Genetics and Genomics in Nursing: A curriculum evaluation toolkit

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Introduction

The field of genetics and genomics is revolutionizing how human health and disease are conceptualized, diagnosed, and managed. Nurses need to be prepared at both the pre-licensure and graduate level and must be confident and competent in their ability interpret and use genetic and genomic data. Education in genetics and genomics must be integrated into nursing curricula at all levels of academic preparation to ensure that all nurses are able to apply genomic concepts to routine patient care.

Currently, many nursing programs do not specifically incorporate genetics and genomics into their nursing curricula or do so inconsistently. There are established competencies for the incorporation of genetics and genomics into nursing, including: *Essential Genetic and Genomic Competencies for Nurses* (Jenkins, J., & Calzone, K. A., 2007) and *Essential Competencies of Genetics and Genomics for Nurses with Graduate Degrees* (Greco, K. E., Tinley, S. & Seibert, D 2012). These genetic and genomic nursing competencies are complementary to existing nursing competencies, such as the American Association of Colleges of Nursing Essentials and can be used as a basis for curriculum mapping. It is well-recognized that nursing students who are ready to practice upon graduation will require knowledge of genetics and genomics including: family health history collection, risk assessment and referrals, personalized health care and understanding of tailored therapies, knowledge of responses to treatment and identifying those at risk, assisting patients in understanding results and making informed decisions regarding treatments, explanation of available treatment options to patients, understanding of the role of direct-to-consumer genetic test marketing, knowledge of ethical, social, and legal issues surrounding genetics, and environmental factors, social determinants of health and genetics. The wide applicability of genetics in all areas of healthcare will dictate for genetics and genomics to be incorporated into a wide range of courses.

To facilitate implementation of genetics and genomics throughout the existing nursing curricula it is important to examine content both at the course level and program level. We have created a framework based on available genetics and genomics competencies to allow course instructors to evaluate the depth and breadth of genetics and genomics concepts within individual courses, and a method for programs to evaluate the overall inclusion of genetics and genomics in nursing education at the level individual programs.

Jenkins, J., & Calzone, K. A. (2007). Establishing the essential nursing competencies for genetics and genomics. *Journal of Nursing Scholarship, 39*(1), 10-16.

Greco, K. E., Tinley, S. & Seibert, D. (2012) Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees. Silver Spring, MD: American Nurses Association and International Society of Nurses in Genetics.

Directions for use

This toolkit is designed to allow evaluation of genetics and genomics content in current nursing curriculum at both the course and program level.

This evaluation is in two parts:

Section 1: Course Evaluation Self-Assessment

We have provided a tool for course instructors to review their courses for genetics and genomics content based on competencies established by Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators, 2nd edition, 2008 for prelicensure nursing and Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees (2011) for advanced practice nursing. Course instructors should evaluate their course using the appropriate spreadsheet. The instructor will evaluate each competency and rank the incorporation of genetics and genomics concepts on this scale:

- Absent – It is absent in my course but should be there;
- Satisfactory – It is present in my course but could be better;
- Exceptional – It is present in my course and to an appropriate degree;
- N/A – Not applicable

An important note about the inclusion of genetic and genomic material and related genetics competencies: Not all competencies are expected to be included in any given course. These competencies represent areas of knowledge that are relevant to genetics and genomics in nursing throughout the entire curriculum and, thus, nursing practice.

After assessing incorporation of each competency, we also ask instructors to reflect on existing implementation of genetics and genomics content in their course, and opportunities for growth and expansion of genetics and genomics content. To aid in planning for content adjustments, we inquire about resources and skills needed to incorporate genetics and genomics content in their course.

Section 2: Genetics and Genomics Curriculum Review Program Evaluation

To assist nursing programs in evaluating their curriculum we also have generated a tool to aggregate the individual course self-assessments. Once each course has been evaluated by the course instructor, programs are encouraged to identify gaps in the overall curriculum by noting the absence or presence of a competency in all the courses. We suggest the following:

Please score incorporation of each competency based on responses from the self-evaluation of genetics and genomics for each course. Use this form to assess gaps in your program's curriculum.

Use the following rubric:

- 1 = Absent - It is absent in my course but should be there
- 2 = Included - It is present in my course but could be better; Exceptional - It is present in my course and to an appropriate degree;
- Leave blank = Not applicable. Do not factor these into your final scores

Once all courses have been entered programs will be able to identify areas of completeness and incompleteness in their curriculum.

Definitions and Examples

As genetics and genomics is further integrated into the standard of care, nurses will find increased exposure to terminology specific to the field. The terms below are used throughout the toolkit, and are defined to aid in the use of the curriculum evaluation. For additional definitions in genetics please visit the Talking Glossary of Genetic Terms (genome.gov).

Mendelian disorders – Disorders that are result of variants in a single gene and are heritable.

Multifactoral disorders – Disorders that are a result of a combination of multiple genes, or genes and environment.

Genetic and genomic information – Genetic and genomics information includes known variants, prior genetic diagnostic data, family history information.

Genetic and genomic technology – Includes techniques for diagnosis (sequencing and cytogenetic techniques), and current/potential therapies.

Inherited predisposition – Carrying the increased likelihood of acquiring a specific trait/phenotype because of an inherited genetic variant. Having a variant does not always mean pathogenesis.

Course Evaluation Self-Assessment

Part 1: Prelicensure Competency Evaluation

Note: An excel version of this spreadsheet is available to make collection and recording of this information easy (see attachments on our Genetics and Genomics Webpage).

Please indicate level to which each competency is incorporated in your course below.

Use the scale:
Absent – It is absent in my course but should be there;
Satisfactory – It is present in my course but could be better;
Exceptional – It is present in my course and to an appropriate degree;
N/A – Not applicable

Domain		Competency	Absent	Satisfactory	Exceptional	N/A
Professional Responsibilities	1	Recognize when one's own attitudes and values related to genetic and genomic science may affect care provided to clients.				
	2	Advocate for clients' access to desired genetic/genomic services and/or resources including support groups				
	3	Examine competency of practice on a regular basis, identifying areas of strength, as well as areas in which professional development related to genetics and genomics would be beneficial.				
	4	Incorporate genetic and genomic technologies and information into registered nurse practice				
	5	Demonstrate in practice the importance of tailoring genetic and genomic information and services to clients based on their culture, religion, knowledge level, literacy, and preferred language				
	6	Advocate for the rights of all clients for autonomous, informed genetic- and genomic-related decision-making and voluntary action.				
Nursing Assessment: Applying/Integrating	7	Demonstrates an understanding of the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness.				

Genetic and Genomic Knowledge	8	Demonstrates ability to elicit a minimum of three generation family health history information				
	9	Constructs a pedigree from collected family history information using standardized symbols and terminology.				
	10	Collects personal, health, and developmental histories that consider genetic, environmental, and genomic influences and risks.				
	11	Conducts comprehensive health and physical assessments which incorporate knowledge about genetic, environmental, and genomic influences and risk factors.				
	12	Critically analyzes the history and physical assessment findings for genetic, environmental, and genomic influences and risk factors.				
	13	Assesses clients' knowledge, perceptions, and responses to genetic and genomic information.				
	14	Develops a plan of care that incorporates genetic and genomic assessment information.				
Identification	15	Identifies clients who may benefit from specific genetic and genomic information and/or services based on assessment data.				
	16	Identifies credible, accurate, appropriate, and current genetic and genomic information, resources, services, and/or technologies specific to given clients.				
	17	Identifies ethical, ethnic/ancestral, cultural, religious, legal, fiscal, and societal issues related to genetic and genomic information and technologies.				
	18	Defines issues that undermine the rights of all clients for autonomous, informed genetic- and genomic-related decision-making and voluntary action.				
Referral Activities	19	Facilitates referrals for specialized genetic and genomic services for clients as needed.				
Provision of Education, Care, and Support	20	Provides clients with interpretation of selective genetic and genomic information or services				
	21	Provides clients with credible, accurate, appropriate, and current genetic and genomic information, resources, services, and/or technologies that facilitate decision-making				
	22	Uses health promotion/disease prevention practices that: • Consider genetic and genomic influences on personal and environmental risk factors.				

		• Incorporate knowledge of genetic and/or genomic risk factors (e.g., a client with a genetic predisposition for high cholesterol who can benefit from a change in lifestyle that will decrease the likelihood that the genetic risk will be expressed)				
	23	Uses genetic- and genomic-based interventions and information to improve clients' outcomes.				
	24	Collaborates with healthcare providers in providing genetic and genomic health care				
	25	Collaborates with insurance providers/payers to facilitate reimbursement for genetic and genomic healthcare services				
	26	Performs interventions/treatments appropriate to clients' genetic and genomic healthcare needs				
	27	Evaluates impact and effectiveness of genetic and genomic technology, information, interventions, and treatments on clients' outcome.				

Reflection on genetics and genomics in the course.

What opportunities are there for growth and expansion of genetics and genomics in the course?

Resources and skills needed to expand on genetics and genomics content in your course.

Part 2: Advanced Practice Competency Evaluation

NOTE: An excel version of this spreadsheet is available to make collection and recording of this information easy (see attachments on our Genetics and Genomics Webpage).

Please indicate level to which each competency is incorporated in your course below.

Use the scale:
Absent – It is absent in my course but should be there;
Satisfactory – It is present in my course but could be better;
Exceptional – It is present in my course and to an appropriate degree;
N/A – Not applicable

Domain		Competency	Absent	Satisfactory	Exceptional	N/A
Risk assessment and interpretation	1	Identify clients with inherited predispositions to diseases as appropriate to the nurse's practice setting.				
	2	Analyze a pedigree to identify potential inherited predisposition to disease.				
	3	Estimate risks for Mendelian and multifactorial disorders in affected families as appropriate.				
	4	Use family history and pedigree information to plan and conduct a targeted physical assessment.				
	5	Interpret the findings from the physical assessment, family history, laboratory findings, diagnostic tests, and/or radiology results that may indicate genetic/genomic disease, disease risk, or the need for a genetics/genomics referral.				
	6	Refer at-risk family members for assessment of inherited predisposition to disease.				
Genetic Education, Counseling, Testing, and Results Interpretation	7	Incorporate clients' attitudes, values, and beliefs rooted in varying ethnic, cultural, social, and religious backgrounds when communicating genetic/genomic information.				
	8	Provide genetic/genomic information that is appropriate to client's level of health literacy and numeracy.				
	9	Educate clients about possible risks, benefits, and limitations of genetic testing and/or therapy.				
	10	Provide anticipatory guidance to assist clients in the decision-making process related to genetics/genomics.				
	11	Obtain informed consent for genetic testing and/or therapy.				
	12	Assess the influence of genetic/genomic risk and disease on family communication and functioning.				

	13	Assess the clinical and psychosocial outcomes, including benefits, limitations, and risks of genetic/genomic information and/or therapies, for clients.				
	14	Support client coping and client use of genetic/genomic information in promoting health, reducing risk, managing symptoms, and/or preventing illness.				
	15	Provide genetic/genomic education and counseling appropriate to practice setting.				
	16	Select appropriate genetic/genomic tests and/or studies.				
	17	Communicate results of genetic/genomic screening and/or testing at a level that clients can understand.				
Clinical Management	18	Apply knowledge about the interaction of genetic/genomic and environmental factors to the care of clients.				
	19	Make appropriate referrals to genetic professionals or other health care resources.				
	20	Evaluate effectiveness of prevention, risk reduction, health promotion, and disease management interventions related to genetics/genomics.				
	21	Manage care of clients, incorporating genetic/genomic information and technology (e.g., risk-based genetic screening and testing, prescription of pharmacogenomic-based drugs, gene-targeted therapy, and use of genetic/genomic information in symptom management).				
	22	Collaborate with genetic specialists, health professionals, and those in relevant disciplines to develop a comprehensive plan to evaluate and manage clients with genetic/genomic disease or risk.				
Ethical, Legal, and Social Implications	23	Facilitate ethical decision-making related to genetics/genomics congruent with the client's values and beliefs.				
	24	Inform health care and research policy related to ELSI issues in genetics/genomics.				
	25	Implement effective strategies to resolve ELSI issues related to genetics/genomics.				
	26	Apply ethical principles when making decisions regarding management of genetic/genomic information identified through clinical or research technologies.				
Professional Role	27	Integrate best genetic/genomic evidence into practice that incorporates client values and clinical judgment.				
	28	Mentor other nurses in the application of genetics/genomics to nursing care within their practice setting.				
	29	Identify genetic/genomic learning needs of other health professionals and disciplines.				
	30	Conduct educational interventions to address the genetic/genomic learning needs of health professionals and clients.				
	31	Participate in the development of professional practice guidelines related to genetics/genomics.				
Leadership	32	Contribute a nursing perspective to genetic/genomic clinical and policy discussions.				

	33	Facilitate an organizational climate that is responsive to genetic/genomic discoveries.				
	34	Use care delivery strategies which incorporate genetic/genomics.				
	35	Influence health policy at the local, state, national, and international levels related to genetics/genomics.				
Research	36	Participate in the application and translation of genetic/genomic research in nursing practice and/or education.				
	37	Identify genetic/genomic health care methods and outcomes that can be influenced by nursing.				
	38	Collaborate with researchers in relevant disciplines in the conduct, dissemination, and/or translation of genomic inquiry and research.				

Reflection on genetics and genomics in the course.

What opportunities are there for growth and expansion of genetics and genomics in the course?

Resources and skills needed to expand on genetics and genomics content in your course.

Genetics and Genomics Curriculum Review Program Evaluation

Prelicensure degree programs

Note: An excel version of this spreadsheet is available to make collection and recording of this information easy (see attachments on our Genetics and Genomics Webpage).

Please score incorporation of each competency based on responses from the self-evaluation of genetics and genomics for each course. Use this form to assess gaps in your program's curriculum.

Use the following rubric:
1 = Absent - It is absent in my course but should be there
2 = Included - It is present in my course but could be better; Exceptional - It is present in my course and to an appropriate degree;
Leave blank = Not applicable. Do not factor these into your final scores

Domain		Competency	Course 1	Course 2	Course 3	Course 4	Add additional courses
Professional Responsibilities	1	Recognize when one's own attitudes and values related to genetic and genomic science may affect care provided to clients.					
	2	Advocate for clients' access to desired genetic/genomic services and/or resources including support groups					
	3	Examine competency of practice on a regular basis, identifying areas of strength, as well as areas in which professional development related to genetics and genomics would be beneficial.					
	4	Incorporate genetic and genomic technologies and information into registered nurse practice					
	5	Demonstrate in practice the importance of tailoring genetic and genomic information and services to clients based on their culture, religion, knowledge level, literacy, and preferred language					

	6	Advocate for the rights of all clients for autonomous, informed genetic- and genomic-related decision-making and voluntary action.					
Nursing Assessment: Applying/Integrating Genetic and Genomic Knowledge	7	Demonstrates an understanding of the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness.					
	8	Demonstrates ability to elicit a minimum of three generation family health history information					
	9	Constructs a pedigree from collected family history information using standardized symbols and terminology.					
	10	Collects personal, health, and developmental histories that consider genetic, environmental, and genomic influences and risks.					
	11	Conducts comprehensive health and physical assessments which incorporate knowledge about genetic, environmental, and genomic influences and risk factors.					
	12	Critically analyzes the history and physical assessment findings for genetic, environmental, and genomic influences and risk factors.					
	13	Assesses clients' knowledge, perceptions, and responses to genetic and genomic information.					
	14	Develops a plan of care that incorporates genetic and genomic assessment information.					
Identification	15	Identifies clients who may benefit from specific genetic and genomic information and/or services based on assessment data.					
	16	Identifies credible, accurate, appropriate, and current genetic and genomic information, resources, services, and/or technologies specific to given clients.					
	17	Identifies ethical, ethnic/ancestral, cultural, religious, legal, fiscal, and societal issues related to genetic and genomic information and technologies.					
	18	Defines issues that undermine the rights of all clients for autonomous, informed genetic- and genomic-related decision-making and voluntary action.					
Referral Activities	19	Facilitates referrals for specialized genetic and genomic services for clients as needed.					
Provision of Education, Care, and Support	20	Provides clients with interpretation of selective genetic and genomic information or services					
	21	Provides clients with credible, accurate, appropriate, and current genetic and genomic information, resources, services, and/or technologies that facilitate decision-making					

22	Uses health promotion/disease prevention practices that: • Consider genetic and genomic influences on personal and environmental risk factors.					
	• Incorporate knowledge of genetic and/or genomic risk factors (e.g., a client with a genetic predisposition for high cholesterol who can benefit from a change in lifestyle that will decrease the likelihood that the genetic risk will be expressed)					
23	Uses genetic- and genomic-based interventions and information to improve clients' outcomes.					
24	Collaborates with healthcare providers in providing genetic and genomic health care					
25	Collaborates with insurance providers/payers to facilitate reimbursement for genetic and genomic healthcare services					
26	Performs interventions/treatments appropriate to clients' genetic and genomic healthcare needs					
27	Evaluates impact and effectiveness of genetic and genomic technology, information, interventions, and treatments on clients' outcome.					

Advanced degree programs

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Please score incorporation of each competency based on responses from the self-evaluation of genetics and genomics for each course. Use this form to assess gaps in your program's curriculum.

Use the following rubric:
1 = Absent - It is absent in my course but should be there
2 = Included - It is present in my course but could be better; Exceptional - It is present in my course and to an appropriate degree;
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Domain		Competency	Course 1	Course 2	Course 3	Course 4	Add additional courses
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	2	Analyze a pedigree to identify potential inherited predisposition to disease.					
	3	Estimate risks for Mendelian and multifactorial disorders in affected families as appropriate.					
	4	Use family history and pedigree information to plan and conduct a targeted physical assessment.					
	5	Interpret the findings from the physical assessment, family history, laboratory findings, diagnostic tests, and/or radiology results that may indicate genetic/genomic disease, disease risk, or the need for a genetics/genomics referral.					
	6	Refer at-risk family members for assessment of inherited predisposition to disease.					
Genetic Education, Counseling, Testing, and Results Interpretation	7	Incorporate clients' attitudes, values, and beliefs rooted in varying ethnic, cultural, social, and religious backgrounds when communicating genetic/genomic information.					
	8	Provide genetic/genomic information that is appropriate to client's level of health literacy and numeracy.					
	9	Educate clients about possible risks, benefits, and limitations of genetic testing and/or therapy.					

	10	Provide anticipatory guidance to assist clients in the decision-making process related to genetics/genomics.					
	11	Obtain informed consent for genetic testing and/or therapy.					
	12	Assess the influence of genetic/genomic risk and disease on family communication and functioning.					
	13	Assess the clinical and psychosocial outcomes, including benefits, limitations, and risks of genetic/genomic information and/or therapies, for clients.					
	14	Support client coping and client use of genetic/genomic information in promoting health, reducing risk, managing symptoms, and/or preventing illness.					
	15	Provide genetic/genomic education and counseling appropriate to practice setting.					
	16	Select appropriate genetic/genomic tests and/or studies.					
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Clinical Management	18	Apply knowledge about the interaction of genetic/genomic and environmental factors to the care of clients.					
	19	Make appropriate referrals to genetic professionals or other health care resources.					
	20	Evaluate effectiveness of prevention, risk reduction, health promotion, and disease management interventions related to genetics/genomics.					
	21	Manage care of clients, incorporating genetic/genomic information and technology (e.g., risk-based genetic screening and testing, prescription of pharmacogenomic-based drugs, gene-targeted therapy, and use of genetic/genomic information in symptom management).					
	22	Collaborate with genetic specialists, health professionals, and those in relevant disciplines to develop a comprehensive plan to evaluate and manage clients with genetic/genomic disease or risk.					
Ethical, Legal, and Social Implications	23	Facilitate ethical decision-making related to genetics/genomics congruent with the client's values and beliefs.					
	24	Inform health care and research policy related to ELSI issues in genetics/genomics.					
	25	Implement effective strategies to resolve ELSI issues related to genetics/genomics.					
	26	Apply ethical principles when making decisions regarding management of genetic/genomic information identified through clinical or research technologies.					
Professional Role	27	Integrate best genetic/genomic evidence into practice that incorporates client values and clinical judgment.					
	28	Mentor other nurses in the application of genetics/genomics to nursing care within their practice setting.					

	29	Identify genetic/genomic learning needs of other health professionals and disciplines.					
	30	Conduct educational interventions to address the genetic/genomic learning needs of health professionals and clients.					
	31	Participate in the development of professional practice guidelines related to genetics/genomics.					
Leadership	32	Contribute a nursing perspective to genetic/genomic clinical and policy discussions.					
	33	Facilitate an organizational climate that is responsive to genetic/genomic discoveries.					
	34	Use care delivery strategies which incorporate genetic/genomics.					
	35	Influence health policy at the local, state, national, and international levels related to genetics/genomics.					
Research	36	Participate in the application and translation of genetic/genomic research in nursing practice and/or education.					
	37	Identify genetic/genomic health care methods and outcomes that can be influenced by nursing.					
	38	Collaborate with researchers in relevant disciplines in the conduct, dissemination, and/or translation of genomic inquiry and research.					

Appendices

i. Conversations in Genetics Webinar Series

Conversations in genetics is an educational series directed at nurses of all levels focusing on topics tailored to the specific needs of nurses and highlighting the roles of nurses in genetics, as well as the utility of genetics in existing nursing roles. This program provides real world examples of genetics in nursing, collaboration with interprofessional teams, and resources and guidelines to support the nursing workforce. Our goal in creating this series was to provide practical education to empower nurses to use genetics and genomics in practice, as well as to provide education to nursing school faculty responsible for integrating this content into nursing school curricula.

The full series is available to the public on the Johns Hopkins University School of Nursing YouTube page on the #GeneticsGenomics channel:
https://www.youtube.com/hashtag/geneticsgenomics

What We Say Matters

Moderator: Joann Bodurtha, MD, MPH
Panelists: Carolyn Applegate, MGC, CGC
Colleen Giofreda, BS
Lindsay Kwong, DNP, FNP-C
Carla McGruder, MS, CGC
Nicole Thompson, MS, CGC
Crystal Tichnell, MGC, RN

Description: The panel addresses questions about discussing family history and genetic issues with patients and the public. They discuss best practices for how nurses and other address sensitive issues, strong emotions, and cultural differences related to genetics. They also discuss how to incorporate genetic risk and family resilience in health assessments.

Link: https://www.youtube.com/watch?v=GFhiOEN7aoY

Resources:

1. Reed EK et al. Three things every nurse practitioner can do to integrate genetics into practice. J Am Assoc Nurse Pract. 2019 Jan;31(1):6-7. doi: 10.1097/JXX.0000000000000182. PMID: 30624360.
2. Hull LE et al. Revisiting the Roles of Primary Care Clinicians in Genetic Medicine. JAMA. 2020 Oct 27;324(16):1607-1608. doi: 10.1001/jama.2020.18745. PMID: 32970138.
3. Knowing Is Not Enough - Family Health portrait http://www.hhs.gov/familyhistory/index.html
4. Understanding a pedigree - https://vimeo.com/399304375

Engaging Nurses in Genetics, the Future is Here

Speakers: Dr. Alison Metcalfe
Dr. Christine Patch

Description: The field of genetics and genomics is revolutionizing how human health and disease are conceptualized, diagnosed and managed. In this webinar Drs. Patch and Metcalfe will discuss practical methods and guidance to empower nurses to use genetics and genomics in real-world care.
Link: https://www.youtube.com/watch?v=VaVKfHvbfLs
Resources:

1. Metcalfe A. Sharing Genetic Risk Information: Implications for Family Nurses Across the Life Span. J Fam Nurs. 2018 Feb;24(1):86-105. doi: 10.1177/1074840718755401. PMID: 29490585.
2. Tonkin E et al. A Roadmap for Global Acceleration of Genomics Integration Across Nursing. J Nurs Scholarsh. 2020 May;52(3):329-338. doi: 10.1111/jnu.12552. Epub 2020 Apr 17. PMID: 32301236; PMCID: PMC7202994.
3. Tonkin E et al. A Maturity Matrix for Nurse Leaders to Facilitate and Benchmark Progress in Genomic Healthcare Policy, Infrastructure, Education, and Delivery. J Nurs Scholarsh. 2020 Sep;52(5):583-592. doi: 10.1111/jnu.12586. Epub 2020 Jun 27. PMID: 32592453; PMCID: PMC7721977.
4. 100,000 Genomes Project - https://www.genomicsengland.co.uk/about-genomics-england/the-100000-genomes-project/

Genetics of the Newborn Screen

Moderator: Carolyn Applegate, MGC, CGC
Panelists: Johnna Watson, RN, BSN
Celide Koerner, RN, MSRD
Kristen Byrnes, CRNP
Gerald Raymond, MD

Description: Our panel answers questions about newborn screening in the state of Maryland. We discuss the purpose and process of newborn screening to identify inborn errors of metabolism and welcome a family to discuss their experience with receiving positive screening results. The discussion will center around the important roles nurses have in identification and management of families and infants from pre-test to results.

Link: https://www.youtube.com/watch?v=4R-zblv6n1A

Resources:

1. Baby's First Test https://www.babysfirsttest.org/newborn-screening/states/maryland
2. Association of Women's Health, Obstetric and Neonatal Nurses (AWHONN) Position Statement, Newborn Screening. Journal of Obstetric, Gynecologic, & Neonatal Nursing, 2016;45:135–136. https://doi.org/10.1016/j.jogn.2015.11.004

3. https://www.newbornscreening.info/
4. ACMG ACT Sheets and Algorithms https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets_and_Algorithms.aspx
5. DeLuca J. et al. Implications of newborn screening for nurses. Journal of Nursing Scholarship. 2013; 45(1):25-33. https://doi.org/10.1111/jnu.12005

Using Genetics in Clinical Cardiology

Moderator: Crystal Tichnell. MGC, RN
Speakers: Jodie Ingles, MPH, FHRS, FCSANZ
Chris Semsarian, AM, MBBS, PhD, MPH, FRACP, FRCPA, FAHMS, FAHA, FHRS, FCSANZ

Description: Our speakers discuss topics in genetics related to cardiology.

Link: https://www.youtube.com/watch?v=loj-ZQ5JYL0

Resources:

1. Sudden Arrythmia Death Syndromes Foundation https://www.sads.org/
2. Towbin JA, et al. 2019 HRS expert consensus statement on evaluation, risk stratification, and management of arrhythmogenic cardiomyopathy. Heart Rhythm. 2019 Nov;16(11):e301-e372. doi: 10.1016/j.hrthm.2019.05.007. Epub 2019 May 9. PMID: 31078652.
3. Ingles J et al. Genetic Testing in Inherited Heart Diseases. Heart Lung Circ. 2020 Apr;29(4):505-511. doi: 10.1016/j.hlc.2019.10.014. Epub 2019 Nov 29. PMID: 31813745.
4. Stiles MK et al. 2020 APHRS/HRS expert consensus statement on the investigation of decedents with sudden unexplained death and patients with sudden cardiac arrest, and of their families. Heart Rhythm. 2021 Jan;18(1):e1-e50. doi: 10.1016/j.hrthm.2020.10.010. Epub 2020 Oct 19. PMID: 33091602; PMCID: PMC8194370.

Demystifying Genetic Testing

Moderators: Jen Wilder, RN, DNP
Teresa Romeo Luperchio, PhD
Speaker: Weiyi Mu, ScM, CGC

Description: Genetic counselor Weiyi Mu discusses considerations when ordering genetic testing, reading and interpreting genetic laboratory reports.

Link: https://www.youtube.com/watch?v=i_bhT4ecwLk

Resources:

1. Find a genetic counselor: https://findageneticcounselor.nsgc.org/
2. Biesecker LG, Green RC. Diagnostic clinical genome and exome sequencing. N Engl J Med. 2014 Sep 18;371(12):1170. doi: 10.1056/NEJMc1408914. PMID: 25229935.
3. Clinvar https://www.ncbi.nlm.nih.gov/clinvar/

Genetic testing lookup tools

4. Concert Genetics https://www.concertgenetics.com/
5. NIH Genetic Test Registry https://www.ncbi.nlm.nih.gov/gtr/

Oncology

Moderator: Jen Wilder, RN, DNP
Speakers: Grace-Ann Fasaye, ScM, CGC
David Euhus, MD

Description: Genetics and genomics knowledge is crucial to understanding cancer development, diagnosis, prognosis, and treatment. This webinar presents basic concepts of genetics in cancer development, treatment, and counseling as an interprofessional team of nurse, genetic counselor, and oncologist.

Link: https://www.youtube.com/channel/UCxOWRTH3CfPYUORRvb5yLgw

Resources:

1. NCCN Guidelines for Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic https://www.nccn.org/guidelines/guidelines-detail?category=2&id=1503
2. Risk calculator https://ask2me.org/calculator.php (Multiple risk calculators exist, each has different strengths based on patient and family history)
3. American Society for Clinicians in Oncology (Advanced practice level) https://www.asco.org/practice-policy/cancer-care-initiatives/genetics-toolkit
4. City of Hope Intensive Course in Genomic Cancer Risk Assessment (Advanced practice level) https://www.cityofhope.org/education/health-professional-education/cancer-genomics-education-program/intensive-course-in-cancer-risk-assessment-overview
5. Oncology Nursing Society (general nursing and oncology nursing) https://voice.ons.org/topic/genetics-genomics

Sharing information with relatives

6. CDC Example Letter for Patients to Share Genetic Testing for Hereditary Breast and Ovarian Cancer (HBOC): https://www.cdc.gov/genomics/implementation/toolkit/hboc_letter_family_members.htm
7. Kintalk(https://kintalk.org)

*Family follow-up testing **(Examples only, no specific laboratories are endorsed or recommended in this document)*

8. https://www.invitae.com/en/family/ **
9. Low-cost testing options for patients without insurance coverage - Color Laboratories https://www.color.com/learn/family-genetic-testing-program**

ii. Pharmacogenomics: Implications for Nurses

Johns Hopkins University School of Nursing Virtual Grand Rounds

Speaker: Nicole Mollenkopf

Description: In the Johns Hopkins School of Nursing Virtual Nursing Grand Rounds, Dr. Nicole Mollenkopf, discusses “Pharmacogenomics: Implications for Nurses”. In this webinar, Dr. Mollenkopf will address pharmacogenomics, discuss genetic variations that have the potential to influence safe and effective medication use, and review resources available on pharmacogenomics information.

Link: https://www.youtube.com/watch?v=oHo6xyTZtt4

Resources:

1. PharmGKB – https://www.pharmgkb.org/
2. Clinical Pharmacogenomics Implementation Consortium (CPIC) – https://cpicpgx.org/

iii. Genetics in Nursing Resources

Nursing Specific Resources

Resource	Description
Nursing specific	
International Society for Nurses in Genetics - https://www.isong.org/	Professional Society focused on genetics in nursing
Omics Nursing Science & Education Network - https://omicsnursingnetwork.net/	Resources for utilizing omics technologies in nursing research
Method for introducing a new competency: Genomics (MINC) Project - https://genomicsintegration.net/index.php	Tool kit for introducing genomics into practice
Global Genomics Nursing Alliance - http://www.g2na.org/	International nursing group to integrate genomics into nursing practice
Online Journal of Issues in Nursing - http://ojin.nursingworld.org/MainMenuCategories/ANAMarketplace/ANAPeriodicals/OJIN/JournalTopics/FirstGeneticsNowGenomics	First Genetics Now Genomics special issue (2008)
Online Journal of Issues in Nursing - http://ojin.nursingworld.org/MainMenuCategories/ANAMarketplace/ANAPeriodicals/OJIN/JournalTopics/GeneticRevolution	Genetic Revolution special issue (2000)
Oncology Nursing Society - https://www.ons.org/	Contains resources including: Genomics and Precision Learning Libarary (Quick reference site for ONS Genomics Advisory Board - includes professional learning tools for oncology nurses, videos, podcasts, clinical references, articles and links to outside Genetics and Genomics Resources), Genomics Foundation site (Genomics taxonomy)
Education and CEUs	
Jax education and learning - https://www.jax.org/education-and-learning	Various educational modules in genetics
Human and Mammalian Genetics and Genomics: The McKusick Short Course - https://www.jax.org/education-and-learning	This course is a modern survey of heredity, disease, genetics in experimental animals and humans, and molecular genetics in the diagnosis and treatment of inherited disorders. The program combines JHU research expertise in medical genetics with JAX's unique knowledge and experience in experimental genetics using the mouse system.
Information	

CDC Office of Genetics and Disease Prevention - https://www.cdc.gov/genomics/default.htm	Broad genetics and genomics resources for all audiences
NHS, National Genetics Education and Development Center – Telling Stories - http://www.tellingstories.nhs.uk/	Educational modules in the form of recorded stories to provide real examples of how genetics and genomics is encountered in real life. This resource is geared toward health professionals.
Surgeon General's Family Health History Initiative - http://www.hhs.gov/familyhistory/index.html	Online tool and information to aid the public in collecting their own family health history.
The University of Utah, Genetic Science Learning Center - http://learn.genetics.utah.edu/	Online educational materials covering a broad range of topics in genetics. Modules are available at no cost.
National Birth Defects Prevention Network - http://www.nbdpn.org/	Network to provide education, resources, data and surveillance of birth defects in the US
MedlinePlus Genetics (previously known as Genetics Home Reference) - https://medlineplus.gov/	Public health information on genetics and genomics
Genetic and Rare Diseases Information Center (GARD) - https://rarediseases.info.nih.gov/	GARD provides the public with access to current, reliable, and easy-to-understand information about rare or genetic diseases in English or Spanish.
Clinical trials - https://clinicaltrials.gov/	Information on publicly and privately funded clinical trials
Genetic Alliance - http://www.geneticalliance.org/	Advocacy in Genetics
Understanding Rare Chromosome and Gene Disorders - https://www.rarechromo.org/	UNIQUE: Networking resources for families with rare genetic disorders and informative fact sheets
Orphanet - https://www.orpha.net/consor/cgi-bin/index.php	Rare disease and orphan drug information
National Society of Genetic Counselors - https://www.nsgc.org/	Professional society for genetic counselors, also contains information on how to find a genetic counselor.
American Society of Human Genetics (ASHG) - www.ashg.org	Professional society for those focused on genetics.
Global Genetics and Genomics Community (G3C) - https://www.genomicscases.net/en	Online simulations and G3C resource list - Unfolding Case Studies of patient cases
Genetics and Genomics Competency Center (G2C2) - https://genomicseducation.net/	NIH portal of educational resources
Gene Reviews - https://www.ncbi.nlm.nih.gov/books/NBK1116/	Point of care resource for genetic disorders

NHGRI - https://www.genome.gov/For-Health-Professionals/Provider-Genomics-Education-Resources	Healthcare Provider Genomics Education Resources
Online Mendelian Inheritance in Man (OMIM) - https://www.omim.org/	Comprehensive compendium of genes and phenotypes
EuroGen Test - http://www.eurogentest.org/index.php?id=160	Information on genetic testing in Europe
Genetics tools	
ACMG - https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Practice-Guidelines.aspx	Clinical practice guidelines
Clinvar - https://www.ncbi.nlm.nih.gov/clinvar/	Clinvar is a public archive of reports about human phenotypes and their relationship to health outcomes
Clingen - https://clinicalgenome.org/	Public resource that defines clinical relevance of genes and variants
Varsome - https://varsome.com/	Tool to provide functional annotation of variants and connect researchers, and data aggregator for DNA variants.
Gnomad - https://gnomad.broadinstitute.org/	Provides population data of variants from aggregate whole exome and whole genome sequencing data
phenodb - https://phenodb.org/	myPhenoDB is a freely-accessible website that allow clinical researchers to store standardized phenotypic information, diagnosis, and pedigree data and then run analyses on VCF files from individuals, families or cohorts with suspected Mendelian disease.
Genescout - https://genescout.omim.org/	GeneScout is a tool to search genomic regions identified by chromosome microarray to show the genes and their associated phenotypes within the regions of interest.
Genematcher - https://genematcher.org/	GeneMatcher is a freely accessible web site designed to enable connections between clinicians and researchers from around the world who share an interest in the same gene or genes.
Variant matcher - https://variantmatcher.org/	Tool to connect patients and clinicians who have interest in the same variant/'unsolved' exome. Affiliated with the Baylor-Hopkins Center for Mendelian Genomics.

CONCERT genetics - https://www.concertgenetics.com/	Various tools and information regarding genetic testing. Genetic test search allows for parameter query of testing available.
UCSC genome browser - https://genome.ucsc.edu/	Tools, datasets, and visualization of genomes - contains various species and many data from different sources. Offers the ability to build your own custom genome 'session' to examine genomic features in regions of interest.
Human Gene Mutation Database - http://www.hgmd.cf.ac.uk/ac/index.php	Database of all published gene mutations
Decipher - https://decipher.sanger.ac.uk/	DECIPHER (DatabasE of genomiC varIation and Phenotype in Humans using Ensembl Resources) is an interactive web-based database which incorporates a suite of tools designed to aid the interpretation of genomic variants.
ELSI	
Human genome project archived site - https://web.ornl.gov/sci/techresources/Human_Genome/elsi/index.shtml	Human Genome project information on ethical issues - archived from 2003
Policy issues in Genetics - NHGRI - https://www.genome.gov/about-genomics/policy-issues	Information on policy issues regarding genetics and genomics (discrimination, genome editing, disparities, consent, privacy and other issues)
Pharmacogenomics	
PharmGKB - https://www.pharmgkb.org/	PharmGKB is an NIH-funded resource that provides information about how human genetic variation affects response to medications. PharmGKB collects, curates and disseminates knowledge about clinically actionable gene-drug associations (i.e., gene-drug pairs) and genotype-phenotype relationships. When using PharmGKB, you will see different types of information. These are detailed in the sections below.

Clinical Pharmacogenomics Implementation Consortium (CPIC) - https://cpicpgx.org/	CPIC is an international consortium of individual volunteers and a small dedicated staff that create, curate, and post freely available, peer-reviewed, evidence-based, updatable, and detailed gene/drug clinical practice guidelines (click here for all CPIC publications). CPIC guidelines follow standardized formats, include systematic grading of evidence and clinical recommendations, use standardized terminology, are peer-reviewed, and are published in a leading journal (in partnership with Clinical Pharmacology and Therapeutics) with simultaneous posting to cpicpgx.org, where they are regularly updated. CPIC started as a shared project between PharmGKB and the Pharmacogenomics Research Network (PGRN) in 2009. CPIC guidelines are indexed in PubMed as clinical guidelines, endorsed by ASHP and ASCPT, and referenced in ClinGen and PharmGKB.