

Genomics and Genetics in Healthcare

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Clinical Objectives



- Understand the importance of genomics to provide effective nursing care
- Integrate genetic knowledge and skills into nursing practice
- Utilize additional online resources for genomics

What Would you Tell Your Patients When They Ask You:

- “How is Muscular Dystrophy passed from one generation to the next?”
- “How has genomics research affected the treatment of cancer?”
- “Are there genetic tests for Alzheimer’s disease?”
- “Do I need genetic counseling for a family history of DVT and positive Factor V Leiden?”

Genetics

- Nurses need to understand the basics of genetics and genomics in order to help counsel or refer their patients
- Genetic diagnosis and research quickly changes with new technologies and discoveries
- Current information is available on internet resources

Definitions

- Genetics is the study of individual genes
- Genes are sequences of DNA, at specific positions on chromosomes
- They provide critical codes that translate into proteins
- Genomics is the interaction of **all** genes and environmental factors

Genes: Basic Units of Heredity

- Genes are sections of DNA that provide the instruction to our bodies to to make up who we are
- Genes are in 48 chromosomes, passed from parent to child by the sperm and the egg
- 23 pairs of chromosomes carry all of our genetic information

Mutations

- Many diseases result from abnormal mutations in certain genes
- Inheriting a genetic mutation from one parent doesn't always put you at risk for disease
- Sometimes it takes only one damaged copy of a gene, but sometimes it takes two

Inheriting Disease

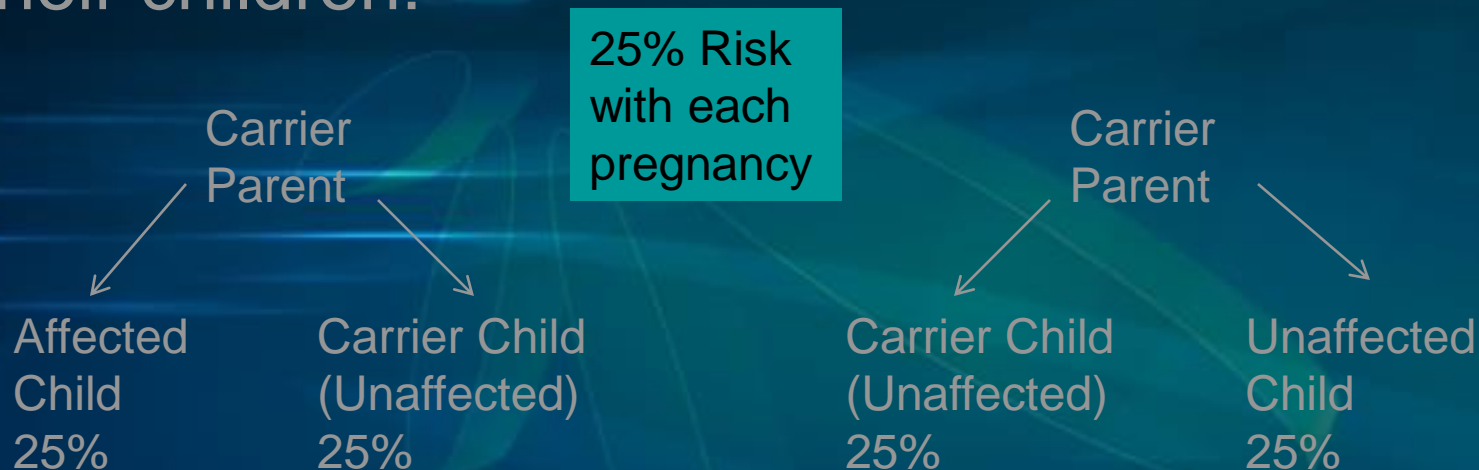
- Autosomal Recessive (contained in chromosome pairs 1 through 22)
- Autosomal Dominant
- X-Linked (contained in the sex chromosome)
- Mitochondrial Inheritance

Autosomal Recessive

- It usually takes two copies of a mutated version of the gene (one from each parent) for the person to develop the trait or disease
 - Hemochromatosis (an iron storage disorder)
 - Cystic Fibrosis
 - Sickle Cell Anemia, Thalassemias
 - Tay Sachs, Gaucher Disease, Fanconi Anemia (typically Jewish diseases)
 - Most Metabolic Diseases

Inheritance Pattern

- People who carry one copy of a recessive mutation are called carriers for that trait
- They do not have the disease, but can pass it on to their children:

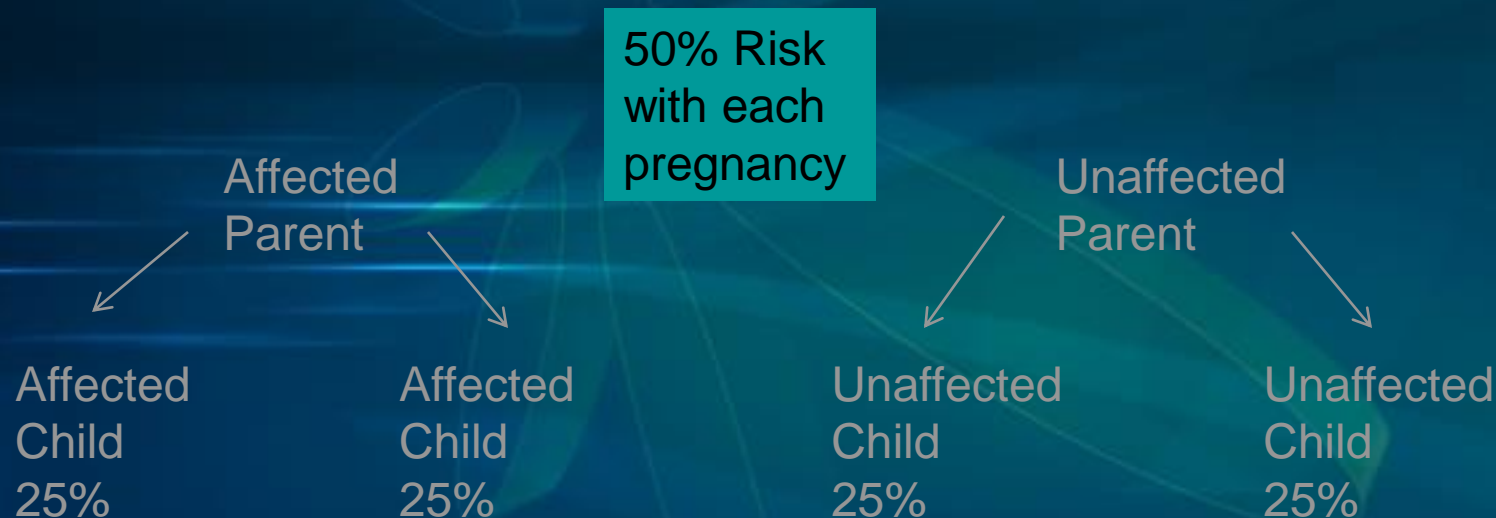


Autosomal Dominant

- Only one parent has to pass on the altered gene for that trait to be expressed
 - Familial adenomatous polyposis (polyps that predispose to colon cancer)
 - Huntington's Disease
 - Familial Hypercholesterolemia
 - Marfan's Syndrome
 - Neurofibromatosis
 - Von Hippel-Lindau Syndrome

Inheritance Pattern

- The risk of passing on a dominant gene is much greater than a recessive gene

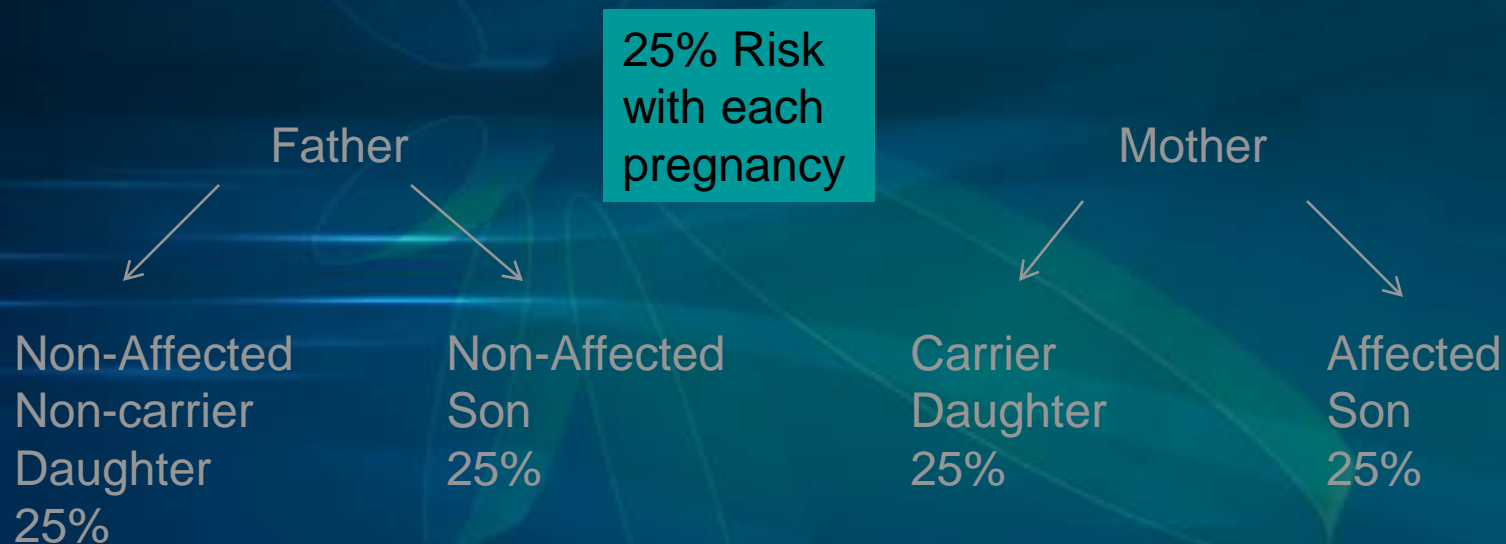


X-Linked

- X and Y chromosomes contain many genes that the autosomal chromosomes do not
 - Males have one Y chromosome and one X
 - Females have two X chromosomes
- Men and women differ in how they inherit certain diseases
 - Hemophilia
 - Color blindness
 - Fragile X can cause hereditary mental retardation

Inheritance Patterns

- Men are more likely to get X-linked disorders because they have no normal X



Mitochondrial Inheritance

- Each cell has many mitochondria that produce energy for the cell
- Inherited from our mother's egg (none in sperm)
- Mitochondria have their own DNA that can be altered and cause disease
 - Maternally Inherited Diabetes
 - Deafness
 - Muscular Dystrophy
 - Neuronal Epilepsy

Diagnosis

- Karyotype is an organized picture of the chromosomes found in a cell. It can detect extra, missing, or abnormal chromosomes:
 - Trisomy 21 (Down's Syndrome)
 - Alzheimer's Disease
 - Klinefelter Syndrome
 - Turner Syndrome

Deletions

- During cell division, some chromosomes may break and DNA segments may be deleted
- Bands on each chromosome are mapped and can be identified by a number
- A letter p means the long arm of the chromosome was broken, or q means the short arm was affected
- If the material deleted is essential, it results in disease

Examples

Deletions

- Cri du chat, 5p
- WAGR, 11p
- Rubinstein-Taybi, 16p
- Miller-Dieker and Smith-Magenis, 17p

Duplications

- Charcot-Marie-Tooth, 17p

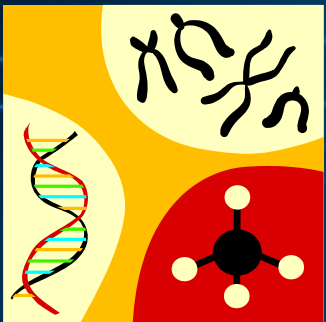
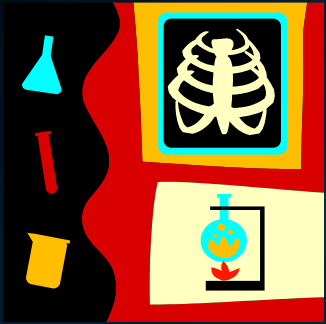
Complex Inheritance

- Many human inherited diseases result from effects and interaction of two or more genes
- Can have protective genes or those with negative effects causing illness or predisposition to illness
 - Coronary Artery Disease
 - Hypertension, Stroke
 - Mental illness
 - Various forms of cancer
 - Arthritis

Multifactorial Risks

- Health, growth and development include environmental component
- Chemical exposure and the internal environment of body can affect risk of deletions
 - DNA mutations can be spontaneous, or caused by chemicals, viruses, or radiation
- Older mothers have more risk of mosaicism, (some abnormal cell development after gamete production)

Pharmacogenomics



- The study of how an individual's genetic inheritance affects the body's response to drugs by combining traditional pharmaceutical sciences with annotated knowledge of genes, proteins, and single nucleotide polymorphisms (DNA sequence variations)

Genetic Testing

- DNA-based tests examine the DNA molecule itself
- Obtained from any tissue, or from blood
 - Biochemical tests examine enzymes and other proteins
 - Microscopic examinations look at stained or fluorescent chromosomes
 - Pre-implantation genetic diagnosis (PGD) screens for genetic flaws among embryos used in vitro fertilization

Pros

- Genetic testing can clarify diagnosis and enable appropriate treatments (e.g. cancer cell characterization)
- Information for families weighing their risks for familial genetic diseases
- People with high risk for preventable illness could improve lifestyle or environment
- Used in newborn screening
- Forensic/identity testing

Cons

- Costs can range from hundreds to thousands of dollars and insurances rarely cover them
- Commercialized gene tests for adult-onset disorders (Alzheimer's disease and cancers)
- Companies are targeting healthy people
- Results give only a probability of developing the illness
- Genetic testing is not regulated in U.S.

Human Genome Project

- Current Information for health care providers available at www.doegenomes.org
 - Ethical, Legal, and Social Issues
 - Genetic Counseling
 - Pharmacogenomics
 - Minorities, Race, and Genetics
 - Gene Testing (more than 900 tests available)

Resources

- Diagram of DNA: The Molecule of Life at <http://www.ornl.gov/hgmis>
- Mayo Clinic Genomics website- terminology <http://mayoresearch.mayo.edu/biobank/glossary-of-terms.cfm>
- National Coalition for Health Professional Education in Genetics at <http://www.nchpeg.org/content.asp?dbsection=about>

More Resources

- Centers for Disease Control (and family history tool) at www.cdc.gov/genomics/update/current.htm
- www.infogenetics.org
- www.geneclinics.org
- www.geneticalliance.org disease information, support and advocacy groups

Questions for Nurses

- Would you consider genetic testing based on your patient's diagnosis?
- Is genetic testing available for that disease?
- What lab is doing that kind of testing?
- Is it diagnostic or research testing?
- Where and how do you send in a sample?

Ethical Questions To Consider

- Should this patient be referred for genetic counseling?
- Is the cost of testing worth the benefit?
- Is genetic testing and research truly confidential?
- Will having a genetic condition affect a person's ability to get health or life insurance?

Conclusion

- Have you developed basic competency with genetic terms and concepts?
- It is vital to educate and promote genomics in schools of nursing and among staff nurses
- Genomics is an important tool in health promotion and for nursing care in any setting

