Genetic Counseling Past and Future

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MotherToBaby AZ

What is Genetic Counseling?

- Communication process
  - Finding the best way to help the patient understand and apply information
- Health professionals who work with families who have or are at risk of having a member with a birth defect or inherited disorder
- MD or MS degree in genetics

Genetic Counseling Profession - Past

- 1947 - Sheldon Reed, a geneticist, coined the term “genetic counseling” to describe the process of providing genetic information and support to families
- 1969 – The first genetic counseling training program was opened in at Sarah Lawrence College by Melissa Richter
- 1971- First eight students graduated, six went into the field

https://www.lohud.com/story/opinion/2017/03/16/birth-genetic-counselor-prescient-melissa-richter/99267638/

Genetic Counseling Profession - Now

- Today
  - Approximately 4,000 ABGC certified genetic counselors
- Graduate programs
  - 41 graduate programs in U.S. with 4 in Canada
    Programs in U.K, Australia, Japan, S. Africa, Netherlands, Norway, Saudi Arabia, Israel, Cuba, France, Taiwan
- Future
  - 16 graduate programs are in the process of becoming established

What Information is Provided?

- **Genetic counselors have advanced training in medical genetics *and* counseling to guide and support patients seeking more information about such things as:**
  - How inherited diseases and conditions might affect them or their families
  - How family and medical histories may impact the chance of disease occurrence or recurrence
  - Which genetic tests may or may not be right for them, and what those tests may or may not tell
  - How to make the most informed choices about healthcare

Where Do Genetic Counselors Work?

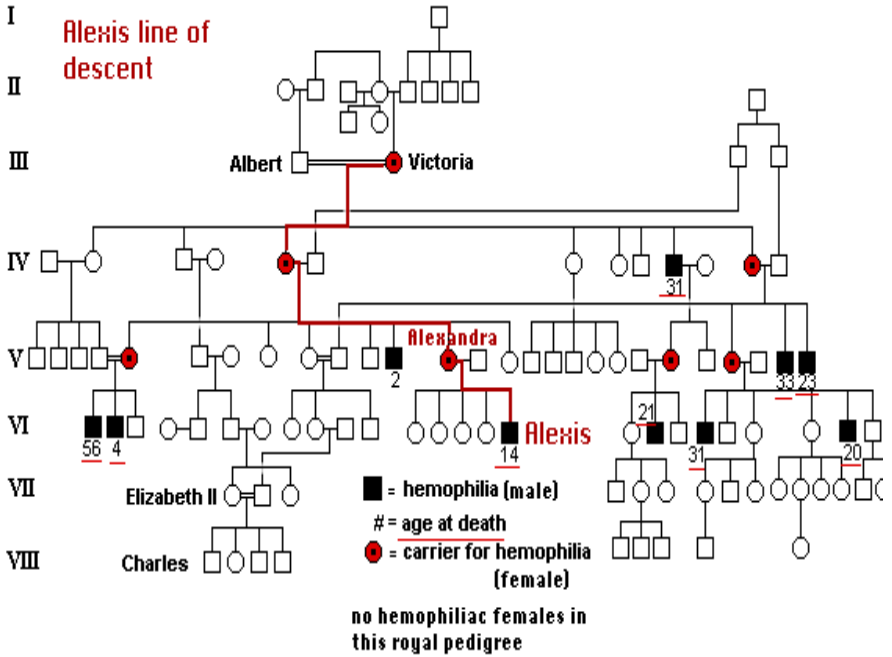
- **Most genetic counselors work in a clinic or hospital, and often work with geneticists, obstetricians, oncologists and other physicians. Genetic counselors can work in a variety of settings including (but not limited to):**
  - Assisted reproductive technology/infertility – for patients looking to become pregnant
  - Cancer – for patients with cancer and their family members
  - Cardiovascular – for patients with diseases of the heart or circulatory system and their family members
  - Laboratory – call out test results, write policy and procedures, organize/conduct research
  - Industry – genetic testing, pharmaceutical and more
  - Neurology – for patients with diseases of the brain and nervous system and their family members
  - Pediatric – for children and their family members
  - Prenatal and preconception – for women who are pregnant or thinking about becoming pregnant
  - Public Health – state health departments, federal agencies (NIH, CDC)

Provision of Genetic Counseling

- **Personal health history**
- **Family history**
  - Chromosome abnormalities
  - Genetic diseases or conditions that “run in the family”
  - Birth defects
  - Intellectual impairment
  - Multiple pregnancy losses/infertility
  - Ethnicity
    - Jewish, Mediterranean, Asian, African

Use of Family History

- Develop rapport
- Historical health of client and family
- Establish pattern of inheritance, calculate risks for family members
- Identify environmental factors such as occupation, education, access to resources and support systems
- Education of client and family
- Determine testing options
- Explore medical management and surveillance options



Different Counseling Areas

- Prenatal
- Teratogens
- Newborn Screening
- Pediatrics
- Cancer

Screening/Testing During Pregnancy

- **Ultrasound**
  - Nuchal translucency 11-13 weeks 6 days gestation
  - Level II sonogram at 18-20 weeks gestation
- **Diagnostic testing**
  - Amniocentesis
  - CVS
- **Carrier screening**
  - Ethnicity-based
  - Universal
- **Serum screening**
  - 1st and 2nd trimester screening
  - Cell-free fetal DNA (NIPT/S)

Exposures During Pregnancy: What is a teratogen?

- **A teratogen is any medication, chemical, infectious disease, or environmental agent that might interfere with the normal development of a fetus and result in the loss of a pregnancy, a birth defect, or adverse pregnancy outcomes.**

- **Potential effects:**
  - Pregnancy loss (miscarriage, stillbirth)
  - Pregnancy complications (preterm labor or delivery)
  - Poor fetal growth
  - Pattern of birth defects
  - Intellectual dysfunction; altered social behavior
  - Complications in the newborn (neonatal abstinence syndrome)

Newborn Screening

- State public health agencies have been administering NBS programs for over 40 years (initially starting with PKU in the 1960s) using dried blood spot cards collected from the newborn heel stick
- By April 2011, all states reported screening for at least 26 disorders on an expanded and standardized uniform panel. The most thorough screening panel checks for 80 disorders (California)
- AZ currently tests for 29 disorders, including hearing
- Most are metabolic disorders that respond to dietary and medical treatment
- Abnormal results are reported back to the pediatrician for treatment
- They are also used for evaluating current NBS tests and developing new ones, clinical and forensic testing, and epidemiologic studies

Pediatric Genetics

- **Usually involves MD geneticist to diagnosis condition in newborn or child such as:**
  - Birth defect
  - Genetic syndrome
  - Chromosomal abnormality
  - Metabolic disorder
  - Intellectual disability
  - Developmental delay/disability

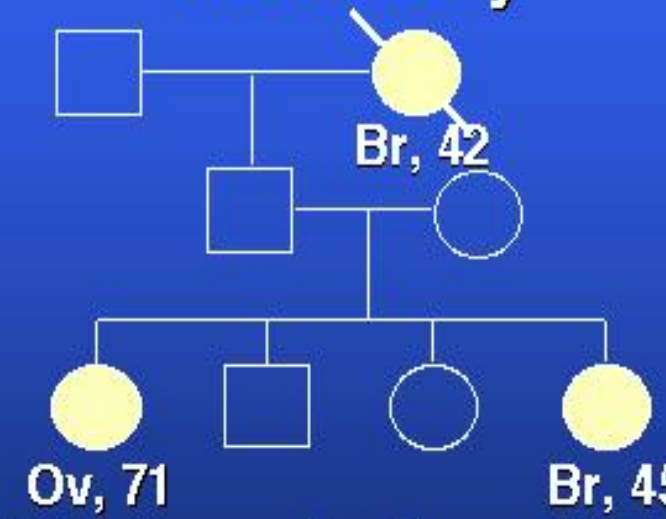
Cancer Genetics

- **Familial cancer syndromes account for about 10% of all cancers**

- **What to look for in the family history:**
  - Two or more close relatives with the same type of cancer (on the same side of the family)
  - Cancer diagnosed at an earlier age than usual
  - Cancer diagnosed more than once in the same person (more than one primary cancer, not a cancer recurrence)
  - Cluster of cancers associated with a known familial cancer syndrome (such as breast and ovarian)
  - Many cases of cancer in a family, more than can be accounted for by chance
  - Evidence of autosomal dominant inheritance

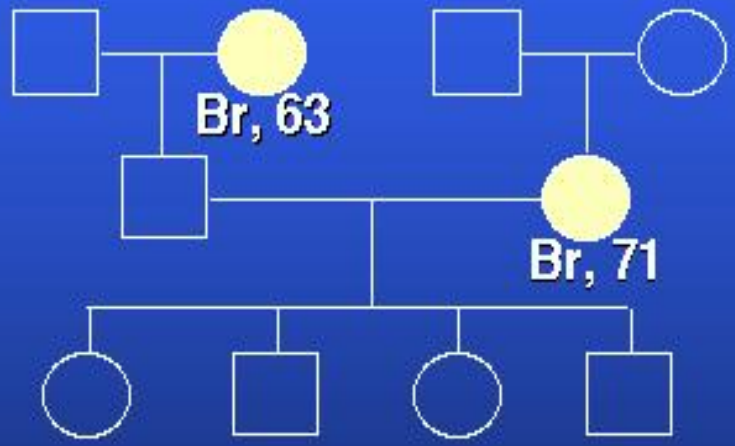
Family History of Hereditary Breast and Ovarian Cancer

Hereditary



- Two or more family members with breast cancer before age 50 or ovarian cancer at any age
- One family member with breast cancer before age 50 or ovarian cancer at any age, plus Ashkenazi ancestry

Sporadic



- None of the breast cancer is diagnosed before age 60
- No ovarian cancer
- No clear pattern on one side of family or other

New Engl J Med 2000;342:564-571

The Future

Whole Exome/Genome Sequencing

- **Used to identify variants that can lead to diagnosis and/or determine risk of disease**
- **American College of Medical Genetics recently produced document suggesting five categories to classify variants:**
  - Known pathogenic variants
  - Those that are likely to be pathogenic
  - Those of unknown significance
  - Those that are likely to be benign
  - Benign variants
- **Remaining questions:**
  - Cost-effectiveness
  - Accuracy and yield
  - Effective integration of genome-based diagnosis in medical care

Personalized Medicine/Pharmacogenomics

- **Use of individualized assessment for medication use by DNA analysis of metabolism genes**
- **Examples of pharmacogenetics include:**
  - Warfarin used as an anticoagulant but requires periodic monitoring and is associated with bleeding problems.
  - Genotyping for SNPs (single nucleotide polymorphisms) in 2 genes involved in the action and metabolism:
    - Cytochrome P450 enzyme (CYP 2C9), which metabolizes warfarin
    - Vitamin K epoxide reductase gene (VKORC1)
  - **Testing enables more accurate dosing that take into account the age, gender, weight, and genotype of an individual.**

MotherToBaby Arizona

- Free and confidential resource that provides up-to-date pregnancy and breastfeeding exposure information to health care providers and patients
- Also counsel for pre-conception and adoption
- Participate in educational outreach and national, multi-site research studies
  - 520-626-3410 or 888-285-3410
- OTIS is the national organization
  - 866-626-6847 or https://mothertobaby.org

MotherToBaby
ARIZONA
Medications & More During Pregnancy & Breastfeeding
Ask The Experts

University of Arizona Genetic Counseling Graduate Program

- Reinstitution of GCGP which existed from 1995-2005
- Currently awaiting accreditation by the Accreditation Council for Genetic Counseling (ACGC) and anticipate opening in fall of 2018
- Is a 22-month, 63 credit program which includes both academic and clinical training
- Most programs are small (5-15 students due to limitation of clinical rotations)
- We plan to accept 5 students/year
- Information is available at: http://precisionhealth.uahs.arizona.edu/education
- https://www.nsgc.org/

“A good decision is based on knowledge and not on numbers”

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