

Slide 1

Genetics the Forefront of Healthcare

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Drexel  
College of Nursing & Health  
Professions

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Slide 2

Objectives

Describe the impact of genetics and genomics and how nurse educators can prepare students to apply the information into practice by:

- *Identifying patients and families with, or at risk of, genetic conditions*
- *Indications for referral to specialist*
- *Ordering and understanding genetic test results and recommending treatments*
- *Implications for patient with condition and for other family members*

Identify challenges of new genetic technologies and the impact on patient care and families

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Slide 3

Human Genome Project

- It June 26, 2000 that US President Bill Clinton and UK Prime Minister Tony Blair announced the successful completion of a "draft sequence" of the human genome.
- The project itself had begun a decade earlier, through government funding from the US Department of Energy and the National Institutes of Health, and a framework of international collaboration

Image  
<http://neetmagazine.com/content/article/human-genome>

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Slide 4

**International Hap-Map Project Overview**

- The haplotype map, or "HapMap," is a tool that allows researchers to find genes and genetic variations that affect health and disease.




image  
• <http://www.genome.gov/10001688>

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**Nursing Faculty is the Key**

- In the 21st century, it is imperative that nurse educators in advanced practice expand upon the science of genetics and new technologies surrounding the science
- The new technology should explore the impact on healthcare of the individual and families.
- Government U.S. public surveys have indicated an interest in using genomic technology in their health care
- Nurses need to know the science to communicate on an interdisciplinary level



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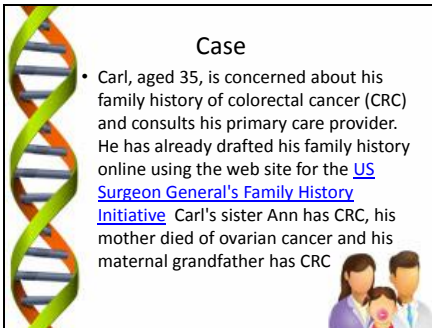
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**Case**

- Carl, aged 35, is concerned about his family history of colorectal cancer (CRC) and consults his primary care provider. He has already drafted his family history online using the web site for the [US Surgeon General's Family History Initiative](#). Carl's sister Ann has CRC, his mother died of ovarian cancer and his maternal grandfather has CRC



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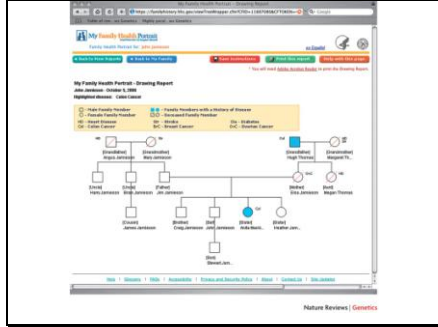
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### Case

- Carol is 10 weeks pregnant. At her first appointment she tells her obstetrician that her paternal aunt has two sons with a learning disability.
- A nurse practitioner student who is a friend has reassured Carol that there would be no risk to her baby because the problem is "on her dad's side".

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The most important is collection of the patient's history and physical exam

### MEDICAL HISTORY

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### Family Medical History

- Congenital anomalies
- Mental retardation
- Known genetic disease
- Metabolic disorders
- Chronic serious illness
- Seizures
- Hemoglobinopathies
- Hearing impairment
- Multiple miscarriages
- Multiple stillbirths
- Significant learning disorders
- Psychiatric issues
- Ancestry, ethnicity
- Consanguinity



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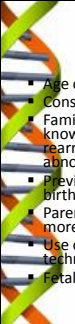
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### Pregnancy History

- Age of mother
- Consanguinity
- Family members with known chromosomal rearrangements or abnormalities
- Previous premature births
- Parental infertility ( 3 or more miscarriages)
- Use of reproductive technology
- Fetal ultrasounds
- Amniotic fluid abnormalities
- Blood screening (triple/quadruple screen )
- Medications or drug abuse
- HELLP syndrome
- Bleeding Disorders (Deficiency Anti-thrombin, Factor V Leiden, protein S and C)
- **Known Teratogens**



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### The Neonatal History

- Symmetric intrauterine growth restriction
- Large for gestational age without a reason
- Hearing loss
- Persistent hyperbilirubinemia
- Poor adaptation to the environment
- Hypotonia or hypertonia
- Seizures
- Remarkable metabolic screen results
- Major organ disruption



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### The Pediatric History

- Developmental delay,
- Learning disabilities,
- Congenital anomalies,
- Hearing or visual impairments,
- Dysmorphic features,
- Metabolic disorders



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...our patients with genetic eye

### PHYSICAL EXAM



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### Physical Examination

- General dysmorphology
- -Minor and major anomalies
- Growth parameters and neurological evaluation
- >3 minor anomalies - 20% risk of major anomaly or mental retardation
- -Single major anomaly- think genetic investigation!



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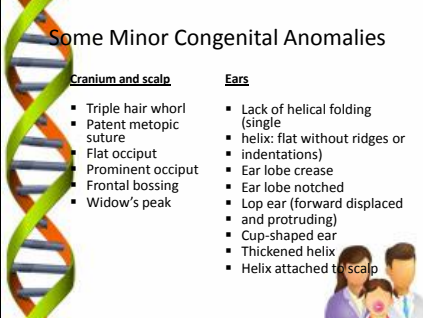
### Some Minor Congenital Anomalies

**Cranium and scalp**

- Triple hair whorl
- Patent metopic suture
- Flat occiput
- Prominent occiput
- Frontal bossing
- Widow's peak

**Ears**

- Lack of helical folding (single helix: flat without ridges or indentations)
- Ear lobe crease
- Ear lobe notched
- Lop ear (forward displaced and protruding)
- Cup-shaped ear
- Thickened helix
- Helix attached to scalp



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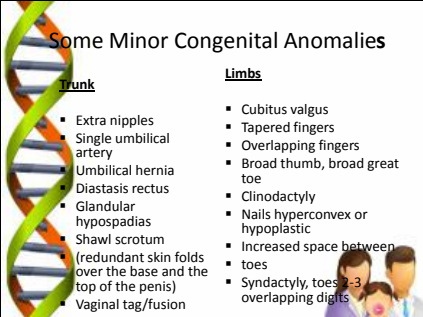
### Some Minor Congenital Anomalies

**Trunk**

- Extra nipples
- Single umbilical artery
- Umbilical hernia
- Diastasis rectus
- Glandular hypospadias
- Shawl scrotum (redundant skin folds over the base and the top of the penis)
- Vaginal tag/fusion

**Limbs**

- Cubitus valgus
- Tapered fingers
- Overlapping fingers
- Broad thumb, broad great toe
- Clinodactyly
- Nails hyperconvex or hypoplastic
- Increased space between toes
- Syndactyly, toes 2-3 overlapping digits



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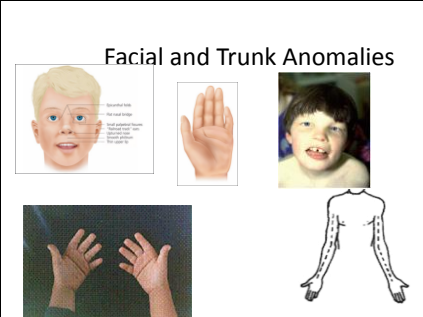
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### Facial and Trunk Anomalies



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**Major Congenital Anomalies**

- Cleft lip, cleft palate
- Congenital heart disease
- Brain anomalies, neural tube defects
- **Omphalocele**
- Specific skin lesions
- Generalized dysmorphism and growth restriction
- Asymmetry (facial, skeletal, limb)
- Hepatosplenomegaly

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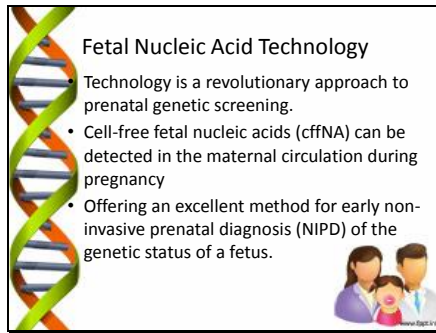
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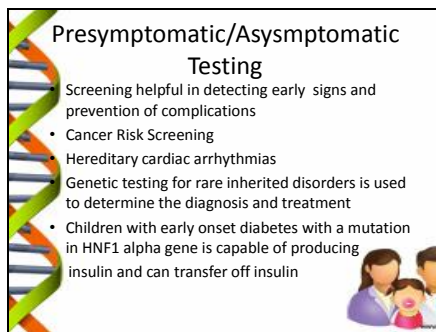
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### FISH Testing

- Fluorescence in situ hybridization:
- Uses fluorescent labeled chromosome-specific DNA segments
- **\*You need to know what you are fishing for\***
- Probe lights up when exposed to ultraviolet (UV) light.
- Viewed under scope to identify missing, additional or rearranged chromosomal material



© Copyright 2003, Karolinska Institute, Stockholm, Sweden. Group: Ricki Berg (PhD) D13  
Note: This karyotype was prepared using a FISH technique known as "chromosome painting". As well as labeling of chromosomes from chromosome 22, chromosome 9 and has been collected material from chromosome 8.

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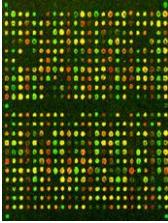
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### Whole genome array comparative genomic hybridization

- A tool used to sift through and analyze the information contained within a genome.
- Same genes in all cells; All genes not active at same time
- Detect abnormal numbers of DNA sequences (deletions, duplications) of very small segments
- Efficient method to confirm clinical suspicion of certain conditions
- Very useful with DD/MR with minor anomalies



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### New Era of the Internet



- Direct to consumer testing a new paradigm
- Sold via the internet and not gone under clinical evaluation
- No direct consult with healthcare provider
- Value of the test and ramifications not clear and challenges to health care providers



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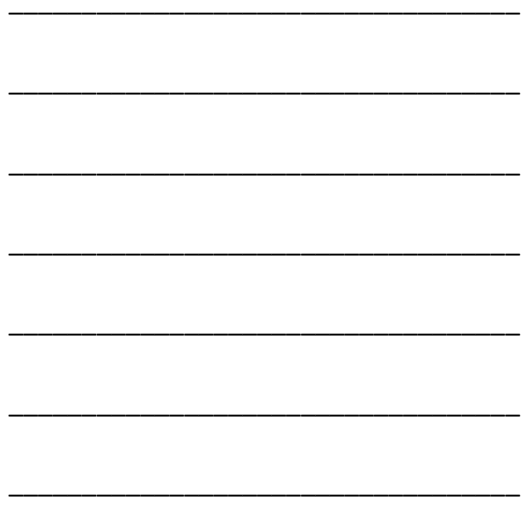
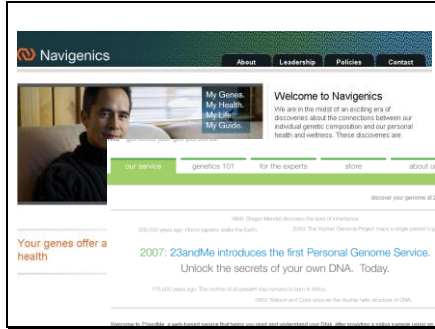
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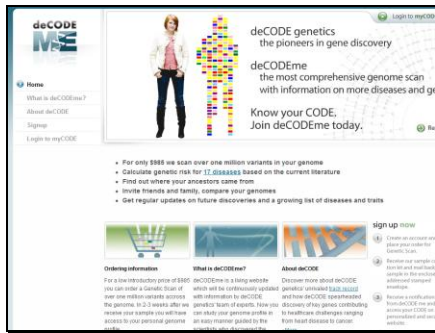
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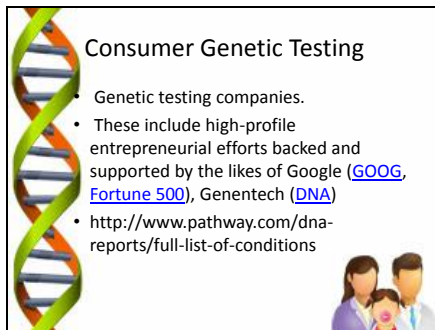
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Could genetics improve prescribing drugs?



**JAMA**  
ORIGINAL CONTRIBUTION

Association of Cytochrome P450 2C19 Genotype With the Antiplatelet Effect and Clinical Efficacy of Clopidogrel Therapy

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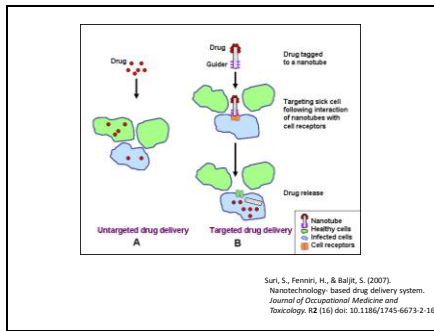
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### Role of APN in Genetic Referral

- Know when to refer
- Clarify and explain referral and results to family
- Educate and support family and patient
- Refer to support groups
- Follow-up in primary care
- Coordinate specialty care and work with interdisciplinary team
- Educate school personnel
- Advocate for educational services



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### Psychosocial Issues Related to a Genetic Diagnosis

- Emotional response to the news
  - Feelings of stigmatization
  - Fear of discrimination
- Family implications; impact on family and work relationships
- Implications for reproductive choices
  - Community and culture
  - Diminished access to medical services
  - Relationship to the medical community



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### Genetic Information Nondiscrimination Act (GINA)

- A federal law (passed 2008) that prevents health insurers and employers from discriminating based on an individual's genetic information
- The bill is intended to allow Americans to take advantage of the benefits of genetic testing without fear of losing their health insurance or their jobs



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### Summary of APN Competencies in Genetics

- Recognition the importance of family history
  - Clues in physical exam
  - 3 generation pedigree
- Referral for genetic testing and understanding the implications of genetic testing
- Understanding of new technologies and pharmacogenetics
- Interdisciplinary communication and referral for support
- Knowledge of the psychological, social, legal and ethical implications



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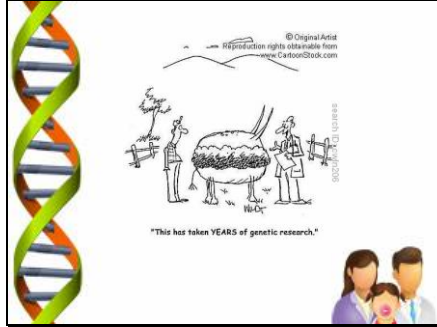
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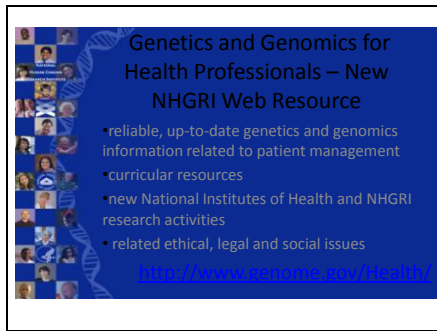
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### Other Clinical Resources

- GeneTests [www.genetests.org](http://www.genetests.org)
- Genetic Alliance [www.geneticalliance.org](http://www.geneticalliance.org)
- Genetics and Rare Diseases Information Center (GARD) [www.genome.gov/10000409](http://www.genome.gov/10000409)
- NCI [www.cancer.gov](http://www.cancer.gov)
- National Human Genome Research Institute <http://www.genome.gov/17517037>

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