Midterm reminder April 13
Covers lectures 10-18 (this includes Professor Roelink’s April 6 lecture)

Office hours this week:
Tu & Th 2-4 PM
No office hours Tu, April 7
Garriga midterm review session
April 12, 5-7 PM 100 GPB

Preimplantation genetic diagnosis (PGD), genetic counseling and eugenics
Reading: pp 200-202, 316

Outline
Preimplantation Genetic Diagnosis (PGD)
- in vitro fertilization (IVF)
- blastomere isolation
- Fluorescence in situ hybridization (FISH)
  and Polymerase Chain Reaction (PCR)

Genetic counseling

Eugenics

But first, in vitro fertilization (IVF)

Who uses IVF?
Couples that are infertile
Couples at risk for having babies with chromosomal abnormalities and genetic diseases

Blastomere Isolation
After IVF, 1-2 cells, known as blastomeres, can be removed from the 8-cell embryo.
Preimplantation Genetic Diagnosis (PGD)

Two techniques used to test blastomeres:
1. Fluorescence in situ hybridization (FISH)
2. Polymerase chain reaction (PCR)

Fluorescence in situ hybridization

FISH of isolated blastomere
Color-chr.
Blue-18
Red-21
Green-13
Yellow-X
White-Y

Blastomere with trisomy 21

Polymerase Chain Reaction (PCR)

PCR and cutting with a restriction enzyme
Restriction enzyme - + - + - +
Outline
Preimplantation Genetic Diagnosis (PGD)

Genetic counseling
What do counselors do?
Who needs counseling?
Pedigrees

Eugenics

Genetic Counseling
Family history
Order genetic tests
Evaluate results
Help understand and reach decisions

Who needs genetic counseling?
- Standard prenatal test abnormal
- Abnormal karyotype
- If either parent already has a child with inherited disease
- If either parent has close relative with inherited disease
- Three or more miscarriages or babies that die in infancy
- If woman is 35 or older
- Couple in at risk ethnic or racial group

Generate a family tree or pedigree.
Define the disorder as:
Autosomal recessive
Autosomal dominant
X-linked recessive
X-linked dominant

Autosomal recessive
Disease is expressed in matings between carriers (heterozygotes).
Expressed by males and females with equal frequency
Most affected individuals have unaffected parents.
Increased frequency with inbreeding.

Autosomal dominant
Huntington’s Disease (HD)
Affected person has an affected parent
Expressed by males and females with equal frequency
No skipping of generations
X-linked recessive

Trait usually expressed only in males.

Sons of carrier females have a 50% chance of being affected.

Affected males do not have affected sons.

Trait skips generations.

X-linked dominant
Congenital Generalized Hypertrichosis

Daughters, but not sons, inherit the trait from their fathers.

Daughters and sons can inherit the trait from their mothers.

The trait is present in each generation or is lost.

Outline
Preimplantation Genetic Diagnosis (PGD)

Genetic counseling

Eugenics
Definition
History

What is Eugenics?

Idea based on breeding of plants and animals

Improving human traits by controlled breeding

Attempts to limit mental illness and other negative traits

History of Eugenics

Started in England by Sir Francis Galton

“the more suitable races or strains of blood a better chance of prevailing speedily over the less suitable.”

History of Eugenics

Charles Davenport started the Eugenics Records Office in the US

1910 published Eugenics: The science of Human Improvement though Better Breeding
History of Eugenics

1920s Fitter Families Contests at state fairs

1907 first sterilization law passed in Indiana to sterilize “unfit” individuals

1924 18 states have sterilization laws

1924 Carry Buck case in Virginia

1927 “Buck vs Bell” decision upheld by U.S. Supreme Court

"...It is better for all the world, if instead of waiting to execute degenerate offspring for crime, or to let them starve for their imbecility, society can prevent those who are manifestly unfit from continuing their kind…… Three generations of imbeciles are enough."

Oliver Wendell Holmes

By 1935, 30 states had eugenic sterilization laws

21,000 people had been sterilized

50% in California

In 1942, Supreme Court struck down law allowing forced sterilizations of criminals

Some sterilizations continued into the 1970s

2002 Virginia Governor apologizes for the Buck vs Bell case

"The eugenics movement was a shameful effort in which state government never should have been involved."

Mark Warner
History of Eugenics

Science behind eugenics discredited by 1930s

Eugenics records Office closed in 1939

In 1927, Rockefeller Foundation provides funds for Kaiser Wilhelm Institute of Anthropology, Human Genetics, and Eugenics in Berlin

In 1933, Hitler charged the medical profession with the task of implementing a national program of race hygiene. An act permitting sterilization of feebleminded, mentally ill, epileptics, and alcoholics was passed.

Within a year, more than 50,000 sterilizations were ordered.

1939 400,000 people had been sterilized.