



What Is Genetic Counseling?

“Helping individuals and families understand how genetics affects their health and lives”



What does the career involve?

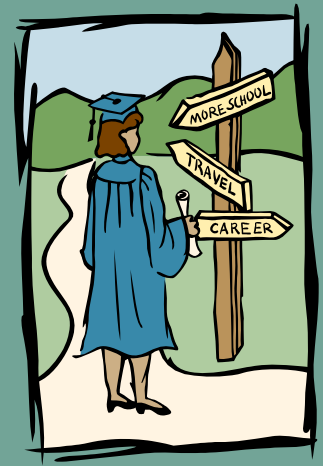
- Explore family histories to identify risks
- Reducing risks
- Discuss diseases
- Review testing options
- Help families with decision making
- Explore the impact of genetic condition
- Educate the community





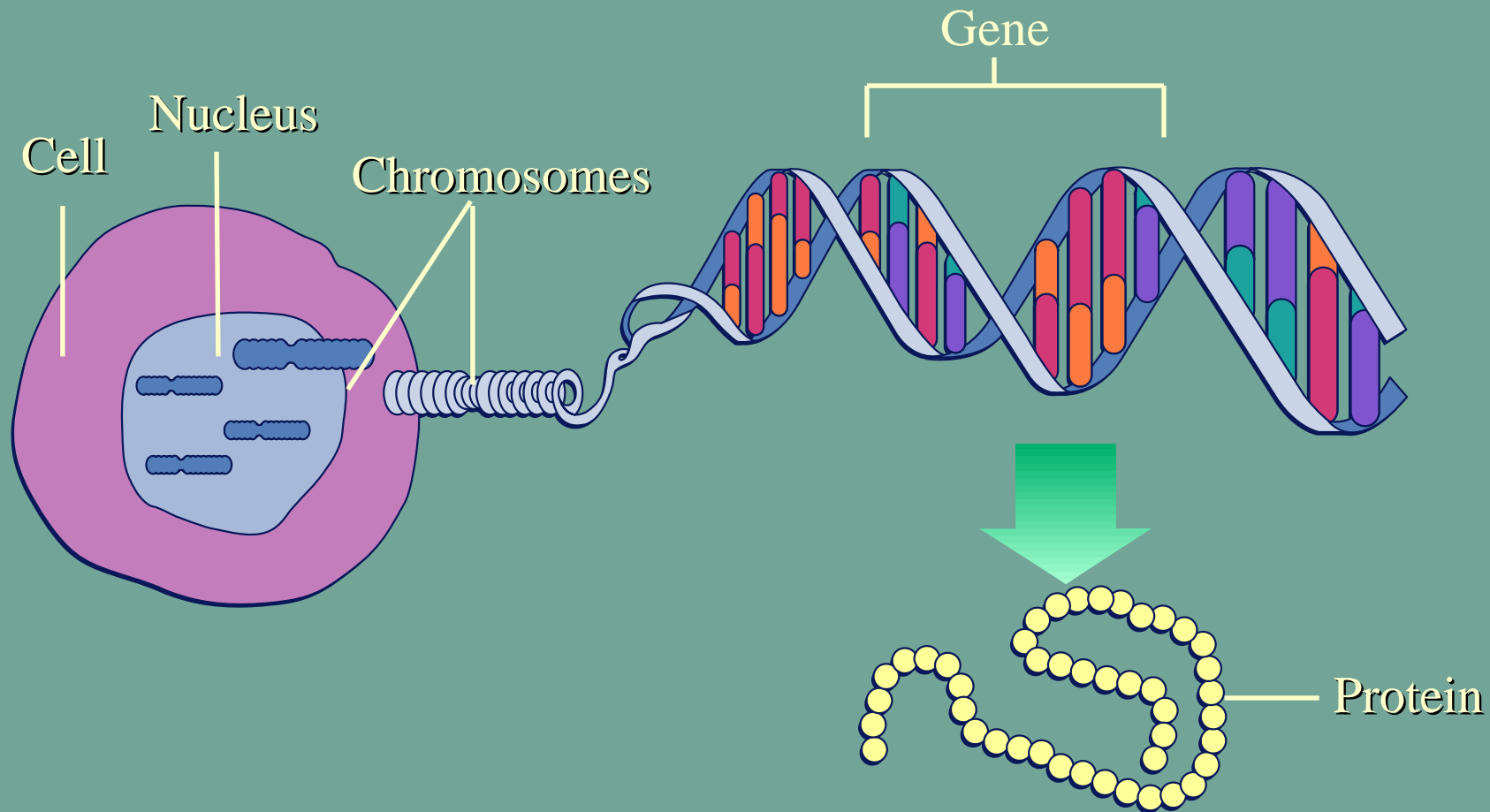
Career Options

- Clinical
 - Pediatric Clinic
 - Prenatal Clinic
 - Adult Specialty Clinic
- Public Health
- Education
- Research
- Consulting
- Industry
 - Pharmaceuticals

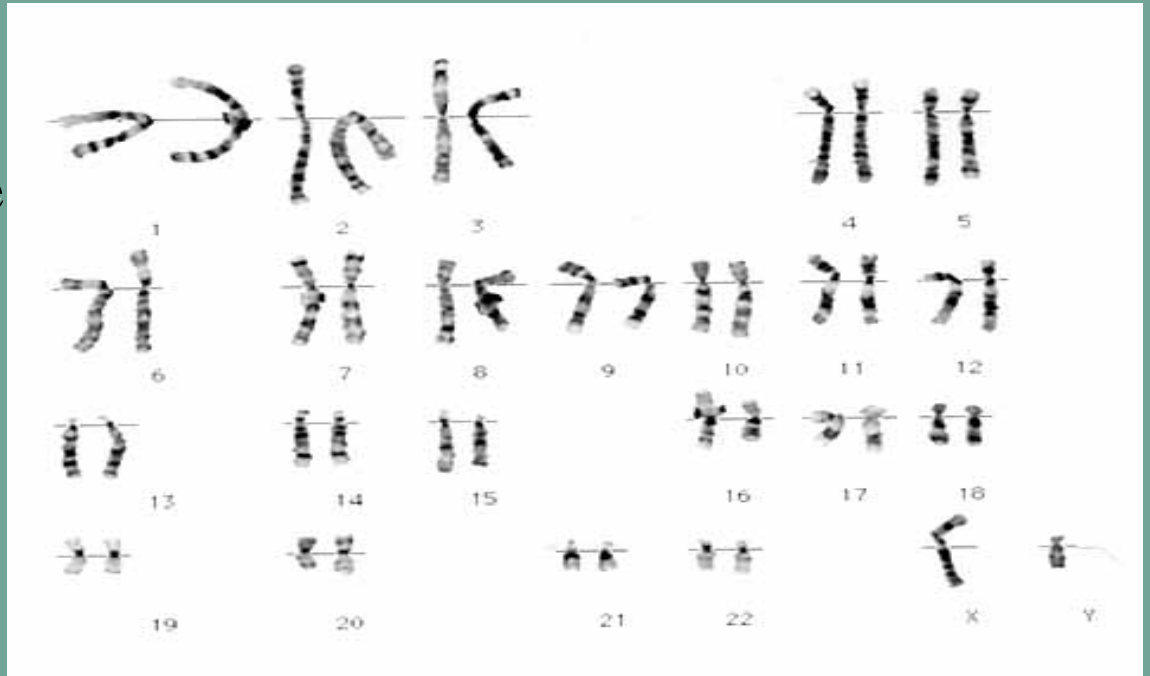
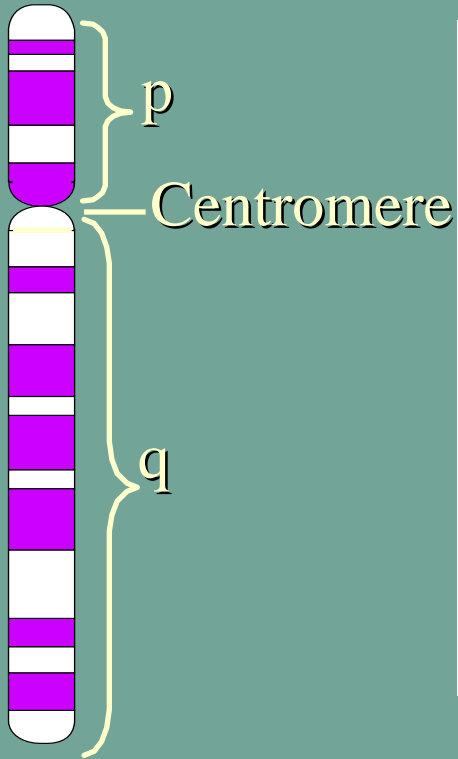




The Basics

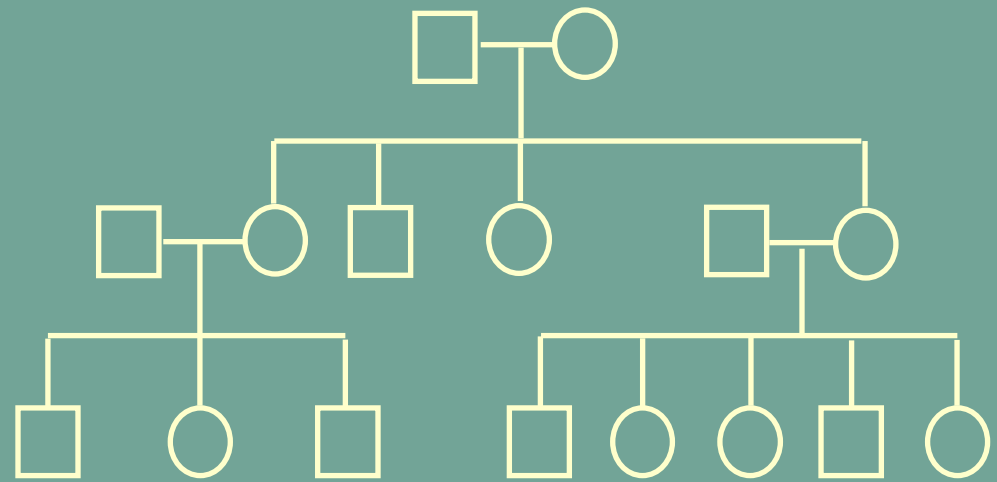


Karyotype



Family History - Pedigree

- Birth defects?
- Handicaps?
- Known genetic conditions?
- Multiple miscarriages?
- Sudden deaths?
- Cancer?
- Ethnicity?
- Consanguinity?



□ = Males

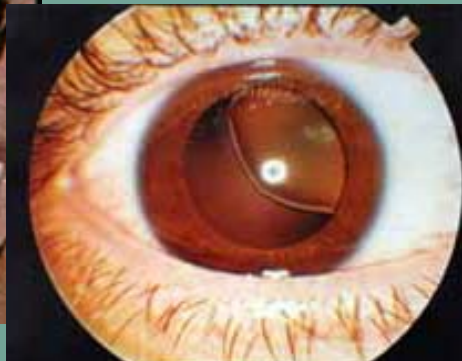
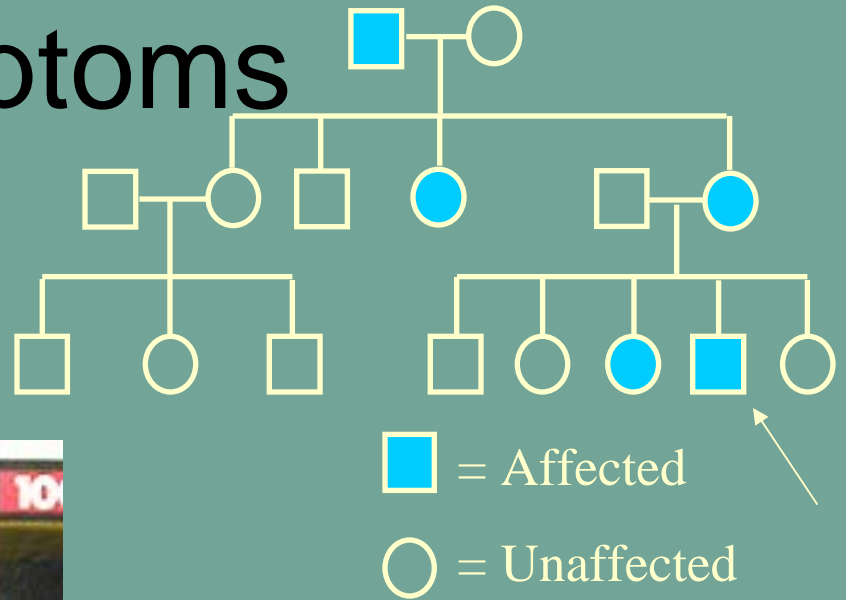
○ = Females



Pediatric Genetics Clinic

- Work as a team with a pediatrician trained in genetics, a nurse, and possibly a social worker or dietician.
- See a wide variety of patients.
- Work with the team to come up with a diagnosis based on symptoms and family history.

Case 1 Symptoms



Pectus excavatum



arachnodactyly

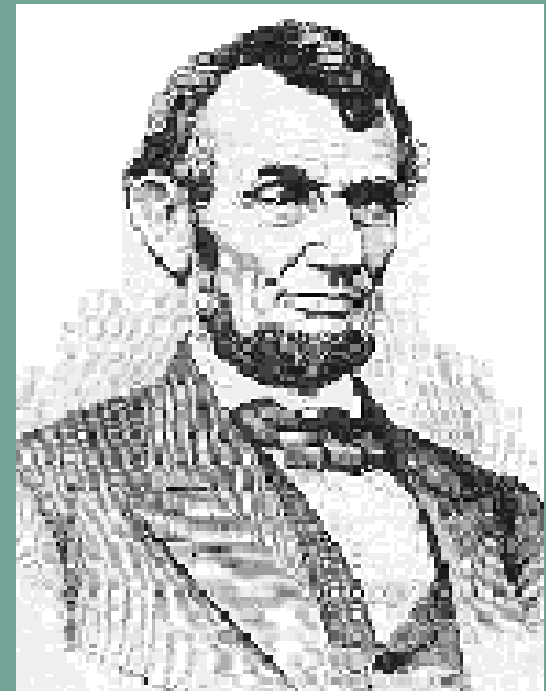


Dilation of aorta

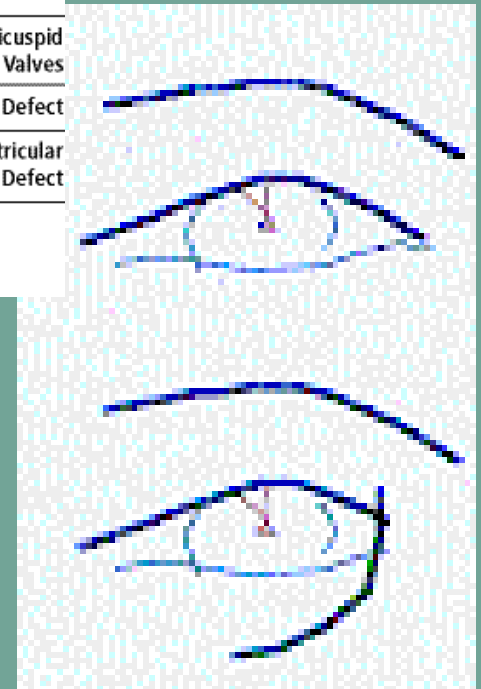
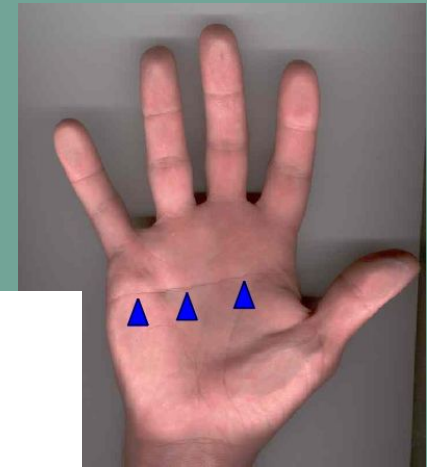
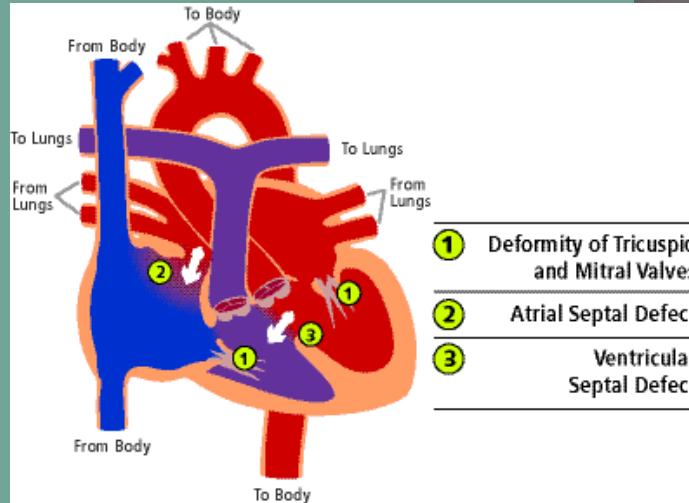


Marfan Syndrome

- Autosomal Dominant Inheritance
- Connective Tissue Disorder
- Gene Testing Available
- FBN1 (70-90% detection rate)
- 100% Penetrance
- Variable Expressivity
- ~1:10,000 Incidence
- Mutation is *de novo* in 25% of cases



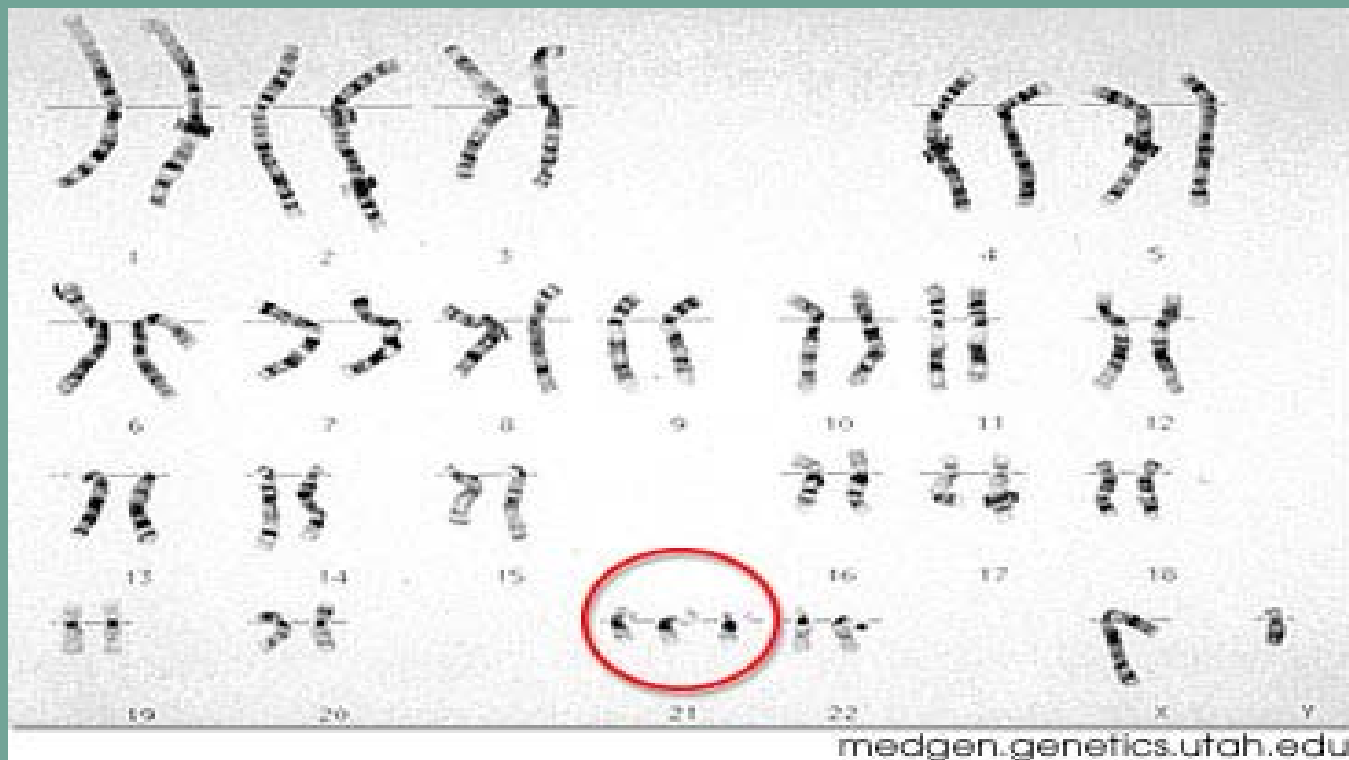
Case 2 Symptoms



- Developmental Delays
- Low muscle tone
- Flat facial profile
- Heart defects

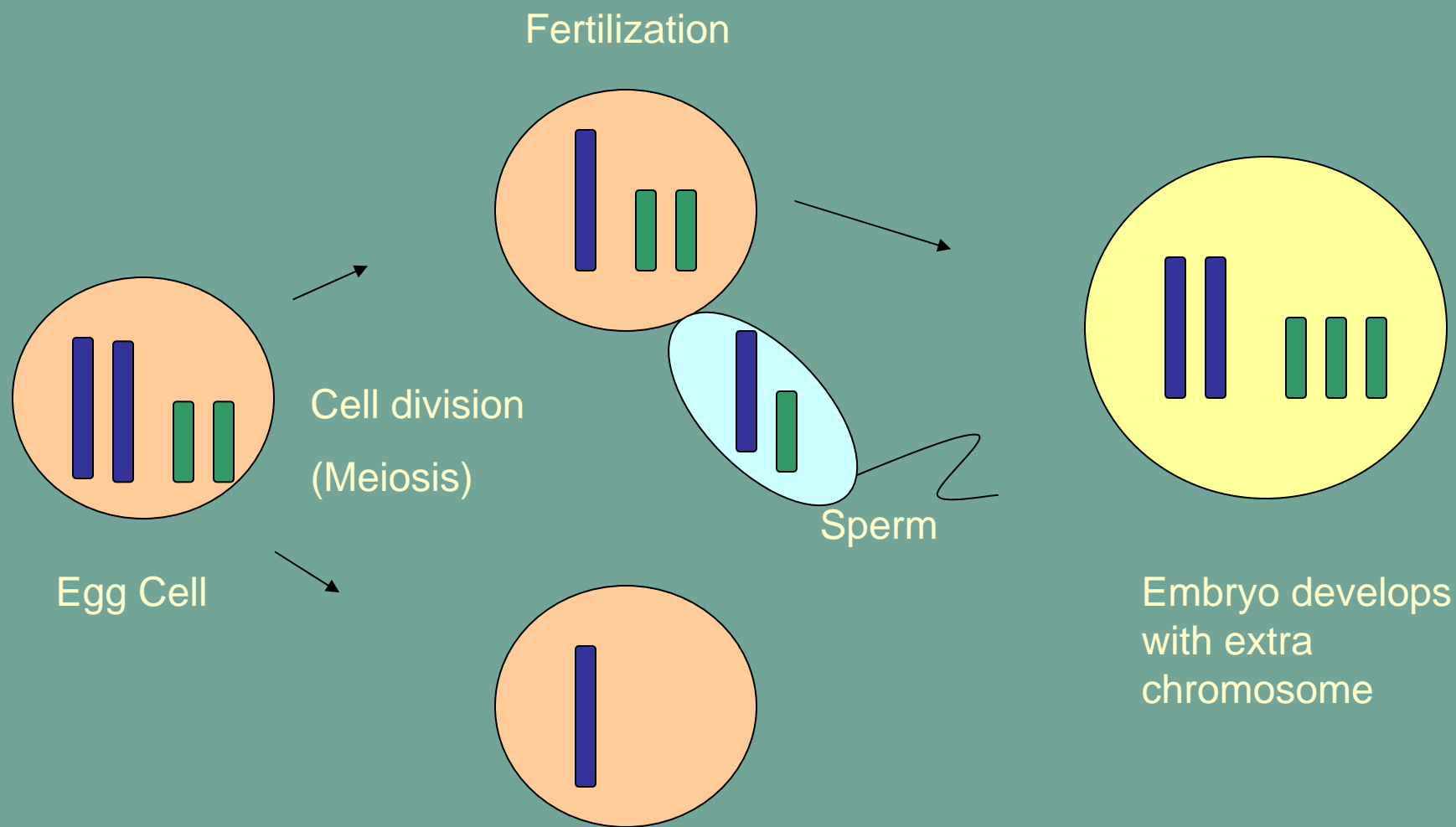


Trisomy 21: Down Syndrome





Nondisjunction

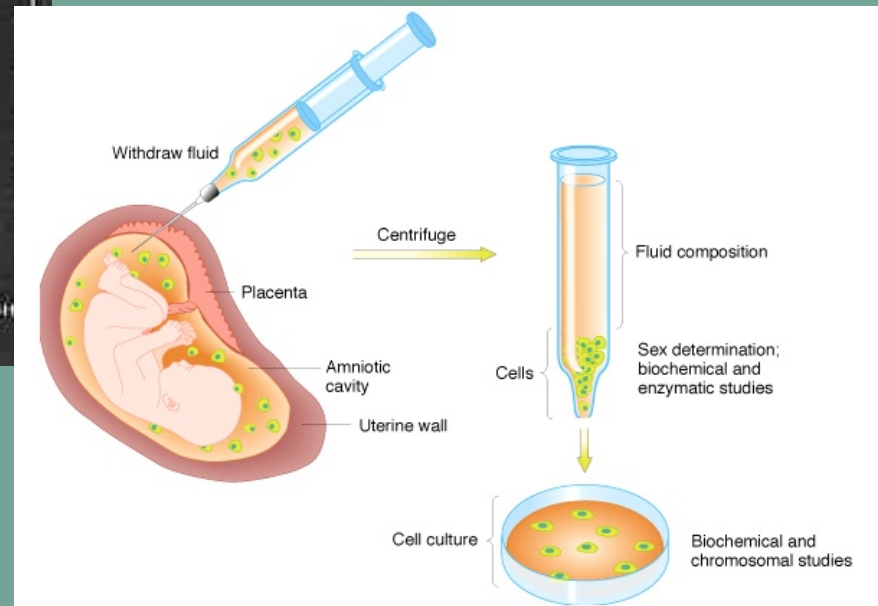
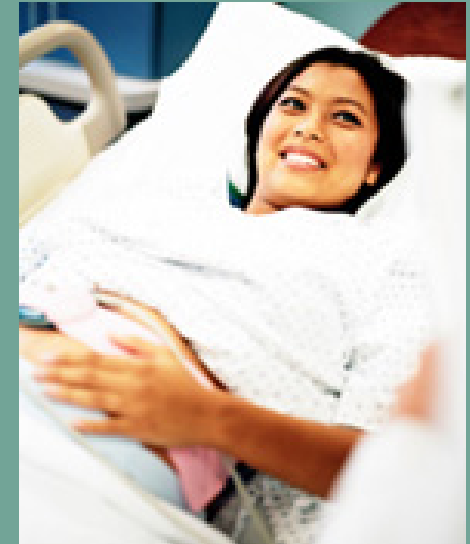




Prenatal Clinic

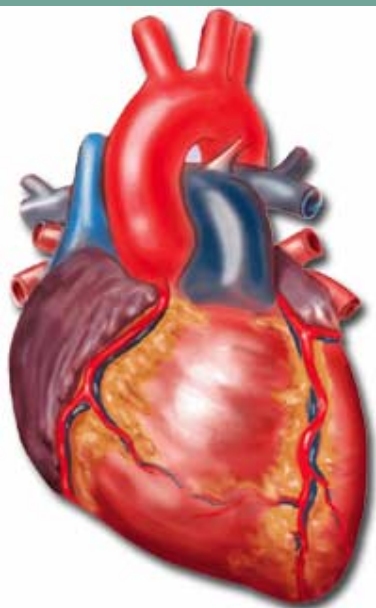
- Work as a team with an obstetrician, sonographer, and nurse.
- Most clinics are “high risk” pregnancy clinics that have much experience with difficult situations.
- See patients for advanced maternal age (AMA), teratogen exposures, abnormal ultrasound, or family history of a genetic condition.

Discuss Testing and Screening Options

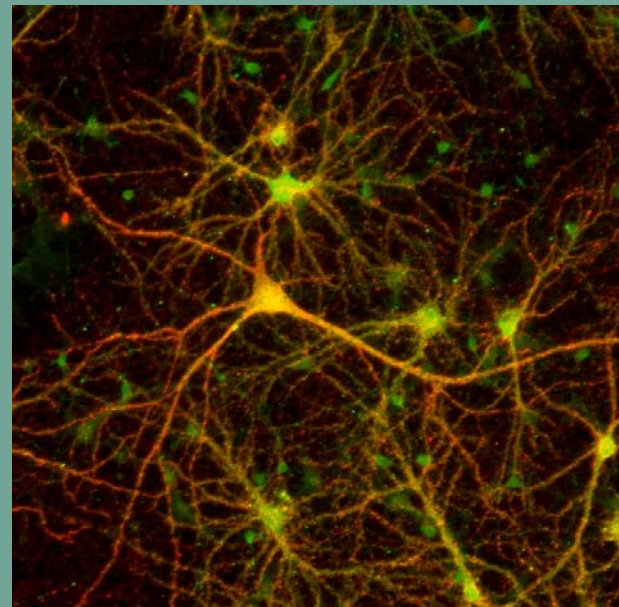




Adult Specialty Clinic



- Oncology
 - Breast Cancer
 - Colon Cancer
- Cardiology
 - Heart Disease
- Muscular Dystrophies
- Neurology
 - Huntington Disease
 - Alzheimer Disease



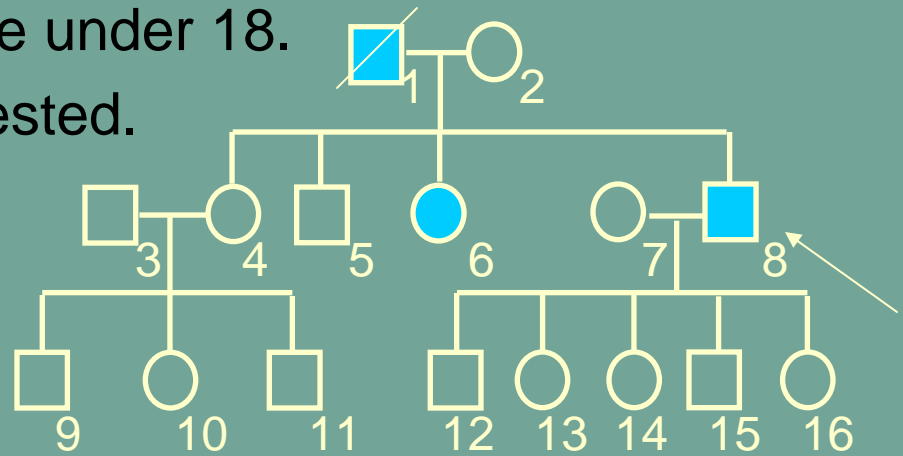


Huntington Disease

- Degenerative neurological disorder.
- Symptoms usually between 35 and 44 years old.
- Death 15-20 years after onset.
- Symptoms are both behavioral/emotional as well as physical.
- Involuntary movements, drunken gait.
- Depression, irritability, aggressive outbursts, social withdrawal, short term memory loss.
- No cure, some drugs are available to suppress symptoms.
- Autosomal dominant inheritance.

Case 3: An Ethical Dilemma

- A family that is affected with Huntington disease comes in for counseling.
- Pt #8 is recently diagnosed and is here to test his children. The oldest boy #12 is 19 years old, the other four children are under 18.
- Pt #4 has not been tested.
- Pt #4 does not want testing and does not want her adult children to know about this family history.



■ = Affected

○ = Unaffected



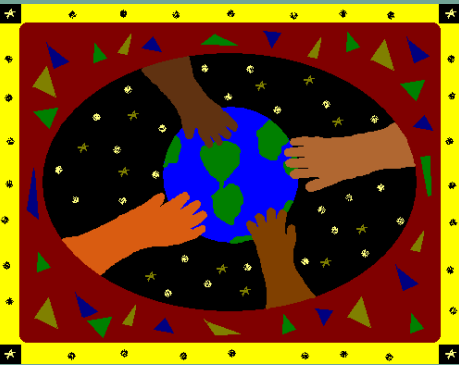
Genetic Counselor's Role

- Discuss the disease.
- Review testing options.
- Help the family in the decision making process.
- Explain inheritance.
- Find resources/support for the family.
- What are the benefits for testing?
- What are the disadvantages of testing?
- What ethical dilemmas are involved?
- How do you feel about Pt #4 not wanting to tell her children?

The Rewards of the Job

- An opportunity to work in science and have interaction with individual patients and their families.
- After college there is only two years of graduate training.
- The median salary for a genetic counselor was reported as \$71,100 in 2007.
- Named a 2008 US News & World Report “Best Career”
- About 90% of genetic counselors report satisfaction with
 - The intellectual challenges of the field
 - Learning opportunities
 - Patient contact/counseling
 - Opportunities for personal growth
 - Autonomy in their current position

(Based on the 2004 NSGC Professional Status Survey)



Prepare to be a genetic counselor

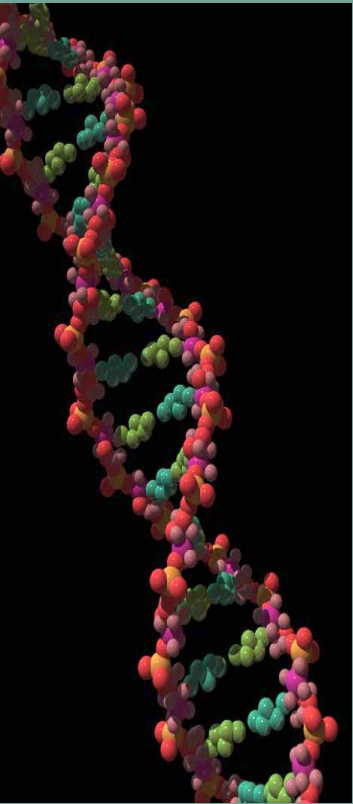


- Learn about the field
 - Read about:
 - Human genome research and ethical dilemmas
 - Treatments and tests for genetic conditions
 - Contact a genetic counselor
 - Talk about roles and responsibilities and the rewards of the job
 - Ask if you can observe in clinic
- Gain experience working with people
 - Volunteer—rape crisis, pregnancy counseling, Special Olympics
 - Be a teaching and/or research assistant
 - Take on leadership roles
- Obtain recommendations
 - From faculty and community supervisors



Find out more...

- Find a genetic counselor in your city or state
 - National Society of Genetic Counselors
 - www.nsgc.org
 - VoiceMail: 610-872-7608
 - Press 1: Career Information
 - Press 2: Job Connection
- Apply to Genetic Counseling Graduate Programs
 - <http://www.abgc.net/genetics/abgc/accred/tr-prog1.shtml>





Other Careers in Genetics

- Clinical Geneticist
 - Physicians specializing in the care of individuals and families with genetic conditions
- Medical Geneticist
 - Physicians specializing in genetic research and applications in clinical practice; often also practice as or work closely with clinical geneticists
- Genetic Laboratory Technician
 - Perform cytogenetic, molecular genetic, and other genetic testing in a laboratory setting
- Genetic Laboratory Research Assistant
 - Conduct research under the supervision of a medical geneticist in a research laboratory or a diagnostic or forensics laboratory