# Raising the Level of Nursing Involvement in the National Precision Medicine Initiative: An Example

Ukamaka Marian Oruche PhD, RN, PMHCNS-BC,

Janet S. Carpenter PhD, RN, FAAN,

Jamie Renbarger MD, MS,

Sydney E. Ross MS

## Abstract

**Purpose**: The Precision Medicine Initiative® (PMI) goal of ushering in a new and more effective era of health care that benefits all Americans requires two critical and interdependent components: a cohort assembly of one million or more Americans who reflect the diversity of the United States of America and an interdisciplinary workforce that includes nursing. The purpose of this paper is provide an example of nursing involvement in PM, specifically as related to gathering biospecimens (saliva) from vulnerable, under-studied adolescents with disruptive behavior disorders and their family members. **Source(s) used**: First, we provide a brief description of important concepts related to PM as well as current roles of nurses in PM. Then, we share lessons learned from our feasibility study aimed at increasing the diversity of our state-wide cohort assembly that has provided biospecimens for the Indiana Biobank. **Conclusion**: Nurses can definitely contribute to biobanks in support of the PMI. This paper is a call to action for nurses to take their rightful place in PM.

---

Raising the Level of Nursing Involvement in the National Precision Medicine Initiative: An Example

**Introduction**

The Precision Medicine Initiative® (PMI) is a national initiative aimed at generating empirical evidence to move precision medicine (PM) into clinical practice to optimize health outcomes for all Americans. PM is defined as an approach to disease treatment and prevention that seeks to maximize effectiveness by taking into account individual variability in genes, environment, and lifestyle (National Institutes of Health [NIH], 2015). The word medicine refers broadly to healthcare rather than solely to the field of medicine or to medications. The PMI goal of ushering in a new and more effective era of health care that benefits all Americans requires two critical and interdependent components. First, it requires a cohort assembly of one million or more Americans who reflect the diversity of the United States of America (USA) and are willing to volunteer to provide data, including biospecimens (NIH, 2015). Second, it requires all of the best research and clinical minds from all disciplines to be involved, including nursing (Calzone et al., 2013).

The purpose of this paper is provide an example of nursing involvement in PM, specifically as related to gathering biospecimens (saliva) and other health data from vulnerable, under-studied, volunteer adolescents diagnosed with disruptive behavior disorders and their family members. First, we provide a brief description of important concepts related to PM as well as current roles of nurses in PM. Then, we will share lessons learned from our feasibility study aimed at increasing the diversity of our state-wide cohort assembly that has provided biospecimens for the Indiana Biobank (NIH, 2015).

**Precision Medicine (PM)**

PM has the potential to revolutionize the way we address the burgeoning issues of chronic disease prevention and management, including mental illness. Nearly 50% of American adults are diagnosed with a chronic illness (National Academies Press [NAP], 2012), 25% have multiple chronic illnesses (NAP, 2012), and 13% seek treatment for mental disorders (Department of Health and Human Services, 2012). The number of American children with chronic illness has risen from approximately 2% in the 1960s to over 7% in 2004

(Centers for Disease Control, 2009).  PM aims to increase our understanding of genetic, environmental, and behavioral factors in order to maximize the effectiveness of preventive strategies, diagnosis, and treatment for chronic conditions, including mental illness. If fully achieved, precision medicine will help us fit not only the right drug but the right behavioral treatment to the right patients (Collins, 2015).

During the 2015 State of the Union Address, President Obama launched the PMI, dedicating $215 million to accelerate the science of PM to more quickly translate and usher in this new and more effective era of American healthcare (White House, 2015). In response, the NIH Director, Dr. Francis Collins, formed the PMI Working Group and charged it with developing the blueprint for creating and managing the requisite large research cohort of one million or more Americans referred to as the Precision Medicine Initiative Cohort Program or PMI-CP (NIH, 2015). The Working Group, in collaboration with key stakeholders and the public, has now made its recommendations to Dr. Collins. Importantly, the Working Group supports the goal to build a cohort of one million or more volunteers over the next four years who will agree to share their health data, provide biospecimens, and be re-contacted for future research. Of particular relevance to this paper, the Working Group recommends that the PMI cohort must reflect the diversity of the USA.

The inclusion of a cohort of diverse volunteers is critical to provide the platform for expanding our knowledge of PM approaches that will benefit all Americans. The inclusion of all Americans is particularly relevant to health disparity populations and to segments of our population that are often left out of research and/or practice. According to Healthy People 2020, health disparities adversely affect groups of people who have systematically experienced relatively greater obstacles to health based on their racial or ethnic group; religion; socioeconomic status; gender; age; mental health; cognitive, sensory, or physical disability; sexual orientation or gender identity; geographic location; or other characteristics  historically linked to discrimination or exclusion (Healthy People 2020). Even more than other areas of health and medicine, the mental health field is plagued by disparities in the availability of and access to its services (Pearson et al., 2014). We join Pearson

and her colleagues' (2015) call to action focused on transforming mental health care and outcomes for all Americans particularly in light of the opportunities offered by PM.

**Nursing and PM**

Nurses have been largely missing from the PM/PMI discussion table. In one paper specifically related to pharmacogenetics as one aspect of PM, authors reported that the available nursing literature consisted of mainly narrative reviews with limited discussions of implications for nursing practice, education, or research. The paper concluded with a resounding call to action for nurses to become involved in and contribute to interdisciplinary conversations and initiatives related to personalized health care (Kinsley, Carpenter, and Von Ah, 2013). In addition, there were no appointed nurse members of the PM Working Group and, unless invited stakeholders include nurses or individuals who are able to adequately represent nursing, the nursing perspective will be absent. Nursing involvement is crucial given that nurses (1) have been identified as the most trusted health care providers (Calzone et al., 2013; Gallup, 2015), (2) constitute the largest proportion of the healthcare workforce, and (3) have the most direct contact or interactions with patients and their families.

Nurses routinely reach out to underrepresented and vulnerable populations both through their research and in their clinical practices. Within their research and practice, nurses can make tremendous contributions in ensuring that a diverse volunteer pool is included in the PMI Cohort Program. Although attention to PM is relatively new for nursing compared to medicine, nurses are interested in advancing their science in this area. For example, PM is cited as particularly relevant to symptom science research on the National Institute of Nursing website (NINR, 2016). However, many nurses may be apprehensive or have questions about how and where to start and what kinds of contributions can be made.

**A Nurse-led Study Contributes To a State-wide PM Cohort Assembly**

The authors conducted a nurse-led study to explore the feasibility of collecting biospecimens (saliva) in combination with behavioral data from adolescents with disruptive behavior disorders and their family members. The goal was to expand the racial and diagnostic diversity of our state-wide PM cohort assembly (i.e.,

the Indiana Biobank) to ensure that data and biospecimens from this population were banked for the purpose of being used in future analyses. Our focused genetic association study was part of a larger mixed-method descriptive study of families of adolescents with disruptive behavior disorders that has been described in detail elsewhere (Oruche et al., 2014). Very early on in the planning stages of the study, a senior nurse researcher encouraged the principal investigator to collaborate with a genomics expert. Together, they prepared an institutional review board application that included an optional saliva sample collection (or genomic study) for biobanking. The PI and her research team recruited members of families of adolescents diagnosed with disruptive behavior disorders who were receiving care in the child and adolescent clinic of a large, publicly funded, Midwestern mental health center from October 2012 to April 2013. The larger study included 52 individuals from 15 different families. We gathered data at one time point using self-report questionnaires and in-depth interviews. We obtained parental consent and assent for children under 18 years old to participate.

Subjects who were adolescents with disruptive behavior disorders and/or their family members (e.g., mother, father, and/or sibling) consented to allow collection of up to 10 mL saliva. For each subject, the saliva sample was collected into an Oragene®•DNA saliva collection cup (DNA Genotek Inc., Kanata, Ontario, Canada). This collection kit contained a buffer that kept the DNA in the saliva stable at room temperature until the DNA could be extracted. Research staff sealed the cup to prevent evaporation, labeled it with the participant identification number, and transported it to the biobank for storage.

**Lessons Learned**

Not surprisingly, some study participants declined to participate in the optional genomic study. Of the 52 individuals recruited in the larger study, 40 provided saliva samples. The most common reasons given for declining participation were related to privacy and security of data, fear of government intrusion including impact on their health coverage, and concerns about how data might be used in the future. Even though we clearly articulated reasons for collecting the biospecimens, how they would be stored and used in the future, and how the findings might benefit them or others, 12 participants still declined.

We provide a few practical suggestions about how to increase participation in genomic studies. Transparency and communication are critical to gaining the trust of participants. First, as a key part of transparency, it is important to tell participants that, although efforts will be in place to safeguard their information, as with all research, data may very rarely be exposed, and should that happen they will be immediately notified. It is not helpful to disregard or minimize participants' concerns about the confidentiality and privacy of their data. Based on our experience, participants' conversation around confidentiality and privacy can be passionate and this takes some time. Therefore, allow adequate time to describe the genomic study and answer participants' questions.

Second, investigative teams must understand why they are collecting specimens and be able to explain and communicate this effectively to participants. It will be helpful to prescript this in very clear and simple or lay terms. The communication can be enhanced by recording a video of the same message to use as needed. Given that PM is a heavily patient-driven initiative, the video could show a previous participant sharing why he/she chose to participate. Experts contend that health literacy will be a key defining factor in recruiting a large and diverse cohort (Parker, Bakken, & Wolf, 2016). Equally important is the use of culturally sensitive language (Parker, Bakken, & Wolf, 2016). For example, it will be important to highlight the significance of including the health disparities population, including persons with mental illness and persons from ethnic minorities. Participants need to know the risks and benefits of participation. We need to explain that we have new and excellent methods for understanding how our genetic makeup, family history, and where we are born, live, learn, and work affect our health. But we need to extend this knowledge to other health conditions or diseases or patient populations that we currently do not know how to treat successfully. Therefore, we need to gather as much of this information as possible from many people. Finally, we need to explain that, though the study findings may not benefit them directly, they will likely benefit other people who look like them or share similar health conditions.

We also learned that the success of our project required collaboration with genomic experts. The unique contributions of the genomic expert included conceptualization and development of protocols for collection of

the biospecimen, provision of materials for collecting and storing the biospecimen, provision of storage in a laboratory facility for future DNA extraction, as well as data analyses. Though we had the support of the Clinical and Translational Sciences Institute at our university, the current guidelines from the PMI Work Group were not available at the time of our study (NIH, 2015). Nurses who wish to contribute to biobanks should align their protocols with the PMI Working Group recommendations and guidelines (NIH, 2015).

In our research project, those who provided saliva samples were 50% African American, demonstrating that it is possible to recruit a diverse cohort of volunteers, including ethnic minorities who are typically underrepresented in research. All of the adolescents with disruptive behavior disorders provided their saliva samples. The inclusion of not only these adolescents but also most of their family members is very encouraging for PMI because individuals with mental health problems and their family members are often underrepresented in research for a variety of reasons. For individuals diagnosed with the mental illness, reasons include lack of cognitive capacity or impaired judgement. For family members, reasons for underrepresentation include competing demands, limited access to information about such research projects, or other socioeconomic disadvantages such as lower health literacy (NIH, 2015; Parker et al., 2016). Nurses typically have access to diverse populations and therefore must leverage these opportunities to educate, invite, and include all patients and their families to be part of the PMI. This is the only way to ensure that knowledge generated from the science of PM is generalizable to all Americans, notably including diverse groups affected by mental illness (NIH, 2015; Parker et al., 2016).

**Conclusion**

Nurses can definitely contribute to biobanks in support of the PMI Cohort Program. We successfully gathered saliva samples within the context of a larger research project – a process very similar to current funding opportunities in the pilot phase of the PMI to build biobanks. Few investigators devote their research solely to genomics or precision medicine, and nurses are particularly lagging behind in this area of research. As nurses, we do not have to change our program of research or practice. PMI simply calls us to contribute within the context of primary research or practice – using these as venues to educate, invite, and include our diverse

patients in precision medicine. Nursing's contributions have the added benefit of creating potential pathways for future nurse scientists in genomics. Building the next generation of an excellent and diverse scientific workforce is a priority for the NIH. This paper is a call to action for nurses to take their rightful place in PM.

References

Centers for Disease Control and Prevention. (2009). *The power to prevent, the call to control: At a glance*. Retrieved from http://www.cdc.gov/nccdphp/publications/AAG/pdf/chronic.pdf

Calzone, K. A., Jenkins, J., Bakos, A., Cashion, A. K., Donaldson, N., Feero, G., … Webb. J. (2013). A blueprint for genomic nursing science. *Journal of Nursing Scholarship*, 45(1), 1-9.

Collins, F. (2015, October). Precision medicine: Advancing science, improving lives. Paper presented at the NINR's 30th Anniversary Scientific Symposium, Bethesda Maryland.

Gallup (2015). Social Issues: Nurses still number 1. Retrieved from http://www.gallup.com/poll/187874/americans-faith-honesty-ethics-police-rebounds.aspx

Department of Health and Human Services, Substance Abuse and Mental Health Services Administration. (2013). Results from the 2012 *national survey on drug use and health: mental health findings*.

Healthy People 2020. *Disparities*. Retrieved from http://www.healthypeople.gov/2020/about/foundation-health-measures/Disparities

Institute of Medicine. (2012). Living well with chronic illness: A call for public action. Washington, DC: National Academies Press. Retrieved from. http://iom.nationalacademies.org/~/media/Files/Report%20Files/2012/Living-Well-with-Chronic-Illness/livingwell_chronicillness_reportbrief.pdf

Knisely, M. R., Carpenter, J. S., & Von Ah, D. (2014). Pharmacogenomics in the nursing literature: An integrative review. Nursing Outlook, 62(4), 285-296. Retrieved from http://dx.doi.org/10.1016/j.outlook.2014.03.004.

National Institutes of Health. (2015). *The precision medicine initiative cohort program: Building a research foundation for 21st century medicine*. Retrieved from http://acd.od.nih.gov/reports/DRAFT-PMI-WG-Report-9-11-2015-508.pdf

Oruche, U.M., Draucker, C., Al-Khattab, H., Knopf, A., & Mazurczyk, J. (2014). Interventions for family members of adolescents with disruptive behavior disorders. *Journal of Child and Adolescent Psychiatric Nursing*, 27(3), 99-108. doi: 10.1111/jcap.12078. PMCID: PMC4199581

Parker, R. M., Bakken, S., & Wolf, M. S. (2016). Getting it right with precision medicine initiative: The role of health literacy. Commentary, National Academy of Medicine, Washington, DC: National Academies Press. Retrieved from http://nam.edu/wp-content/uploads/2016/02/Getting-it-Right-with-the-Precision-Medicine-Initiative-the-Role-of-Health-Literacy.pdf

Pearson, G., Hines-Martin, V., Evans, L., York, J., Kane, C., & Yearwood, E. (2015). Addressing gaps in mental health needs of diverse, at-risk, underserved, and disenfranchised populations: A call for nursing action. Archives of Psychiatric Nursing, 29(1), 14-18.

White House. (2015). The precision medicine initiative. Retrieved from https://www.whitehouse.gov/precision-medicine

National Institute of Nursing Research (2016). Precision medicine and NINR-Supported nursing science. Retrieved from http://www.ninr.nih.gov/researchandfunding/precisionmedicine#.VumvOk0VHIU