Contributors

Genomics Nursing: Scope and Standards of Practice, Third Edition, was produced through extensive revisions to the *Genetics/Genomics Nursing Scope and Standards of Practice*, Second Edition (2016). In the intervening years, a multitude of changes have occurred in terms of genetic and genomic capacities, national and global sociopolitical contexts, and the nursing specialty. This document is reflective of those changes as envisioned and translated by a workgroup of experienced genomics nurse specialist clinicians, academicians, researchers, educators, and policy leaders. Our workgroup was guided by a deep commitment to fostering the development of genomics nursing toward a more equitable future for nurses, patients, families, communities, and populations within the context of advancements in precision health.

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ABOUT THE INTERNATIONAL SOCIETY OF NURSES IN GENETICS

The International Society of Nurses in Genetics (ISONG) is a global nursing specialty organization dedicated to fostering the scientific and professional growth of nurses in human genetics and genomics worldwide.

The vision of ISONG: Caring for people's genetic and genomic health throughout the lifespan and across the continuum of health and disease (ISONG, 2022).

The mission of ISONG is to serve both the nursing profession and the public. ISONG fosters and advocates for the scientific and professional development of its members and the nursing community, in the discovery, interpretation, application, and management of genomic information, for the promotion of the public's health and wellbeing. ISONG advocates for public understanding of genomic health and use of genomic information.

ABOUT THE AMERICAN NURSES ASSOCIATION

The American Nurses Association (ANA) is the premier professional organization representing the interests of the 5 million registered nurses in the United States. ANA advances the nursing profession by fostering high standards of nursing practice, promoting the rights of nurses in the workplace, projecting a positive and realistic view of nursing, and by lobbying the Congress and regulatory agencies on healthcare issues affecting nurses and the public. ANA is at the forefront of improving the quality of healthcare for all. Founded in 1896, and with members in all 50 states and United States territories, ANA is the strongest voice for the profession (ANA, n.d.).

ANA represents the interests of all registered nurses and exists to advance the nursing profession by

- Fostering high standards of nursing practice,
- Promoting a safe and ethical work environment,
- Bolstering the health and wellness of nurses, and
- Advocating on healthcare issues that affect nurses and the public.

Scope of Genomics Nursing

The International Society of Nurses in Genetics (ISONG) represents genomics nurses worldwide and is the official professional organization of nurses in genomics in the United States. ISONG is responsible for defining and establishing the global scope of professional nursing practice in genomics. In keeping with ISONG's responsibility, this document broadly describes genomics nursing practice and then delineates the scope of genomics nursing in the United States.

The professional code of ethics that governs practice in the United States (ANA, 2025) holds that health is a universal human right; therefore, the need for nursing is universal. All nurses commit to advancing health, welfare, and safety to achieve and sustain health so that persons and communities develop to their fullest potential and live with dignity. ISONG and ANA recognize that this document derives from a United States-centric perspective and acknowledge that nurses outside of the United States may have scope and standards of practice that vary from this document. In these cases, the International Council of Nurses' (ICN) documentation on standards of ethical professional nursing practice helps provide clarification for genomic healthcare applications for all nurses at the global level (International Council of Nurses, 2021).

DEFINITION OF GENOMICS NURSING

Genomics nursing integrates nursing with the rapidly expanding field of precision healthcare in all contexts. Defining genomics nursing therefore requires an understanding of nursing, genomics, and precision healthcare:

- Nursing refers to "the diagnosis and treatment of human responses and advocacy in the care of individuals, families, and groups, communities, and populations in the recognition of the connection of all humanity" (ANA, 2021, p. 1).

- Per ICN, nursing "encompasses autonomous and collaborative care of individuals of all ages, families, groups and communities, sick or well and in all settings. Nursing includes the promotion of health, prevention of illness, and the care of ill, disabled and dying people. Advocacy, promotion of a safe environment, research, participation in shaping health policy and in patient and health systems management, and education are also key nursing roles" (ICN, 2022).
- Genomics refers to the "study of all the genes in the human genome together, including their interactions with each other, the environment, and the influence of other psychosocial and cultural factors" (American Nurses Association, 2009; National Human Genome Research Institute, 2022).
- Precision health refers to the impact that a person's unique genetic, genomic, and omic composition have on well-being and health within the context of that person's lifestyle and social, economic, cultural and environmental context (Fu et al., 2020).
- Precision healthcare refers to clinical care delivered on the basis of understanding the contribution of an individual's unique disease risks, including genomic information and social context, and treatment responses for the improvement of health outcomes (National Human Genome Research Institute, 2024).

Thus, **genomics nursing is nursing within the context of genomic information and in the provision of precision healthcare**. Genomic nursing involves assessing, intervening, evaluating, protecting, promoting, advocating, educating, and performing research to ensure the health, safety, and well-being of others and society. As such, genomics nursing is concerned with the health and interconnectedness of individuals, families, communities, and populations as it relates to hereditary and non-hereditary genomic conditions.

Genomic conditions are heritable and non-heritable alterations of human health (including anomalies, behaviors, diseases, issues, or predispositions) that arise in whole or in part due to genetic, genomic, or other omic processes. Thus, genomics nurses are concerned with how single-gene disorders, chromosomal abnormalities, copy number variants, gene–gene interactions,

gene–environment interactions (including interactions with social determinants of health [SDOH]), epigenomics, gene expression, and molecular modifications may alter gene expression or otherwise affect human health.

The term "genomics nursing" was chosen with intentionality to address the evolving scientific understanding of the importance of considering the whole genome and its internal and external interactions in the context of precision health. As stated in the previous edition, genetics nursing traditionally involved the care of people with single-gene disorders such as cystic fibrosis or Huntington's disease, and chromosomal disorders such as Down syndrome. However, even single-gene disorders are modified by other genes and the environment. Indeed, the evidence base for genomics continues to rapidly expand, and precision healthcare is advancing both in terms of availability and scope. Thus, the term genomics best reflects the complexity and breadth of the phenomena of concern to genomics nurses. The deliberate selection and use of the term genomics reflects the evolution of the science to incorporate complex network and system approaches and signaling dynamics (Fu et al., 2020; E. Kurnat-Thoma et al., 2021); however, ISONG and ANA acknowledge that

- Nurses, physicians, and counselors may continue to use "genetic" or "genetics" in their professional titles;
- Certifying boards in this field award credentials that use the term genetic or genetics; and
- Many national and international organizations, representing the majority of professionals in this field, still use "genetic" or "genetics" in their names as a reflection of either their individual focus on single-gene and chromosomal disorders or the professional history of the organization and the specialty discipline of clinical (Mendelian) genetics.

While there has been extensive debate around the terminology for recipients of nursing services over the last several decades, inconsistency in the use of such terminology continues. In particular, recent trends toward the use of the term "healthcare consumer" have been considered by this working group. In consultation with the Board of Directors at ISONG, the terms "person" or "patients" were preferred for this revision

cycle due to the economic associations of the term "consumer," which may disempower and disenfranchise individuals without the economic means, ability, or access to freely participate in healthcare markets. Furthermore, the term "consumer" may be inappropriate in global healthcare contexts beyond third-party payer systems. This workgroup and ISONG reaffirm the vision of healthcare as a universal human right, in accordance with the Provision 9 of the *Code of Ethics for Nurses* (ANA, 2025). As precision healthcare models advance, genomics nurses hold an ethical duty to work toward health equity in careful consideration of the differential experiences and opportunities that individuals face based on socioeconomic status and healthcare access within and outside of the United States. Thus, this work group has chosen to focus on the recipients of services as persons rather than as consumers. Recipients of clinical genomics nursing services will be referred to throughout this document as persons or patients. These terms may encompass, for the purposes of this document, individuals, families, communities, and whole populations and includes both those at risk for and with manifested genomic conditions. In particular, when patients are unable to participate in care activities or decision-making (e.g., due to developmental stage such as with infants and children or vulnerability such as altered cognitive capacity), the word "patient" may refer to a legally-authorized representative, patient surrogate decision-maker, guardian, or parent.

Interaction of Genetics and Genomics

The Human Genome Project laid the foundation for enormous advances in the fields of genetics and genomics by completing the first reference sequence of the human genome in 2003. In response, genomics nursing practice has evolved significantly since the publication of ISONG's *Statement on the Scope and Standards of Genetics Clinical Nursing Practice* in 1998. Research efforts are improving understanding of the functions and interactions of all genes in the human genome as well as interactions with environmental factors. This genetic revolution has resulted in a paradigm shift from genetics to genomics, which is much broader and affects all areas of nursing practice. The ability to understand the role of genetics in human health and detection of diseases is a tremendous step towards

better prevention, risk reduction, treatment, and the realization of cures for common diseases and health problems (Green et al., 2020).

Scientific discoveries such as gene editing are enhancing clinical capabilities in diagnosing and delivering advanced treatments for rare, single-gene disorders (Frith, 2020). Likewise, advancements further the ability to predict susceptibility to and prescribe preventive therapies for genetically influenced chronic conditions, such as cancer and obesity (Behravan et al., 2020; Cuevas-Sierra et al., 2019; Rohde et al., 2019). Genomic investigation of infectious diseases, once thought impracticable, has increased the speed of diagnosis, the effectiveness of existing therapies, and the development of new therapies. Particularly during the COVID-19 pandemic, increased reliance on genomic knowledge and technology was brought to the collective forefront of our national awareness. Genomic science and healthcare were used to (1) sequence viral strain epidemiology patterns and monitor severe public health impacts of variants on global populations and individual disease risks; (2) rapidly create mRNA vaccines and novel therapeutics to save life and reduce suffering at a global scale; and (3) develop accurate diagnostic tests to guide clinical care and public health protocols. This expanding knowledge will continue to affect how genomics services are defined and delivered. Indeed, such services extend into an increasing variety of settings. Clinical nursing genomics services may include, among other things,

- Providing and managing comprehensive care, including state-of-the-art, evidence-based risk assessment, genomic screening, diagnosis, counseling, and therapeutic modalities;
- Protection and advocacy of vulnerable and underserved patient populations (e.g., minoritized racial and ethnic groups, sexual and gender minority populations, the very young and very old, people with disabilities, those with low socioeconomic status or educational attainment) to ensure equitable healthcare access and patient satisfaction with genomic services that is culturally responsive and appropriate per the patient's needs and wishes;
- Administering medications and treatments that are tailored to a patient's specific genetic information;

- Facilitating interdisciplinary referrals for the evaluation and treatment of genomic health conditions;
- Interpreting common non-prescribed (direct-to-consumer) genetic tests provided by commercial, non-healthcare entities and supporting patient, family, community, and population education;
- Evaluating, coordinating, and improving healthcare access to genomic specialty services;
- Educating, developing, and maintaining a nursing workforce with appropriate genomics competencies for level of practice and practice setting;
- Developing and optimizing routine nursing and clinical workflows to ensure integration of genomics information with electronic health records (EHR) and clinical decision support (CDS) systems;
- Educating individuals, families, and public and professional populations about genomics; and
- Assessing, evaluating, and developing recommendations for ethical, legal, and social implications (ELSI) of new and existing genomics services and health information technologies.

DESCRIPTION OF GENOMICS NURSING

The genomics nurse focuses on providing nursing care, education, administration, research, advocacy, and policymaking based on an understanding of the underlying genomics of individuals, families, communities, or populations. Genomics nursing practice encompasses a clear focus on the entirety of the human genome, including interactions between genes and between genes and the environment, and its implications on health and nursing care.

Genomics nursing has and will continue to contribute leadership to the development of genomics and precision healthcare models, for example, through

- Development of academic frameworks to guide curricula;
- Nursing workforce diversification, education, and training;

- Formation of and leadership in academic–community and academic–practice partnerships focused on genomics research, practice, education, and outreach;
- Competency development within the field of generalist and specialty nursing;
- Extensive additions to the evidence-base through the conduct of genomics research; and
- Expanding genomics use in multiple clinical practice areas, including pharmacogenetics, newborn screening, infectious disease monitoring, oncology, and many others.

Comprehensive genomics nursing practice is a dynamic process involving extensive interprofessional collaboration. Genomics nursing may occur in any setting, including but not limited to clinical patient care environments, academia, commercial industry, research laboratories and collaboratives, administrative settings, non-profit organizations, community and/or grassroots settings, and policymaking, legislative, and diplomatic forums.

Clinically practicing genomics nurses identify and manage physiological and psychological responses to genomic conditions and provide consultation and education to individuals, families, communities, populations, and healthcare teams. Clinical genomics nursing involves personal and trusting relationships between recipients of care and the nurse. Recipients of genomics nursing care may be individuals, families, communities, or populations including persons at any stage of life, from preconception to postmortem. Individual patients of genomics nurses include people who are symptomatic for a genomic condition, who are at risk for developing a genomic condition, or who may have a child who is at risk. Genomic nurses at varying levels of clinical practice may provide nursing care for patients, for example, by

- Prescribing and administering pharmacogenetic-based therapies and medications;
- Conducting family history assessments to detect the presence of high-risk hereditary and multifactorial genomic conditions;

- Making referrals to appropriate colleagues for follow-up testing, treatment, or care coordination for genomic conditions;
- Ordering and interpreting chromosomal, genetic, and genomic laboratory tests;
- Developing healthcare plans for individuals; and
- Providing counseling and education to individuals, families, communities, and populations.

In addition, genomics nurses may consult with the healthcare team to provide general information to parties interested in genomics or precision healthcare including the public, policymakers, and other stakeholders in public health. Genomics nurses in research generate and disseminate new knowledge about genomics and its implications for human health and nursing. Genomics nurses in academic or educational roles use current research and standards to support student learning objectives and genomics competencies within the specialty and among generalist nurses. In the healthcare policy area, genomics nurses use the evidence base to establish and guide the future direction of genomics nursing and healthcare policy (E. Kurnat-Thoma et al., 2021).

Of note, provision of basic nursing care in genetics and genomics is recognized as a fundamental registered nurse competency as endorsed by 49 key stakeholder organizations (American Nurses Association, 2009). ISONG agrees with the Consensus Panel that all registered nurses should possess a basic level of understanding of genetics and genomics (American Nurses Association, 2009). On the international stage and among professional organizations within and outside of nursing, wide support exists for enhancing the role of nurses in genomic healthcare (Global Genomics Nursing Alliance [G2NA], 2023). In recent years in the United States, genomics has been re-envisioned as part of the scientific basis for nursing rather than considered as a separate essential attribute; this shift in prioritization is due in part to the lack of opportunity for generalist and non-genomic specialty nurses for training (Newcomb et al., 2019). For example, the American Association of Colleges of Nursing (2021) limits the essential competencies of generalist professional nursing related to genetics to a single statement beyond basic scientific knowledge: to "apply individualized information, such as genetic/genomic, pharmacogenetic,

and environmental exposure information in the delivery of personalized health care" (p. 30). The specialist genomics nurse possesses competencies and standards of practice that extend well beyond the application of genomics information in the delivery of care in clinical practice settings. Thus, the genomics nurse has a role in educating and supporting nurses in general practice and the practice of other specialties, as well as other members of the healthcare team, toward enhanced care of persons, families, communities, and populations with or at risk for genomic conditions.

ESSENTIAL ATTRIBUTES OF GENOMICS NURSING

The essential features of genomics nursing practice are

- Attention to the full range of human experiences and responses pertaining to the continuum of health and illness of patients related to genomic conditions;
- Application of genomics knowledge (e.g., evidence base) to the processes of nursing practice, education, research, and policy related to
  - Health education, promotion, maintenance, and restoration
  - Optimization of health and abilities
  - Prevention of illness and injury
  - Alleviation of suffering
  - Informed patient decision-making
  - Complex healthcare systems
  - And when appropriate, a peaceful and dignified death;
- Integration of objective data with knowledge gained from an understanding of the patient's subjective experience with, or risk of, a genomic condition and associated disability or morbidity;
- Maintenance of principles and practices that promote genomic well-being and healing in consideration of the ethical, legal, and social issues (ELSI) associated with genomic conditions or susceptibility to genomic conditions;
- The promotion of social justice for people affected by or susceptible to genomic conditions;

- Establishment of caring relationships with persons and their legally authorized representatives, families, communities, or populations served, including patients, students or trainees, research participants, and policymakers, that facilitate nursing care as appropriate to the situation and setting; and
- Demonstration of leadership through consultation to other members of the healthcare team or stakeholders to support a wide variety of persons in policy, governance, and complex health systems.

Nursing is built on a foundation of knowledge that comprises the dual components of science and art. As a specialty of nursing, genomics nursing is a scientific discipline as well as a professional specialty. At all levels and in all settings, genomics nursing practice is evidence-based, relying on discoveries by nurses and others to continually refine and improve clinical, research, and educational practice outcomes, processes, and value. A number of theories and conceptual frameworks are used for assessing, diagnosing, planning, implementing, and evaluating care that is responsive to the essential attributes of genomics and precision health practice areas (Corwin et al., 2019; Kurnat-Thoma et al., 2021; Kurnat-Thoma et al., 2022; Reed, 2020). These theories and frameworks—derived from nursing, medicine, social sciences, biology, ethics, and other related fields—provide a foundation for understanding, implementing, and evaluating the practice of genomic nurses.

Ethical and moral dilemmas are common in the context of genomics and precision healthcare, including around questions of fairness to all persons (justice). Thus, sensitivity to diversity and health equity in all settings is an essential component of genomics nursing. *The ICN Code of Ethics for Nurses* (International Council of Nurses, 2021) is the framework for ethical nursing practice worldwide. In the United States, *The Code of Ethics for Nurses* (ANA, 2025) provides a framework to guide ethical nursing practice in the United States. In the context of genomic healthcare provision, ANA (2025) requires that genomic nurses have a responsibility to sustain their knowledge and competence in advanced technologies to translate ethical implications of import for clinical

practice, nursing education, research and public policy for specialty areas such as genetic testing, national and international guidelines protecting the rights of the human person, and emergent areas of governance and public health. Genomics nurses adhere to the ethical principles that govern all nursing, with special attention and focus to those aspects of genomics healthcare that require advanced ethical consideration.

Genomics nurses in all settings continually update practice and knowledge in line with evolving standards of care in precision health and genomics, as appropriate to their level of practice, training, professional role, and setting. Genomics nurses in all settings work toward the creation and updating of evidence-based policies and practices to support optimal health and learning outcomes of patients, families, communities, populations, and students or trainees. At the local, state, federal, and international levels, genomics nurses advocate for evidence-based healthcare policies that support the health of persons in need of genomic nursing services. In addition, genomics nurses advocate for enhanced development of the nursing and genomics nursing workforce.

PRACTICE SETTINGS FOR GENOMICS NURSING

Genomics nurses practice in healthcare settings that include but are not limited to: hospitals and their affiliated clinics, academic medical centers, and universities; institutions of higher education and training programs; health and science policy settings; research funding organizations; regional genomic centers; ambulatory and primary healthcare facilities; industrial, community, and school health settings; local, state, and federal government agencies, including those that provide services to underserved and vulnerable populations such as Federally Qualified Health Centers; private industry, including clinical and biotechnology laboratories and pharmaceutical companies; managed healthcare organizations; non-profit foundations and organizations; and healthcare recipient and provider insurance organizations. As advances in genomic technology continue to expand into a wider variety of clinical, academic, and research settings, so too will genomics

nursing practice. Genomics nursing practice has applications across a wide array of settings:

- Genomics nurses provide information about the relevant inheritance patterns, genomic testing results, and familial implications of test results in acute care settings and specialty clinics that focus on chromosomal and single-gene disorders.
- Genomics nurses contribute expertise and skills to public and community health settings.
- Genomics nurses in oncology assess risks; provide genomic counseling; guide interventions for somatic and hereditary cancers; facilitate screening and diagnostic genomic testing; and educate patients, families, and members of the oncology care team on hereditary cancer syndromes, the impact of the hereditary cancer diagnosis on current care, overall patient and family function, future cancer risk, and familial implications.
- Advanced practice genomics nurses evaluate, diagnose, counsel, and manage patients with or at risk for developing a genomic condition within specific patient populations and in collaboration with interprofessional teams of providers.
- Genomics nurses in the non-profit sector provide information, research, patient counseling, leadership, and expertise to foundations and other non-profit organizations dedicated to health or populations with specific genomic conditions.
- Genomics nurse managers, leaders, and nursing staff in Magnet-certified acute care organizations, or who are on Magnet journeys, facilitate integration of genomics services, nursing leadership, and healthcare outcomes into Magnet accreditation processes and requirements.
- Genomics nurses across a wide range of clinical practice environments ensure adoption of the most recent high quality clinical practice guidelines by participating in evidence-based practice and quality improvement projects and translating the strongest genomics research evidence into routine practice.
- Genomics nurses at all levels and in roles ranging from direct patient care to research and education to administration serve on

Institutional Review Boards (IRBs) to facilitate regulatory compliance, ensure patient safety and confidentiality, and protect human subjects across the lifespan who choose to participate in genomics research.

- Genomics nurses of all levels of academic preparation contribute their professional expertise, time, and talents to serve on local, state, federal, and international leadership and advisory groups (e.g., boards, committees) across a variety of multi-sector organizations to advocate on behalf of patients, populations, the profession of nursing, and the specialty of genomics nursing.
- Leaders in genomic nursing develop, initiate, and sustain a variety of regional, national, and international multi-sector collaborations to continuously expand the reach and impact of genomics into healthcare (Kurnat-Thoma et al., 2020).
- Genomics nurse administrators empower their organizations to cultivate and retain highly skilled workforces to develop and participate in new scientific innovations, evidence-based practice, quality improvement, performance improvement, and nursing research activities involving genomics services.
- Genomics nurse researchers and scientists perform research and policy analyses to expand genomics knowledge, facilitate creation of updated clinical practice guidelines, and achieve dissemination and implementation of genomics services into everyday clinical practice and community public health environments.
- Genomics nurses lead the conversation on ethical issues in genomics within the frameworks provided by national and international codes of ethics and advocate for social, health, and nursing policies at the local, state, national, and international level that adhere to the highest standards of ethical practice and quality of care.
- Genomics nurse educators design and effectively deliver genomics nursing content based on established competencies, develop learners' critical thinking and abilities to provide genomics-informed nursing care, utilize assessment and evaluation strategies vetted for genomics education, and advance education for genomics nursing by acting as change agents and leaders and through academic practice partnerships.