

**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**

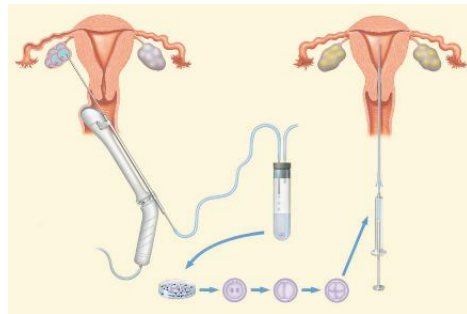
### **Preimplantation Genetic Diagnosis (PGD)**

**-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)**

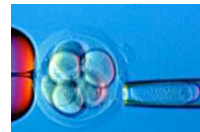


## **Who uses IVF?**

**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.**

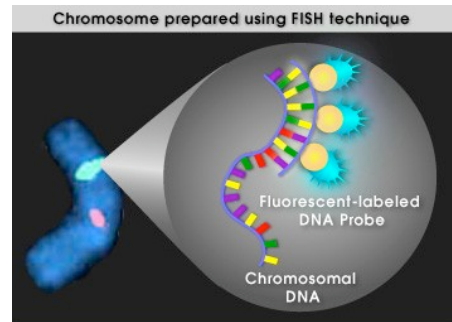
## Preimplantation Genetic Diagnosis (PGD)



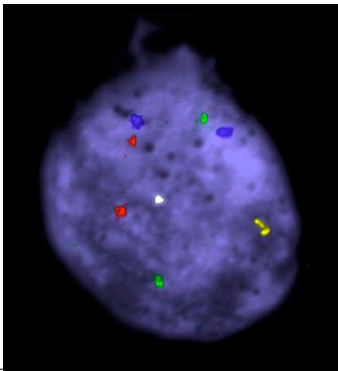
Two techniques used to test blastomeres:

1. Fluorescence in situ hybridization (FISH)
2. Polymerase chain reaction (PCR)

## Fluorescence in situ hybridization

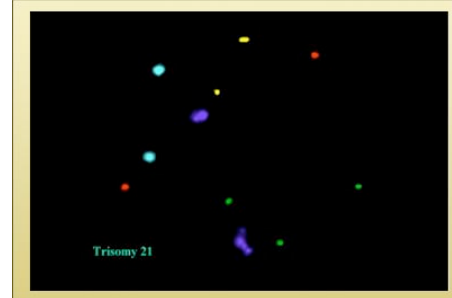


## FISH of isolated blastomere

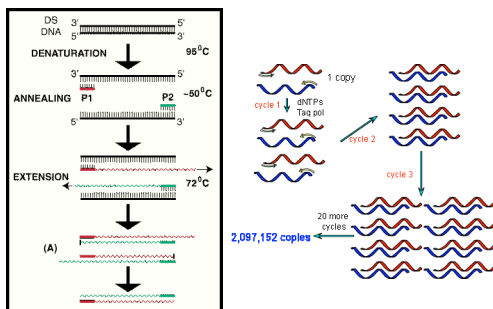


Color-chr.  
**Blue-18**  
**Red-21**  
**Green-13**  
**Yellow-X**  
**White-Y**

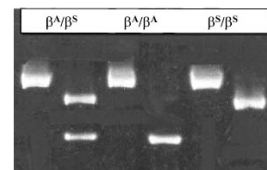
## blastomere with trisomy 21



## 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:

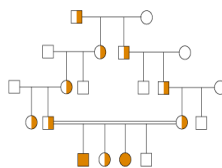
Autosomal recessive

Autosomal dominant

X-linked recessive

X-linked dominant

## Autosomal recessive



Disease is expressed in matings between carriers (heterozygotes).

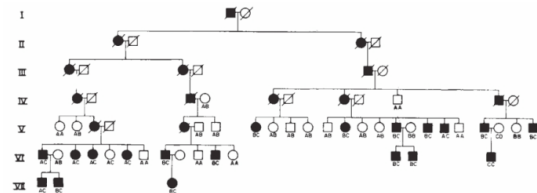
Expressed by males and females with equal frequency

Most affected individuals have unaffected parents.

Increased frequency with inbreeding.

## Autosomal dominant

### Huntington's Disease (HD)

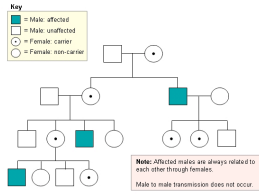


Affected person has an affected parent

Expressed by males and females with equal frequency

No skipping of generations

## X-linked recessive



Trait usually expressed only in males.

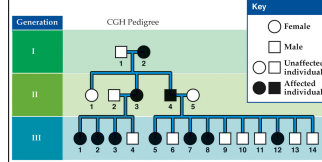
Sons of carrier females have a 50% chance of being affected.

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

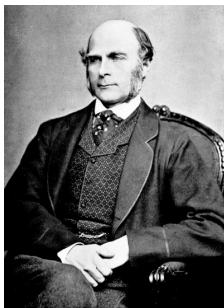
## What is Eugenics?

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



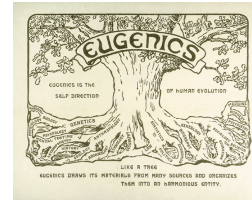
Started in England by Sir Francis Galton

“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 through 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

## History of Eugenics

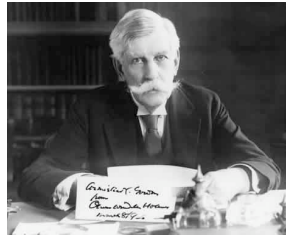
1924 Carry Buck case in Virginia



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



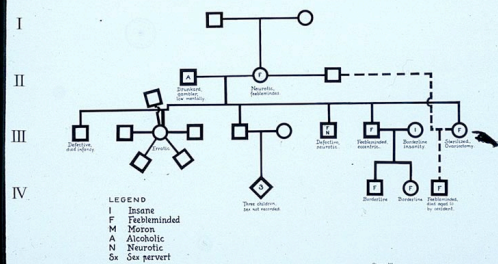
## History of Eugenics

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

THE NEAR BLOOD-KIN OF A FEEBLEMINDED WOMAN STERILIZED BY THE STATE OF CALIFORNIA



## History of Eugenics

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 feeble-minded, mentally ill, epileptics, and alcoholics was passed.

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

1939 400,000 people had been sterilized.