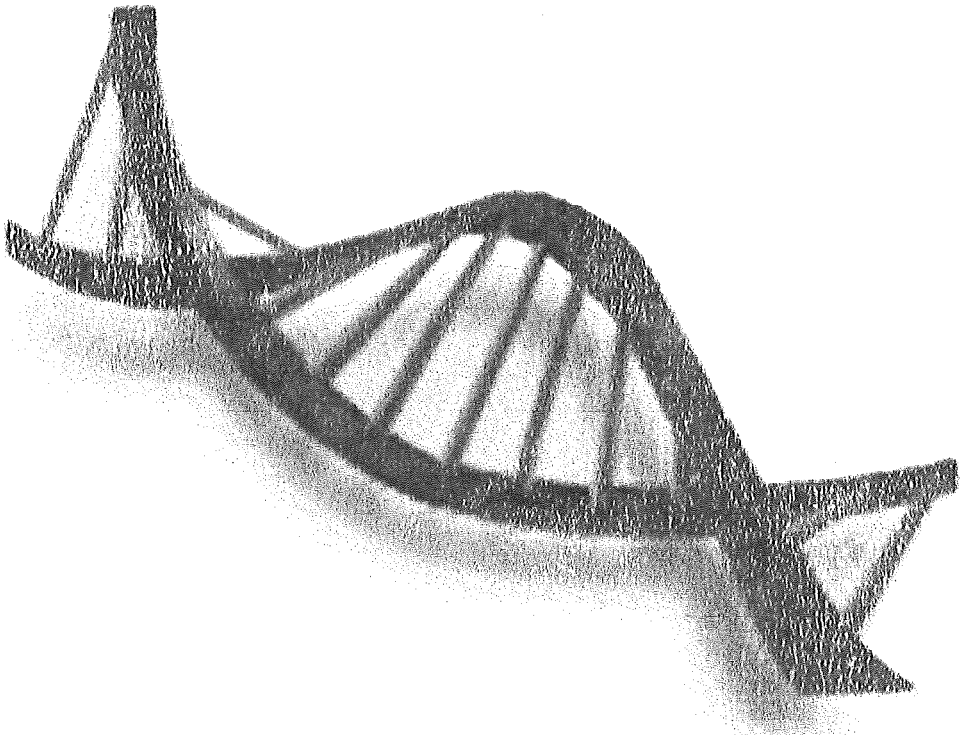
UNDERSTANDING GENETICS & GENOMICS: DEFINITIONS FOR NURSING PRACTICE



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GENETICS & GENOMICS IN THE NEWS

OPEN A NEWSPAPER, LISTEN TO THE NEWS, USE THE INTERNET, OR READ PROFESSIONAL NEWSLETTERS & YOU WILL FIND ARTICLES ABOUT GENETICS & GENOMICS. SEE SOME RECENT EXAMPLES BELOW!

New Genetic Findings Should Revolutionize Cancer Screening

Genes Dictate Folate Benefit in Schizophrenia

Heavy Smoking May Be Genetic

Genes Tied to Endometrial Cancer Outcomes

Marry Me? We're Genetically Different

Valve Disease May Have Genetic Roots

Groups Offer Guidance on Gene Testing in Kids

Genetics and high blood pressure appear to interact to increase deposits involved in the pathology of Alzheimer's

Personal Genome Project: Nature and Nurture, Warts and All

SPECIAL REPORT: PSYCHIATRY--5 Psych Disorders Have Common Genetics (The findings come from a genome-wide analysis of 33,332 cases and 27,888 controls)

Information to Collect

FAMILY HISTORY COLLECTION FORM

A family history generally should include:

the family structure,

medical history of each family member, and

environmental history.

FAMILY STRUCTURE

Identify as many family members as practical, including at least all first-degree relatives (parents, siblings, and children) and second-degree relatives (grandparents, aunts/uncles, and cousins), adding information about more distant relatives as necessary to see patterns in the family.

Indicate how individuals are related to each other within the family (for example, whether relatives are maternal or paternal).

Identify specific types of relationships, including monozygotic (identical) and dizygotic twins (fraternal), individuals adopted in and out of the family, and step- and half-siblings.

Include information about living and deceased relatives.

MEDICAL HISTORY AND DEMOGRAPHIC INFORMATION

The history for each family member should include:

Sex

Age

Any chronic or long-term conditions (or, in a targeted history, the condition[s] of interest)

Age at diagnosis

Relevant interventions or procedures

Cause of death and age at death

Ethnicity or country or countries of origin of the family

In situations where risk appears to be significantly increased or when an unusual diagnosis is reported, clinicians should validate diagnoses by reviewing records (e.g., medical records, pathology reports, death certificates).

LIFESTYLE, DIET, AND ENVIRONMENTAL RISK FACTORS

Depending on how much the clinician already knows about the patient and his or her history and concerns, the family history may include:

important environmental/lifestyle risk factors for disease in relatives, such as smoking, alcohol use, and diet,

the patient's occupation and the occupation of relatives with chronic conditions, and

protective environmental/lifestyle modifications, including lifestyle changes, treatments, and surgeries.

AMA
AMERICAN
MEDICAL
ASSOCIATION

Adult Family History Form

Date _____
Please complete as much of this form as possible and RETURN it before your next appointment. This information may be useful to your doctor prior to your appointment.

(Index)Patient _____

Date of Birth_____ Sex_____ Ethnicity_____

Address_____

Phone number_____ Work number_____

Occupation_____ Highest Grade Completed _____

Name of Spouse _____

Date of Birth_____ Ethnicity_____

Referring Doctor_____

Address_____

Family Doctor_____

Address_____

Reason for Referral_____

Medical Diagnosis (if known) _____

List any Health Problems you (the patient) have:_____

_____

_____

List any Hospitalizations (place, reasons & dates)

Name and Location	Reason	Date
_____	_____	_____
_____	_____	_____
_____	_____	_____

What questions do you have that you would like answered?_____

_____

_____

_____

_____

_____

The Index Patient's Brothers/Sisters and their Children

List your brothers/sisters. Please include stillbirths(sb), miscarriages(m) and those deceased(d).

Name of Sibling	Date of Birth mo/yr	Sex	Present Health	Sibling's Children (list age & sex)

Are any of the above half-brothers/sisters and/or step-brothers/sisters?_____

_____

Are any of the above adopted or foster children?_____

_____

Biological Mother of Index Patient

Name_____ Maiden (family) name_____

Date and place of birth_____ Ethnic origin_____

Present Health_____

_____

_____

Mother's Brothers and Sisters and their Children
(include stillbirths, miscarriages and deceased)

Name of Mother's Sibling	Date of Birth mo/yr	Sex	Present Health	Mother's Sibling's Children (list age and sex)

Are any of the above half-brothers/sisters and/or step-brothers/sisters?_____

_____

Other information of significance_____

_____

_____

Maternal Grandfather

Name _____

Ethnic origin_____ Date & Place of Birth_____

How many brothers?_____ How many sisters?_____

Present Health (if deceased, date and cause of death)_____

_____

Maternal Grandmother

Name _____

Ethnic origin_____ Date & Place of Birth_____

How many brothers?_____ How many sisters?_____

Present Health (if deceased, date and cause of death)_____

_____

Is there anyone else on the maternal side of the family that has any birth defects, mental retardation, or any other health concerns not yet mentioned? List each person affected and identify the problems.

_____

_____

_____

Biological Father of Index Patient

Name_____ Maiden (family) name_____

Date and place of birth_____ Ethnic origin_____

Present Health_____

_____

_____

Fathers's Brothers and Sisters and their Children
(include stillbirths, miscarriages and deceased)

Name of Father's Sibling	Date of Birth mo/yr	Sex	Present Health	Father's Sibling's Children (list age and sex)

_____ _____ _____ _____ _____

_____ _____ _____ _____ _____

_____ _____ _____ _____ _____

Are any of the above half-brothers/sisters and/or step-brothers/sisters?_____

_____

Other information of significance_____

_____

_____

Paternal Grandfather

Name _____

Ethnic origin_____ Date & Place of Birth_____

How many brothers?_____ How many sisters?_____

Present Health (if deceased, date and cause of death)_____

_____

Paternal Grandmother

Name _____

Ethnic origin_____ Date & Place of Birth_____

How many brothers?_____ How many sisters?_____

Present Health (if deceased, date and cause of death)_____

_____

Is there anyone else on the paternal side of the family that has any birth defects, mental retardation, or any other health concerns not yet mentioned? List each person affected and identify the problems.

_____

_____

_____

Family History Questionnaire

Please complete the chart below. Provide as much information as you are able, and use the back or attach additional sheets as needed. Do not include information about family members who are not biologically related to you.

Mother's Side of Family (Biological)				Father's Side of Family (Biological)			
✓ If Living Current Age	✓ If in Good Health	Relationship	1) Cause of death 2) Age at death 3) Health problems	✓ If Living Current Age	✓ If in Good Health	Relationship	1) Cause of death 2) Age at death 3) Health Problems
		Grandmother				Grandmother	
		Grandfather				Grandfather	
		Aunt(s)				Aunt(s)	
		Uncle(s)				Uncle(s)	

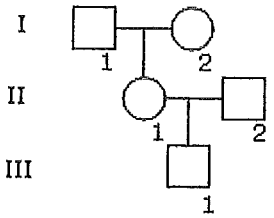
Biological Mother				Biological Father			
✔ If Living	✔ If in Good Health	Name and current age if living	1) Cause of death 2) Age at death 3) Health problems	✔ If Living	✔ If in Good Health	Name and current age if living	1) Cause of death 2) Age at death 3) Health problems

Your Sisters				Your Brothers			
✔ If Living	✔ If in Good Health	Name and current age if living	1) Cause of death 2) Age at death 3) Health problems	✔ If Living	✔ If in Good Health	Name and current age if living	1)Cause of death 2) Age at death 3) Health problems

Your Daughters				Your Sons			
✔ If Living	✔ If in Good Health	Name and current age if living	1) Cause of death 2) Age at death 3) Health problems	✔ If Living	✔ If in Good Health	Name and current age if living	1) Cause of death 2) Age at death 3) Health problems

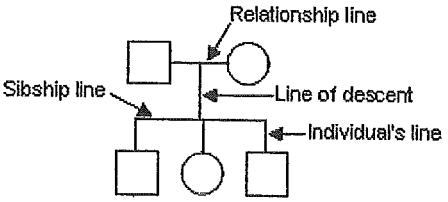
Common pedigree symbols

	Male	Female	Sex Unknown
Individual			
Affected individual			
Multiple individuals	5	5	5
Multiple individuals, number unknown	n	n	n
Deceased individual			
Pregnancy	P	P	P
Proband	P	P	P
Consultand			
Spontaneous abortion	male	female	ECT (if ectopic)
Termination of pregnancy	male	female	
Obligate carrier			

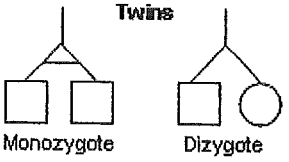


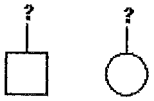
Roman numerals indicate generations; Arabic numerals indicate specific individuals within a certain generation (i.e., individual I-2 is the maternal grandmother of individual III-1).

Relationships

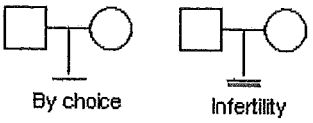


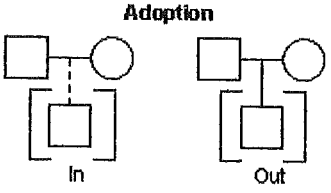
Consanguineous relationship

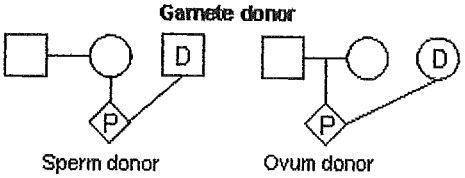


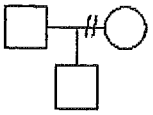


Family history unknown





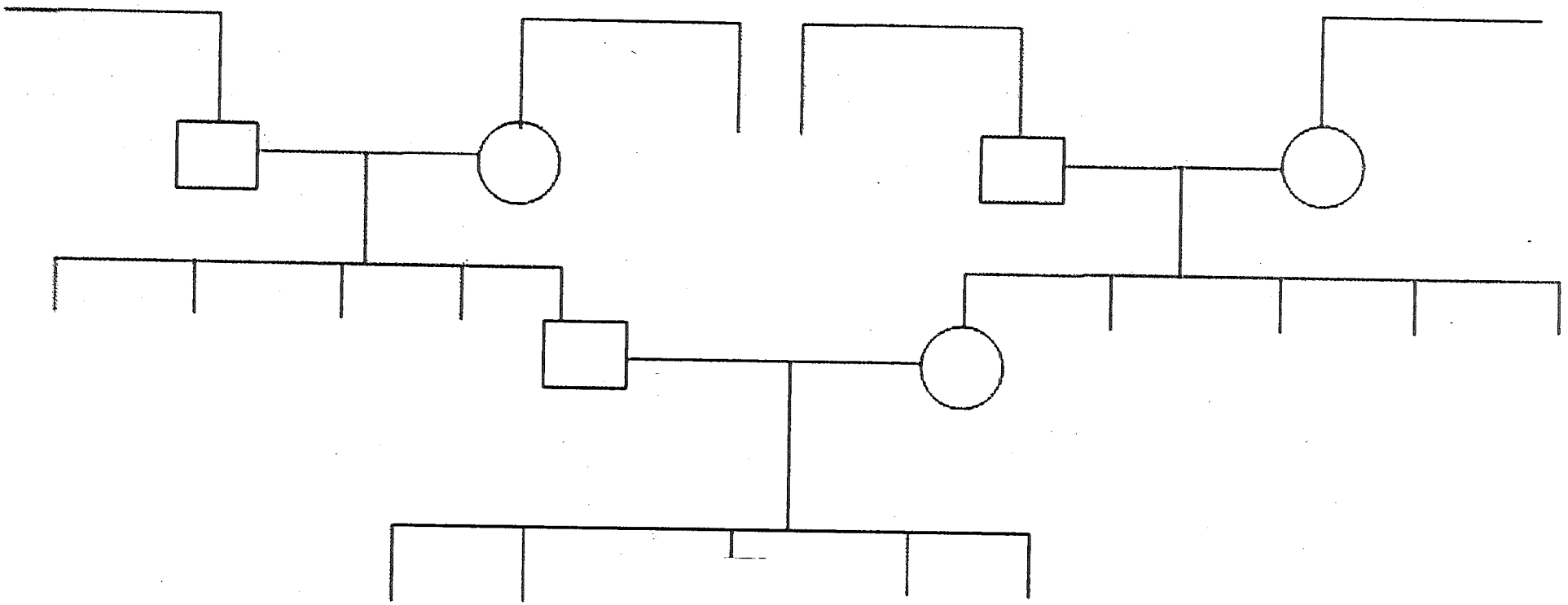




Relationship no longer exists

Symbols adapted from Bennett et al (1995) Recommendations for standardized human pedigree nomenclature. *Am J Hum Genet* 56:745-52.

Your Family History



YOU

NAME: _____

Interpreting the Family History

EVALUATION OF MEDICAL AND FAMILY HISTORY TO INFORM RISK

Because the family history is used to observe patterns in the family and to assess risk, the family history cannot benefit the patient if there is no interpretation and follow-up. Families at increased risk may be offered additional screening or treatment and encouraged to undertake lifestyle modification. Consultation and referral to genetics is appropriate in cases where risk appears to be significantly increased.

More information about how to evaluate the family history, through the use of genetic red flags, follows.

Genetic red flags

Genetic "red flags" are indications that there might be increased genetic risk in an individual or family. The primary red flag for most common diseases is a large number of affected relatives with a close degree of relationship. This can indicate unusually high genetic and/or environmental risk, and the risk for close relatives may be increased dramatically. Generally, the same red flags increase risk for Mendelian and common disorders.

Some genetic red flags include:

Family history of multiple affected family members with the same or related disorders, which may or may not follow an identifiable pattern in the family

> Such a pattern indicates increased risk, whether through genetic or environmental risk factors, or a combination of genes and environment.
>
> Examples: Three family members in two generations with heart disease; a father and son with diabetes

Earlier age at onset of disease than expected

> Disorders that arise at a younger age than expected may occur because of a genetic predisposition that makes an individual more susceptible to environmental exposures.
>
> Examples: Heart disease occurring in the 30s; lung cancer in the 40s

Condition in the less-often-affected sex

> A disorder that occurs in the less common sex may occur because of a genetic predisposition that overrides other hormonal, developmental and environmental factors that contribute to its occurrence.
>
> Examples: Breast cancer in a male; persistent stuttering in a female

Disease in the absence of known risk factors

> Genetic predisposition may lead to the occurrence of a disorder in the absence of obvious environmental factors.

Example: hyperlipidemia in an individual with an ideal diet and exercise regimen

Ethnic predisposition to certain genetic disorders

Some genetic disorders are more common in certain ethnic groups. Awareness of a patient's ethnicity or ancestral background can aid in recommending genetic testing and evaluation of genetic conditions.

Example: lactose intolerance in an individual of African ancestry

Close biological relationship between parents (i.e., consanguinity)

Consanguinity is a relationship by blood or a common ancestor. Because relatives are more likely to share the same genes, children from a consanguineous couple related as first cousins or closer have an increased risk of having an autosomal recessive condition.

Examples: autosomal recessive disorders, including those that are part of the newborn screen (including MCADD and cystic fibrosis), are more common in consanguineous couples.

Fact Sheet

U.S. Department of Labor
Employee Benefits Security Administration
September 2009

The Genetic Information Nondiscrimination Act of 2008 (GINA)

The Genetic Information Nondiscrimination Act of 2008 (GINA) prohibits discrimination in group health plan coverage based on genetic information. GINA is effective for plan years beginning after May 21, 2009 (January 1, 2010 for calendar year plans). Regulations implementing the provisions of GINA were made public on October 1, 2009.

Builds on HIPAA's protections. GINA expands the genetic information protections included in the Health Insurance Portability and Accountability Act of 1996 (HIPAA). HIPAA prevents a plan or issuer from imposing a preexisting condition exclusion provision based solely on genetic information, and prohibits discrimination in individual eligibility, benefits, or premiums based on any health factor (including genetic information).

Additional underwriting protections. GINA provides that group health plans and health insurance issuers cannot base premiums for an employer or a group of similarly situated individuals on genetic information. (However, premiums may be increased for the group based upon the manifestation of a disease or disorder of an individual enrolled in the plan.)

Prohibits requiring genetic testing. GINA also generally prohibits plans and issuers from requesting or requiring an individual to undergo a genetic test. However, a health care professional providing health care services to an individual is permitted to request a genetic test. Additionally, genetic testing information may be requested to determine payment of a claim for benefits, although the regulations make clear that the plan or issuer may request only the minimum amount of information necessary in order to determine payment. There is also a research exception that permits a plan or issuer to request (but not require) that a participant or beneficiary undergo a genetic test.

Restricts collection of genetic information. GINA also prohibits a plan from collecting genetic information (including family medical history) prior to or in connection with enrollment, or for underwriting purposes. Thus, under GINA, plans and issuers are generally prohibited from offering rewards in return for collection of genetic information, including family medical history information collected as part of a Health Risk Assessment (HRA). The regulations provide several examples illustrating GINA's application to HRAs.

An exception is included for incidental collection, provided the information is not used for underwriting. However, the regulations make clear that the incidental collection exception is not available if it is reasonable for the plan or issuer to anticipate that health information will be received in response to a collection, unless the collection explicitly states that genetic information should not be provided.

Other protections. GINA also contains individual insurance market provisions, administered by the Department of Health and Human Services's Centers for Medicare & Medicaid Services, privacy and confidentiality provisions, administered by the Department of Health and Human Services's Office for Civil Rights, and employment-related provisions, administered by the Equal Employment Opportunity Commission (EEOC).

The Role of the Registered Nurse as Related to Genetic Testing Clinical Position Statement

The registered nurse, as part of the multidisciplinary health care team, should participate in the provision of care for women and newborns considering or undergoing genetic testing and treatment. The registered nurse should be prepared to assist clients with accurate and complete information to enable them to make informed decisions related to genetic testing for themselves or for prenatal or newborn purposes. As members of the health care team, registered nurses must be prepared to work with patients in a manner that supports the decision-making process inherent to genetic screening or genetic evaluation, in cooperation with other health care providers, including physicians and genetics counselors and specialists. *

Registered nurses should be familiar with and comply with institutional, state and federal policies to ensure that any medical information that results from genetic testing remains confidential. Fundamental, evidence-based education about genetics and genetic counseling should be included as part of basic nursing education and registered nurses who work in related practice settings are encouraged to consider continuing education in this area in order to maintain the clinical competency necessary to provide optimal care to their clients.

Women should be encouraged to maximize the potential knowledge and health benefits gained from discovering important genetic information for themselves or for prenatal or newborn purposes, however, mandatory requirements for women to undergo testing would be intrusive and inappropriate. Health care providers should not pressure women to undergo genetic testing for themselves or for prenatal or newborn purposes.

Background: Genetic testing and genetic evaluation creates complex ethical, legal and social issues for women and their families. The rapid advances in genetic testing have resulted in the availability of increasing amounts of information for women and their families. As the public becomes more aware of options for genetic testing and its role in disease identification and management, nurses in most areas of practice will be increasingly called upon to address basic genetics-related questions, and women and families will continue to face challenging decisions regarding the results of this information.

Registered nurses must be sensitive to these complexities and be prepared to support women and their families in their decision-making processes. As members of a multidisciplinary team of health care providers involved in genetic testing and screening, nurses must be prepared to understand the implications of the results of genetic testing and provide support for women and families in their decision-making. Nurses who are knowledgeable about the management of genetic information and patient issues can foster patient understanding and assist with adaptation strategies irrespective of the outcomes of genetic counseling or testing.

Some key components related to genetics and genetic testing include:

- Basic mechanisms of heredity and mutation
- Potential risks and benefits of testing
- Fundamental technical components of testing
- Sensitivity and specificity of tests
- Key issues of informed consent and decision-making, including freedom of choice for patients
- Knowledge of available resources, including referrals to genetics counselors or specialists and relevant support groups
- Ethical, legal and social components
- Patient management issues

*AWHONN recognizes that while registered nurses should be prepared to support clients related to genetic testing, they do not and should not be expected to fill the role of a genetics counselor or specialist. Genetics nursing is a separate clinical specialty that focuses on providing nursing care to clients who have known

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References and Resources

American Nurses Association Consensus Panel. (2008). *Essentials of genetic and genomic nursing: competencies, curricula guidelines, and outcome indicators*(2nd ed.). Silver Spring, MD: American Nurses Association. [online]. Available: http://genome.gov/17517037

Bennett, R.L., French, K.S., Resta, R.G. & Doyle, D.L. Standardized human pedigree nomenclature: update and assessment of the recommendations of the National Society of Genetic Counselors. *J Genet Couns* **17**, 424-33 (2008). *Drawing a Family Pedigree.[online]. Available:* http://www.medicine.uiowa.edu/uploadedFiles/Research/Human_Genetics/Pages/Clinical_Genetics/IIHG%20How%20to%20Draw%20a%20Pedigree.pdf

Calzone, K. A., Jenkins, J.(2013). Global Genetics/Genomics Community: Interactive Case Studies. [online]. Activate: http://g-3-c.org/en/

Calzone, K. A., Jerome-D'Emilia, B., Jenkins, J., Goldgar, C., Rackover, M., Jackson, J.,...Feero, W. G. (2011). Establishment of the genetic/genomic competency center for education. *Journal of Nursing Scholarship*, *43*(4), 351-358.

Centers for Disease Control and Prevention (CDC). (2001). *Genomic Competencies for the Public Health Professionals* [online]. Available: http://www.cdc.gov/genomics/

Cincinatti Children's Hospital Medical Center. (2013).*The Genetics Education Program for Nurses (GEPN):Self-paced modules for nurses* [online]. Avaialble at http://www.cincinnatichildrens.org/education/clinical/nursing/genetics/cont/self/default/

Health and Human Services. *My family portrait*[online] Available: https://familyhistory.hhs.gov/fhh-web/home.action

International Society of Genetics and American Nurses Association. (2006). *Genetics/Genomics Nursing: Statement on the Scope and Standards of Practice.* Silver Spring, MD.: Nursesbooks.org.

International Society of Genetics and American Nurses Association. (1998). *Statement on the Scope and Standards of Genetics Clinical Nursing Practice*. Washington, DC: American Nurses Association [online]. Available: http://www.isong.org/

Kahn, S. (2013). Kahn Academy: *Heredity and genetics biology*. [online]. Available: http://www.khanacademy.org/science/biology/heredity-and-genetics

National Center for biotechnology Information. U.S.Library of Medicine. National Institute of Health. (2004). A science primer: Just the facts. Retrieved from http://www.ncbi.nlm.nih.gov/About/primer/pharm.html

National Coalition for Health Professional Education in Genetics (nchpeg). (2013). *Interpreting the family history: Evaluation of medical family history to inform risk*. [online]. Available : http://www.nchpeg.org/

National Institute of Environmental Health Sciences (NIEHS). (2013). *Environmental Genome Project* [online]. Available: http://www.niehs.nih.gov/

National Institute of health (NIH). (2013). [online]. Avaialable: http://www.nih.gov

National Institute of Health (NIH). (2013). National Cancer Institute [online]. Available: http://www.cancer.gov/cancertopics/genetics

National Institute of Health (NIH) National genome Research Institute. (2013). *Genetics/Genomics Competency Center for Education.* [online]. Available: http://www.g-2-c-2.org/index.php

U.S. Department of Labor, Employee Benefits Security Administration. (2009). The genetic information act of 2008 (GINA) [online]. Available: http://www.dol.gov/ebsa/pdf/fsgina.pdf

U.S. National Library of Medicine. (2013). *Genetic Home Reference*: *Your guide to understanding genetic conditions.* [on-line]. Available: http://ghr.nlm.nih.gov/