Genetic Counseling
Chapter 4

Viewing the Chromosomes for Genetic Counseling

- Helps determine the genotype of parents to determine if the unborn child will have a genetic disorder.
- The chances of bearing a child with trisomy increases with mothers age.
- Typically above 40 years chances increase to 1 in 100, while its 1 in 1200 for a mother in 20's.
- Done by 1) Amniocentesis or 2) Chorionic Villi Sampling

Amniocentesis

- Procedure involves obtaining a sample of amniotic fluid from the uterus
- Not performed until about the 12th week of pregnancy
- Involves risk of spontaneous abortion by 0.5%
- Fetal cells in the amniotic fluid are cultured to increase their number and chromosomes are analyzed.
- Biochemical tests on amniotic fluid can test for Alpha fetoprotein (AFP) – excess may indicate neural tube defects in fetus
Chorionic Villi Sampling (CVS)

- Involves obtaining sample of Chorionic Villi (Fetal tissue that forms part of placenta)
- Culture of cells not required since tissue is sampled.
- But biochemical tests cannot be performed.
- Results are obtained quicker
- But risk of spontaneous abortions greater (0.8%)

Karyotyping

- Visual display of chromosomes arranged by size, shape and banding pattern.
- All 22 pairs of autosomes and one pair of sex chromosomes.
- Mitosis in cells is stopped at metaphase, and chromosomes are stained.
- Chromosomes are visualized under a microscope
- Figure in previous slide shows Karyotype of Normal individual and one of Downs syndrome (note 3, 21 chromosomes.
Analyzing the family history

- Since over 400 different chromosomal and biochemical tests can be performed
- Counselors need to determine what tests are warranted
- Usually done using a pedigree chart.
- Males designated by squares and females by circles
- Shading indicates affected individuals

Autosomal Dominant and Recessive Disorders

- In the figure – the child is affected, but neither parent is.
- This happens when the disorder is recessive and the parents are carriers (Aa)
- So if the family pedigree may suggest that the parents are carriers for an autosomal recessive disorder, then appropriate genetic test is suggested.
Autosomal Dominant Disorder

- Child is unaffected, but both parents are affected.
- Can happen through autosomal dominant disorder
- Parents are Aa
Sex-linked Disorders

- Carried on the X or Y chromosome
- X-linked Recessive Disorders:
  - Sons inherit the X-linked recessive allele from mother, since fathers gave the Y chromosome.
  - More males have trait because recessive alleles on the X chromosome are always expressed in males, as the Y chromosome lacks the allele for the disorder.
  - Females who have the condition – inherit an allele from BOTH father and mother.
  - All sons of such a female will have the condition
  - Next slide shows how color blindness is an X-linked recessive disorder

Y-linked Genetic Disorders

- Few genetic disorders are carried on the Y chromosome.
- Best known – determination of gender during development.
- How to diagnose Y-linked conditions?
- Present only in males – passed directly from fathers to sons.
Types of Genetic Testing

• Two types of genetic testing
  – Biochemical testing
  – DNA testing

Testing for a Protein

• Babies with Tay-Sachs disease
• Lack enzyme called Hexosaminidase A (Hex A)
• Test on cells can reveal if the individual is homozygous normal, carrier or has disease.
• Biochemical tests are not as definitive as DNA tests.
Testing DNA

- Two types
  - Using genetic marker
  - Using DNA probe
- Individuals with sickle-cell or Huntington disease, have abnormal sequence of bases on a chromosome.
- Difference is a Genetic Marker
- Restriction enzymes are used to cleave DNA at certain base sequences.
- Differences in fragments reveal who has the mutation or not

![Diagram showing normal and genetic disorder fragmentation patterns of DNA.](image)

DNA Probes

- Single stranded DNA that binds to a complementary piece of DNA – bind to a genetic mutation.
- DNA Chips
  - Glass square that has several rows of DNA probes
  - DNA sample is cut into small pieces using restriction enzymes, tagged with fluorescent dye and converted into single strands.
  - Fragments that bear the mutation will bind to the probes.
Testing the Egg and Embryo

- In vitro fertilization (IVF), eggs are collected from mother
- And sperm from father
- Fertilization takes place in lab glassware
- Allows for the egg or embryo to be tested for any genetic disorder
Testing the Egg

• Meiosis results in an egg and two polar bodies
• Polar bodies have a haploid number of chromosomes.
• When a woman is heterozygous for a recessive genetic disorder, half the polar bodies will receive the mutated allele and the egg will receive the normal allele.
• So if the polar body tests positive for the mutated allele, then the egg will have a normal allele.
• So only normal eggs are used for IVF.

Testing the Embryo

• Embryo – for first two months of development
• Fetus – from second to ninth month
• After fertilization the zygote begins to divide – when the embryo is has six to eight cells – one cell is removed and tested for genetic disorders.
• Only embryos that test negative for genetic disorders are placed in the uterus of the mother and allowed to continue to term