Genetic Counseling: A Profession in the Making

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Definition

...the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.

• Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.

• Education about inheritance, testing, management, prevention, resources and research.

• Counseling to promote informed choices and adaptation to the risk or condition.
Origins

- **1971** – First genetic counseling training program started
- **1979** – National Society of Genetic Counselors (NSGC) formed
- **1981** – First Annual Education Conference held
- **1996** – Professional competencies written and published

www.nsgc.org
Training

• 2 to 3 year master’s degree
  - coursework
  - clinical rotations
  - thesis project
• 30 programs in North America
• Logbook and certification exam
• Licensure (UT, IL, CA)
• Doctorate (Ph.D. or Gc.D.)?
Genetic Counseling Subspecialties

- Prenatal – risk to pregnancy
- Pediatric – diagnosis and recurrence
- Cancer – risk to individual and family
- Research – understanding genetic disease
- Education Outreach – informing the community
Employment

- University Medical Centers
- Private hospitals
- Research grants
- Industry laboratories
- State agencies
- Support organizations

Check out the Professional Status Survey in 2006!
The Genetic Counseling Process

- Contracting
- Genetics education
- Family and medical history
- Risk assessment
- Physical exam
- Testing options
- Diagnosis and natural history
- Psychosocial counseling
- Follow-up
Genetics Education

- DNA, chromosomes, genes
- Deletion, translocation, etc.
- Nondisjunction
- Inheritance patterns
- De novo vs. familial
- Penetrance and variability
Family and medical history

• Three generation pedigree
• Birth defects, mental retardation, genetic conditions
• Personal and family medical history
• Pregnancy history
• Ethnic background
Risk Assessment

- Age
- Maternal serum screening results
- Ultrasound findings
- Physical exam
- Family history
- Population incidence
- Empirical data
Testing Options

Prenatal:
- Screening vs. diagnostic

Pediatric:
- Test availability
- Detection rate
- Cost
- Turnaround time

Adult:
- Predictive vs. diagnostic
- Insurance coverage
- Potential for discrimination
Psychosocial Counseling

- Psychosocial Assessment
- Respond calmly to displays of emotion
- Offer empathy and support
- Engage in appropriate follow-up

Know when to refer...
Follow-up

- Write summary letter to doctor and/or patient
- Arrange additional testing and appointments
- Call doctor and/or patient with test results
- Be available for future questions
- Expose patients to available support resources
Nondirectiveness

- Basic tenet of genetic counseling
- Provide nonbiased information
- Support patient’s ability to make personal decisions
- Avoid imposing personal beliefs or opinions on patient
- Advocate for patient’s right to direct their own care
Case Examples
Case 1 – Multiple Congenital Anomalies

- 16 y.o. patient, first pregnancy
- Referred for ultrasound due to anomalies seen on outside scan
- Accompanied by her mother and maternal grandmother
- **Findings** – cleft lip and palate, pyelectasis
- Psychosocial assessment
Case 1

Before Surgery

Complete unilateral CL

After Surgery

http://www.emedicine.com/ent/topic135.htm
Case 1

Bilateral renal pyelectasis
Case 2 – Chromosome Conditions

- 25 y.o. patient, first pregnancy
- Abnormal maternal serum screen
- 1/60 risk for Down syndrome
- Normal ultrasound
- Amniocentesis
- Findings – Trisomy 21
- Psychosocial Assessment
Case 2

Atrial septal defect (ASD)
Case 2

Preston

DOB: 10/28/05
Case 3 – Rare Anomaly

- 34 y.o. patient, first pregnancy
- Referred due to abnormal finding on outside scan
- Ultrasound identified identical twins, one grossly abnormal
- **Finding** – acardiac twin
- IUFD at 32 weeks
- Psychosocial Assessment
Case 3
My Story

- Took Genetics in Human Affairs, Fall ’00
- Decided to minor in Genetics
- Developed project investigating roles of genetic counselors
- Researched master’s programs and applied Fall ’02
- Graduated with a B.S. in Biological Sciences, Spring ’03
- Earned an M.S. in Genetic Counseling from UNCG, Spring ’05
- Began working at USC, June ’05
ANGEL Study

- Anencephaly: Genetic and Environmental Links
- **Purpose:** Investigate etiology of anencephaly
- **Goal:** Enroll 500 cases nationwide
- **My role:** Create network of genetic counseling professionals to identify, approach, and enroll patients in the study
- **Method:** One-on-one discussions, group presentations, national conferences and meetings, promotional materials
Questions?

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