What is Genetic Counseling?

Shelly Galasinski
UNC - Greensboro
November 21, 2003
Learning Objectives

- Understand the diverse roles of the genetic counselor
- Understand the process of genetic counseling
- Identify situations in which genetic counseling is warranted
- Know where to learn more about genetic counseling
Overview

- Genetic counseling defined
- Areas of practice
- Case studies
  - Prenatal
  - Pediatric
  - Adult
- How to become a genetic counselor
Genetic Counseling Defined

- A process of explaining medical & scientific information about a genetic condition or birth defect to an individual or family
  - Review of family, medical and pregnancy history
  - Perform risk assessments
  - Discuss the inheritance of genetic conditions
  - Discuss diseases, their management, treatment & surveillance options
  - Review testing options
Genetic Counseling Defined

- Explore the impact of genetic disorders on both affected & unaffected family members
- Assist families & individuals as they adjust to the diagnosis
- Use non-directiveness to facilitate decision-making
- Serve as a patient advocate
Areas of Practice

- Prenatal Genetics
- Pediatric Genetics
- Adult Genetics
- Cancer Genetics
- Metabolism Genetics
- Disease Research
- Diagnostic Laboratory
- Support Groups
- Public Health
- Government
- Unlimited opportunities
Prenatal Genetic Counseling

- AMA
- Abnormal screening test
- Family history
- Abnormal ultrasound
- Teratogens
- Ethnicity
- Infertility and/or multiple miscarriages
- Preimplantation diagnosis
A 28-year-old G5P1 Caucasian female is referred for GC regarding a previous child with a posterior urethral valve (PUV)

The patient is currently 12 weeks pregnant
Case Prep

- What is a PUV?
  - Etiology (syndrome, teratogen, isolated?)
  - Recurrence risk
- Recurrent miscarriages
  - Genetic etiologies
- Testing Options
Case Prep

- Anticipated psychosocial issues
  - Lost previous child with PUV at 18 weeks
  - Previous losses

- Population screening
  - Cystic Fibrosis carrier screening
During the Appointment

- Contracting
  - Identify patient concerns and expectations
  - Explain to patient what will happen during appt.

- Patient concerns
  - Recurrence and cause of PUV
  - Previous miscarriages
During the Appointment

- Psychosocial counseling
  - How pt was coping with previous losses
  - Pt reported that mother was dying from cancer
  - Changing jobs

- Pedigree
  - 3 generation family history
  - Birth defects, mental retardation, miscarriages, infertility, stillbirths, ancestry, consanguinity, GU problems
During the Appointment

- **Explanation of PUV**
  - Generally sporadic, low recurrence risk
  - Multifactorial inheritance
  - Reassure patient that she did not cause this

- **Significant family history/risk assessment**
  - Husband's previous wife had 3 miscarriages
  - Mother with breast cancer at 39 y/aunt with breast cancer in 40’s
During the Appointment

- **Address miscarriages**
  - \( \sim 10\%-15\% \) of couples with recurrent miscarriages carry a chromosome rearrangement (translocation, inversion)

- **Parental chromosome studies**
  - Need for husband’s blood sample
  - No insurance

- **Pt reveals past drug use**
  - Concerned that this could cause chromosome change
  - Psychosocial counseling
During the Appointment

- Address family history of breast cancer
  - Make aware of genetic counseling
    - Risk assessment
    - Susceptibility testing for breast cancer gene mutations (BRCA 1 and 2)
    - Risk to other family members
    - Treatment and management
  - Recommendations
    - Early surveillance
    - Share family history with PCP
During the Appointment

- Testing options
  - U/S for PUV
  - Parental chromosome studies
  - Amniocentesis

- Conclusion
  - Summarize information, plan of action
  - Further questions
  - Resources
Follow-up

- Patient letter
- Chart note for medical record
- Reporting of test results
- Additional counseling
Additional Counseling Issue

- Carrier screening for CF
  - Autosomal recessive condition
    - Lungs and digestive tract
  - 1/25-1/30 carrier frequency in Caucasian pop.
  - Risk Calculations
    - Carrier status unknown
      - \((1/25)(1/2)(1/25)(1/2) = 1/2500\)
    - 1 parent carrier
      - \((1)(1/2)(1/25)(1/2) = 1/100\)
Pediatric Genetic Counseling

- Diagnostic evaluation
- Developmental delay
- Mental retardation
- Dysmorphology
- Family history
- Health care maintenance
- Treatment
Case Report #2

- Patient is a 34 month old boy with speech delay, developmental delay, and question of autism
- Seen in genetics clinic with his mom for evaluation
Case Prep

- Obtain and review patient’s medical records
  - Pediatrician
  - Neurology
  - Developmental evaluation
- Research any possible disorders
During the Appointment

- Contracting
- Elicit
  - Medical history
  - Pregnancy history
  - Family history
- Evaluation by Medical Geneticist
  - Dysmorphology/Physical exam
During the Appointment

- Geneticist recommends
  - Chromosome studies
  - Fragile X syndrome testing

- Explain chromosomes and testing to mom
  - What test involves – blood sample
  - What test is looking for
  - Turnaround time
  - Return for results
Follow-up/Results

- Patient letter, chromosome studies
- Testing determines that patient has Fragile X syndrome
- Prepare for results disclosure
  - Explanation of results
  - Natural history of Fragile X
  - Inheritance
  - Support resources
Fragile X Syndrome

- X-linked inheritance
- Affects 1/4000 boys and girls
- Mental retardation
- Autistic like behaviors
  - Speech
  - Abnormal hand movements
  - Sensory integration disorder
Fragile X Syndrome - Phenotype

- Large head circumference
- Large ears
- Long face
- Broad forehead
- Flat feet
- Hyperextensible joints
Review - X-linked Inheritance

- The incidence of the trait is much higher in males than females
  - Carrier females are usually unaffected but some may express the condition with variable severity
- All daughters of affected males are carriers
- Male to male transmission never occurs
Classic Pedigree for X-Linked
Fragile X - Inheritance

- X-linked CGG triplet repeat disorder
  - Multigenerational mutational process
  - Unstable CGG expands over time
- Classification
  - 29 repeats is average
  - 52-200 repeats – premutation “carrier”
  - 200 or greater is full mutation
  - Methylation status
Results - Genetic Counseling

- Results disclosure
- Discussion of Fragile X and inheritance
- Identification of at risk relatives
  - Facilitate communication process
  - How does this information affect the family
- Identification of professional resources
- Provision of written information
Adult Genetic Counseling

- Diagnostic evaluation
- Family history/reproductive questions
- Management
- Treatment
- Susceptibility testing
- Presymptomatic testing
Case Report #3

- John Smith (25 years old) is referred for genetic counseling because his mother (47 years old) was recently diagnosed with Huntington’s disease
- John is interested in testing for HD
- John is recently engaged
Huntington Disease

- Autosomal dominant neurodegenerative disorder that affects the basal ganglia and the central nervous system
- Progressive dementia and involuntary movements
  - Involuntary movements, drunken gait
  - Depression, irritability, aggressive outbursts, social withdrawal, short term memory loss
- Average age of onset (30 to 50 years)
  - Death 15-20 years after onset
Genetic Counseling

- Review feature’s of Huntington’s disease
- Review genetics of HD
  - Autosomal Dominant inheritance
    - Pt at 50% risk of have HD mutation
  - Anticipation
  - Complete penetrance
- No treatment
Presymptomatic testing

- Very accurate
  - Positive DNA test cannot determine the age of onset

- Testing protocol (must be at least 18 years of age)
  - Medical Genetics (3)
  - Neurology (1)
  - Psychiatry (1)
Risks and Benefits

Benefits
- Reduced uncertainty
- Decreased anxiety
- Appropriate diagnosis/education
- Informed family planning
- Informed life planning

Risks
- Increased anxiety
- No medical benefit
- Altered self image
- Discrimination
- Implications for other family members
Additional Issues

- Sharing information with fiancé
  - Relationship
  - Reproductive issues
  - Long term care
- Dealing with mom’s disease
Becoming a Genetic Counselor

- Two year Master’s Degree in Genetic Counseling
  - 24 programs in US (average 6-8 students per class)
  - Didactic courses in molecular, cyto- and clinical genetics, counseling, and risk calculation
  - Clinical Rotations in various subspecialties
  - Laboratory Exposure/ Rotation

- Various Educational Backgrounds
  - Most have majors in biology, or psychology
  - Some may have nursing, research or teaching backgrounds
Becoming a Genetic Counselor

Certification Exam
- American Board of Medical Genetics (ABMG) and the American Board of Genetic Counseling (ABGC)
- Currently offered every 3 years
- Must be board eligible
  - Graduate from ABGC accredited program
  - ABGC approved logbook of clinical cases
- General Exam (ABMG)
- Genetic Counseling subspecialty exam (ABGC)

Continuing Education Credits
National Society of Genetic Counselors (NSGC)
http://www.nsgc.org