, confidentiality protections, options for withdrawal of consent, and data ownership (National Institutes of Health [NIH], n.d.a.). Additionally, potential participants need to be aware that withdrawal of informed consent from future participation may be challenging due to the nature of broad data sharing of de-identified datasets (Paskal et al., 2018). The NIH Genomic Data Sharing Policy of 2014 further requires that investigators seeking funding obtain broad consent from participants of genomic research, which includes data sharing (NCI, 2016; NIH, n.d.b.).

The Genetic Information Nondiscrimination Act (GINA) of 2008 generally makes it illegal for health insurance companies and group health plans in the US to use genetic information in making decisions regarding eligibility or premiums and this information should be provided to potential participants (US Equal Employment Opportunity Commission [EEOC], n.d.). While GINA makes it illegal for employers with 15 or more employees to use an individual's genetic information when making decisions regarding hiring, promoting, firing, or setting terms of employment, those who work for small companies are not afforded these protections (EEOC, n.d.). GINA has other additional limitations related to military and associated health insurances (e.g., Tricare), and to genetic discrimination by companies that sell life, disability, or long-term care insurance (Green et al., 2015). Furthermore, GINA may be misunderstood by the public (Lenartz et al., 2021). State laws, such as Florida House of Representatives (2021) CS/HB 833

and The Florida Senate (2020) H.B. 1189 provide important additional protections and illustrate how policy can be shaped for continued improvement nationally and internationally.

Some US states allow researchers to conduct research studies using dried blood spots from state-mandated public health genetic newborn screening programs. This secondary research use after state testing for rare genetic conditions raises several issues. Ethically, one is that newborn individuals whose samples are used for research and subsequently donated to genomic biobanks do not provide informed consent, or even assent. These infants could face scenarios of having biobanked genomic data re-identified in the future for which they never provided informed consent for donation (Downie et al., 2021; Esquerda et al., 2020).

Data Ownership. Social, legal, and ethical issues related to biobanking data ownership remain incompletely resolved. Dynamic consent models have been proposed that use technology to allow participants to decide whether they agree to broad consent or prefer consent on a study-by-study basis. Broad consent imposes potential for secondary use of genomic data in which the investigators who are knowledgeable about the initial informed consent process may or may not be involved in subsequent research studies. Biorepositories must be ethically responsible for systematically protecting data collections (e.g., via use of an honest broker system) to the extent possible (Harati et al., 2019; Malsagova et al., 2020). Disagreement abounded in the past over ownership of biobanked data, including provider reach-through rights (Bledsoe, 2017; NCI, 2016). However, recent consensus is toward biobank custodianship rather than ownership of data, which complies with the concept that one person may not "own" another, as described in the Universal Declaration of Human Rights (Petrini, 2012; Sotelo et al., 2021). Ethical biobank governance structures also include considerations of potential commercial use of donated biosamples which result in patents and/or monetary profit (Paskal et al., 2018).

Data Sharing. Data sharing is the transfer of biobanked data, including biospecimens, health information, and/or any new data derived from the samples to researchers at another institution or biorepository not affiliated with the biobank or institution of origin (Garrison et al., 2016). Broad consent and data sharing confers less control to participants over their data's use and/or disposition. A general framework is included in broad consent for future research studies, which may be structured to allow wide interpretation. An example is, "research for the investigation of genetic influences on cardiovascular or other metabolic disease." Broad consent models may be acceptable due to the recognition of the importance of the secondary use of specimens, the limitations of specific consent models, and the logistical difficulties of tracking tiered consents and decision about what types of research fall within the scope of such consents. A potential issue of broad consent is that underrepresented racial or ethnic populations may be less willing to participate in health-related research studies with unclear future aims because of past research abuse (Barker, 2013; Claw et al., 2021; Garrison et al., 2016; Shavers et al., 2000).

Disclosure of Future Results. An additional consideration for the use of genomic biobanking data includes whether to inform participants about genetic findings. Even if genomic health information derived from such results proves to be clinically useful to participants, the procedures used to de-identify shared biobank data to maintain participant privacy and data confidentiality may complicate returning results to individual participants. Dynamic consent, which requires reconsenting participants each time their data are used, is one feasible method for the return of any actionable findings, as participant identifiers remain intact. Language about whether results will be disclosed during the initial research study or subsequent studies derived from biobanking must be included in the initial, and each subsequent (if any), informed consent form.

It is the position of ISONG that professional nurses will:

- Maintain knowledge about genomic biobanking research practices and related ethical, legal, and social issues.

- Incorporate methods to evaluate and integrate information about genomic biobanking research into curricula as able.

- Take opportunities to educate the public regarding the purposes, benefits, and risks associated with genomic biobanking research.

- Advocate for the protection of all human subjects in genomic biobanking, particularly for use in genomic research globally.

- Support the formation of trust and respect among researchers and potential participants in biobanking research via obtaining informed consent utilizing a sense of ethical and professional responsibility.

- Contribute to development of genomic biobanking protocols for the safe and ethical collection, management, storage, and dissemination of genomic and corresponding health data.

- Participate in legislation and policy formation for biobanking, especially when related to participant protections, equitable distribution of benefits, and diversity of population representation.

- Utilize expanded skills to:

    - Generate original and translational research through use of existing genomic biobanked data to advance the understanding of diseases and biobehavioral

human responses to diseases, as well as clinical practices that draw upon these findings.

- Spearhead new sample collection endeavors to contribute new genomic and health information data to biobanks for future research use to advance the understanding of diseases and biobehavioral human responses to diseases.

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