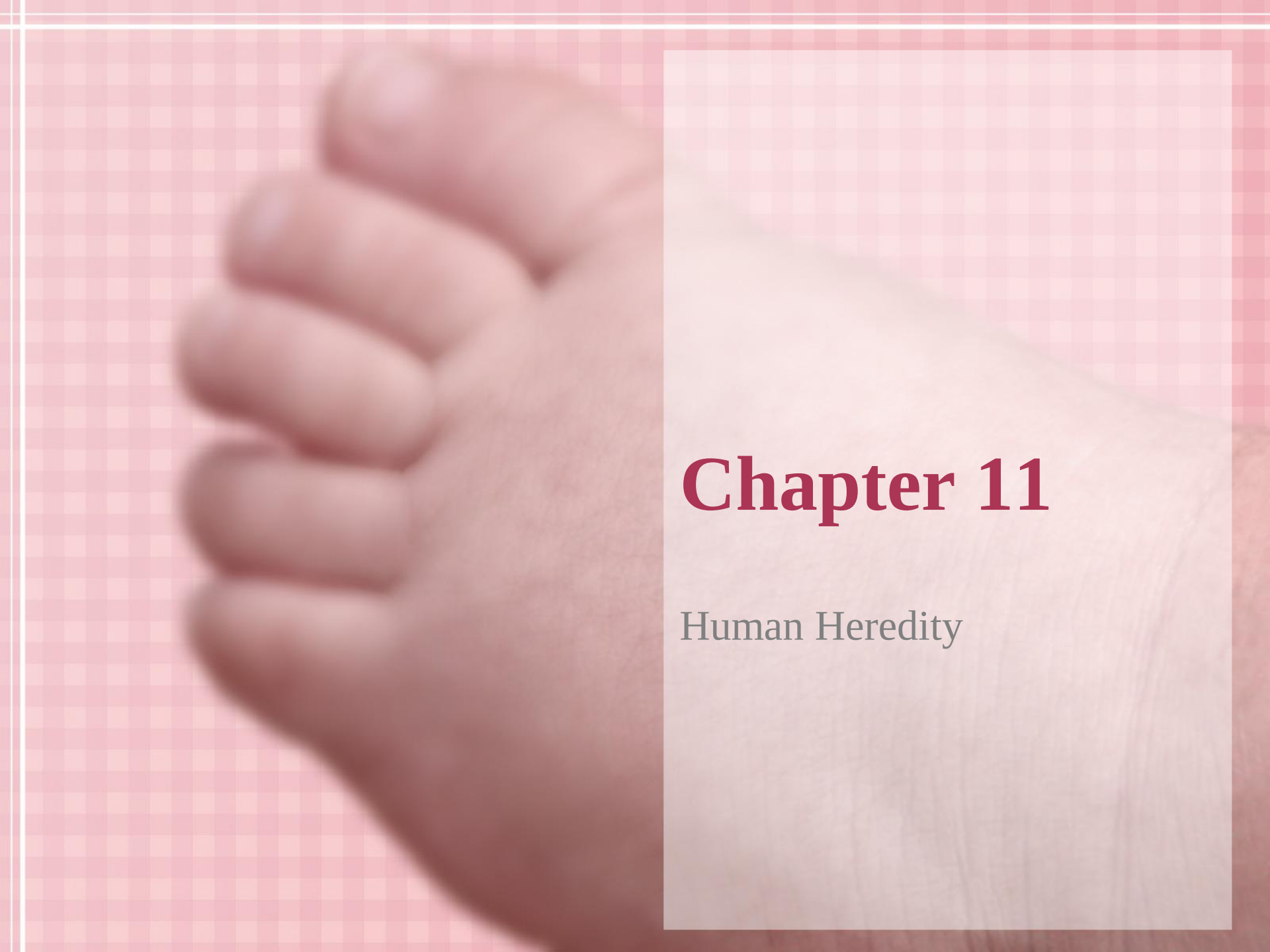


Chapter 11

Human Heredity

11-1 “It runs in the family”

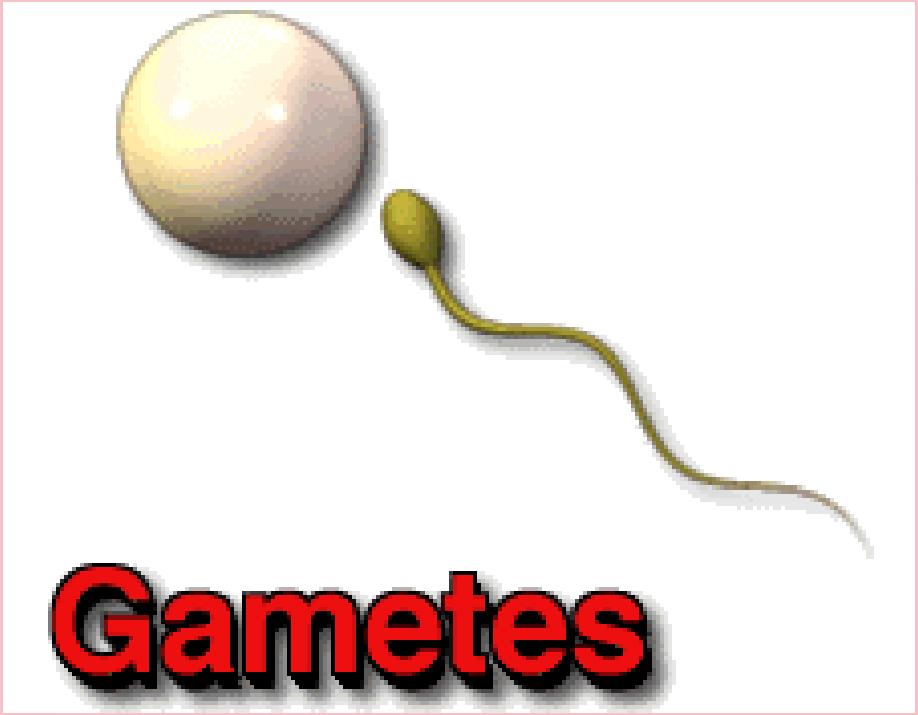
- Many characteristics of human children are genetically determined
- Many human traits are inherited by the action of dominant and recessive allele genes, although other traits are determined through more complicated gene interactions

The Human Organism – A Review

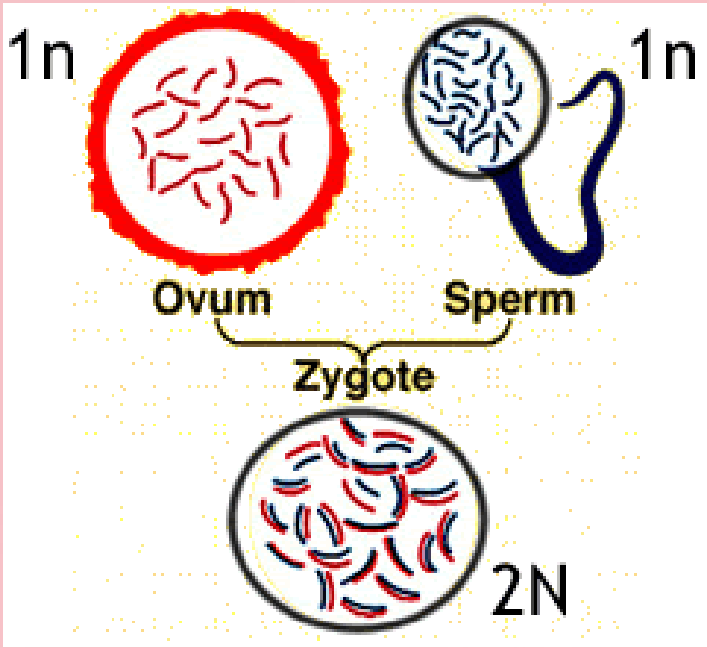
- Diploid cell – 2 sets of homologous chromosomes
  - 46 chromosomes
  - 23 pairs

- 6 billion nucleotide pairs
- 3,000 letters to a page – over 1 million pages

- Gametes – sperm or egg
  - Contain a single copy of a gene



- Zygote – fertilized egg – 46 chromosomes
  - Formed when sperm and egg unite



Human Traits

- Phenotype is only partly determined by the genotype
- Some traits are strongly influenced by environmental factors (non-genetic)
- Examples – exercise and nutrition

- It is important to consider the influence of the environment on the expression of some genes, it must be understood that environmental effects on gene expression are not inherited;

- Genes that are denied a proper environment in which to reach full expression in one generation can, in a proper environment, achieve full potential in a later generation

11-2 The Inheritance of Human Traits

- more than 3,000 human genes have been described

Human Blood Groups

- Multiple alleles – genes with more than two forms
- Example – ABO and Rh blood groups

* Remember – an organism can have two alleles only!

- Blood Type – determined by the presence or absence of certain things in the blood
- RBC (Red blood cell) – can carry two different antigens
- Antigens – molecules that can be recognized by the immune system

Genotypes and Phenotypes

- ii
- I^AI^A or I^Ai
- I^BI^B or I^Bi
- I^AI^B

- Type O
- Type A
- Type B
- Type AB

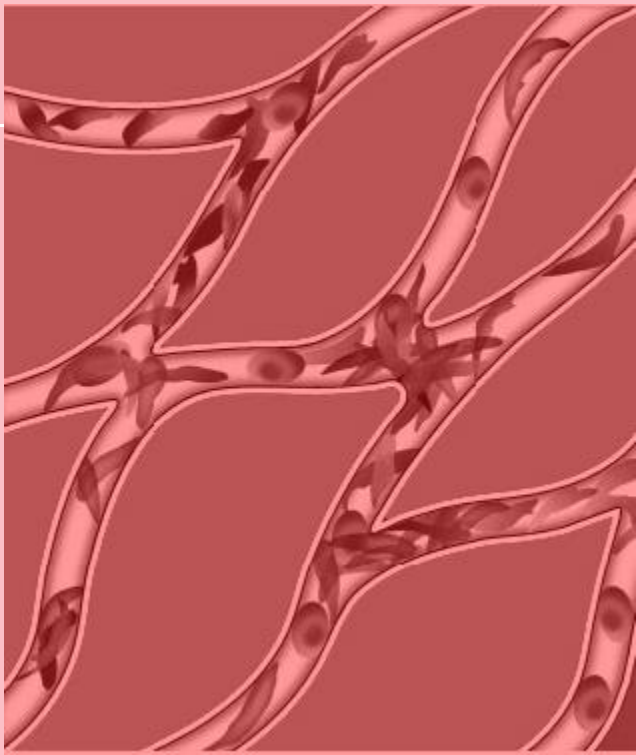
Rh Blood Groups

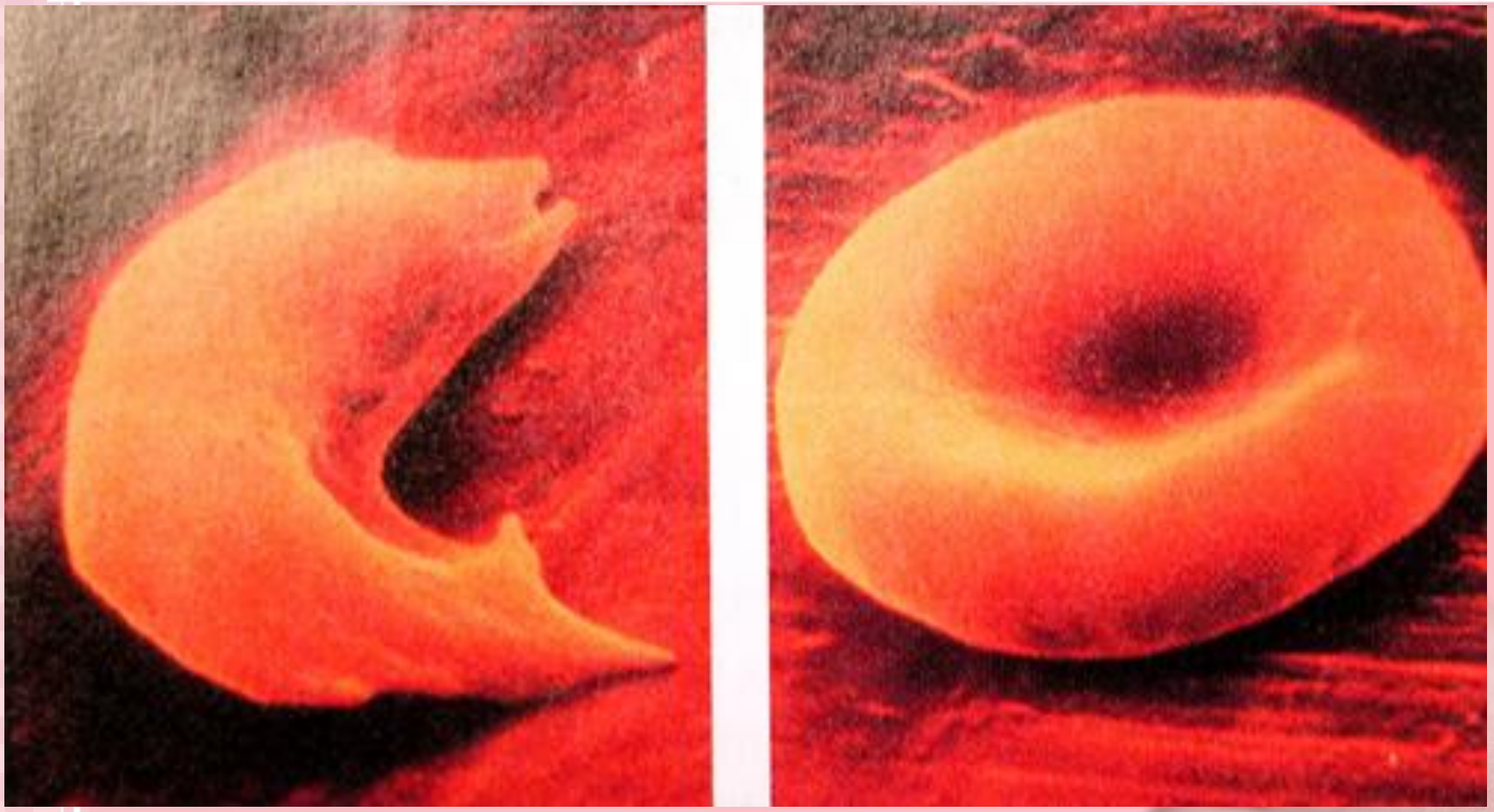
- Rh antigen also on RBC

- Rh^+ - have antigen (Dominant)
- Rh^- - no antigen (Recessive)

Sickle Cell Anemia

- Caused by a change in one of the polypeptides found in hemoglobin (carries oxygen in RBC)
- One nucleotide difference
- Codominant inheritance pattern – H^AH^S
- H^A - normal allele
- H^S – Sickle Cell allele





- Sickle Cell Anemia – common in people of African ancestry and from tropical regions
- Carriers (heterozygous) of Sickle Cell trait ($H^AH^{S)}$ resistant to malaria

11-3 Sex-Linked Inheritance

- Genes on an X chromosome are inherited in a sex linked pattern

Sex-Linked Genetic Disorders

- Gene for the trait is on the X or Y
- X has many genes – Y has few
- Defects easy to spot – appear more in males

Colorblindness

- Recessive X linked disorder
- Cannot distinguish colors
- Dominant Gene – X^C
- Recessive Gene - X^c

Carrier

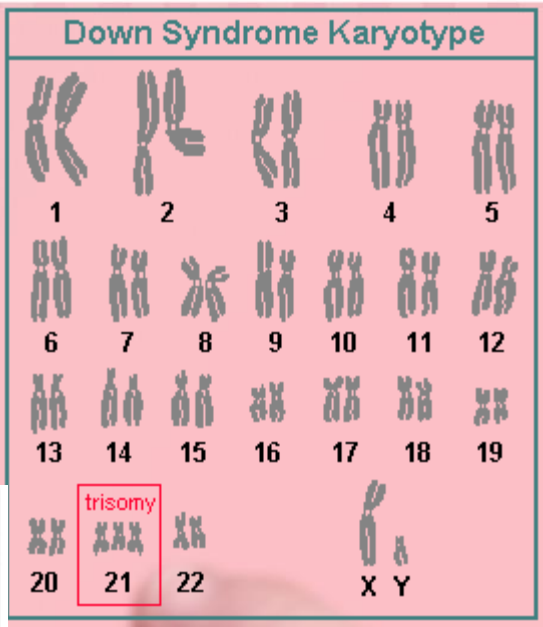
- A heterozygous female – has the gene but does not express it – can pass it on to her children

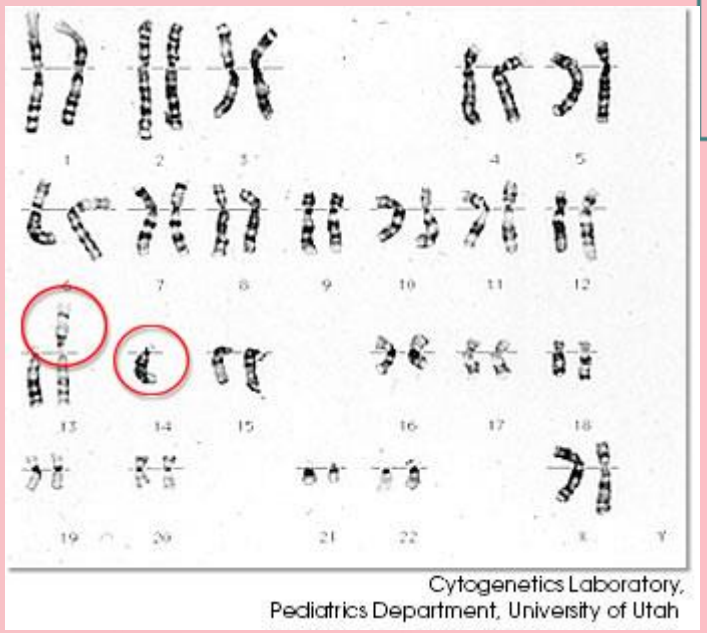
Hemophilia

- Recessive X linked disorder
- Blood does not clot
- X^H – good gene
- X^h – hemophilia gene

11-4 Diagnosis of Genetic Disorders

- Down's Syndrome – Trisomy 21
  - Extra copy of the 21st chromosome
- Can be detected by microscopic examinations of chromosomes
- karyotype




Cytogenetics Laboratory,
Pediatrics Department, University of Utah

Parental Diagnosis

- Amniocentesis – removes fluid from the sac around the baby
  - The fluid can be used to grow cells and make a karyotype
- Chorionic Villus Biopsy – cells are removed from the embryo
  - Faster than amnio.

- CVB and Amnio make it possible to detect chromosomal abnormalities
- Test for: biochemical abnormalities
  - » Presence of certain DNA sequences

- We can detect over 100 disorders
- Knowledge leads to choices and decisions

Genetic Engineering

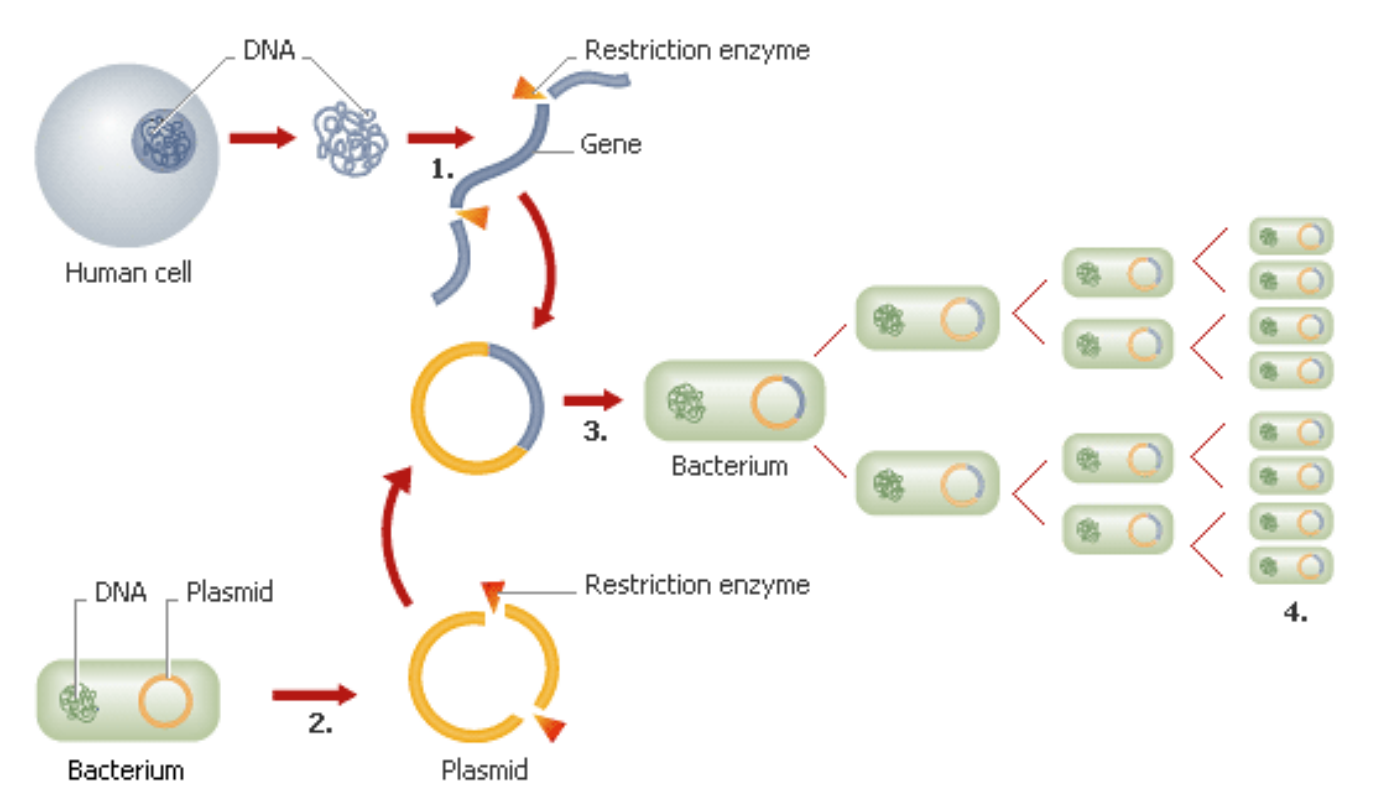
- Biologists can engineer a set of genetic changes directly into an organisms DNA

Restriction Enzymes

- Proteins that cut genes at specific DNA sequences

Engineering New Organisms

- Transgenic – organisms that contain foreign genes



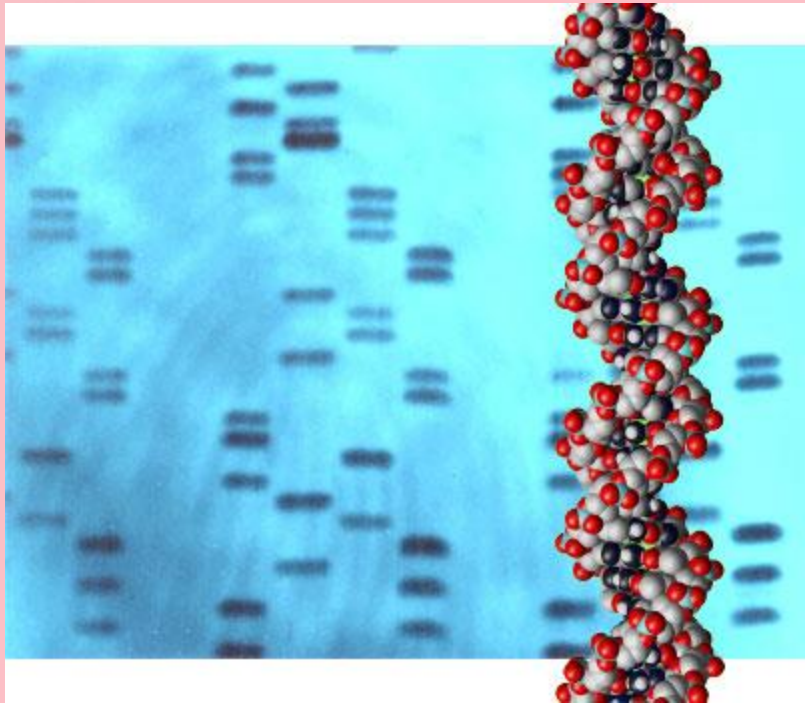
Transgenic Bacteria

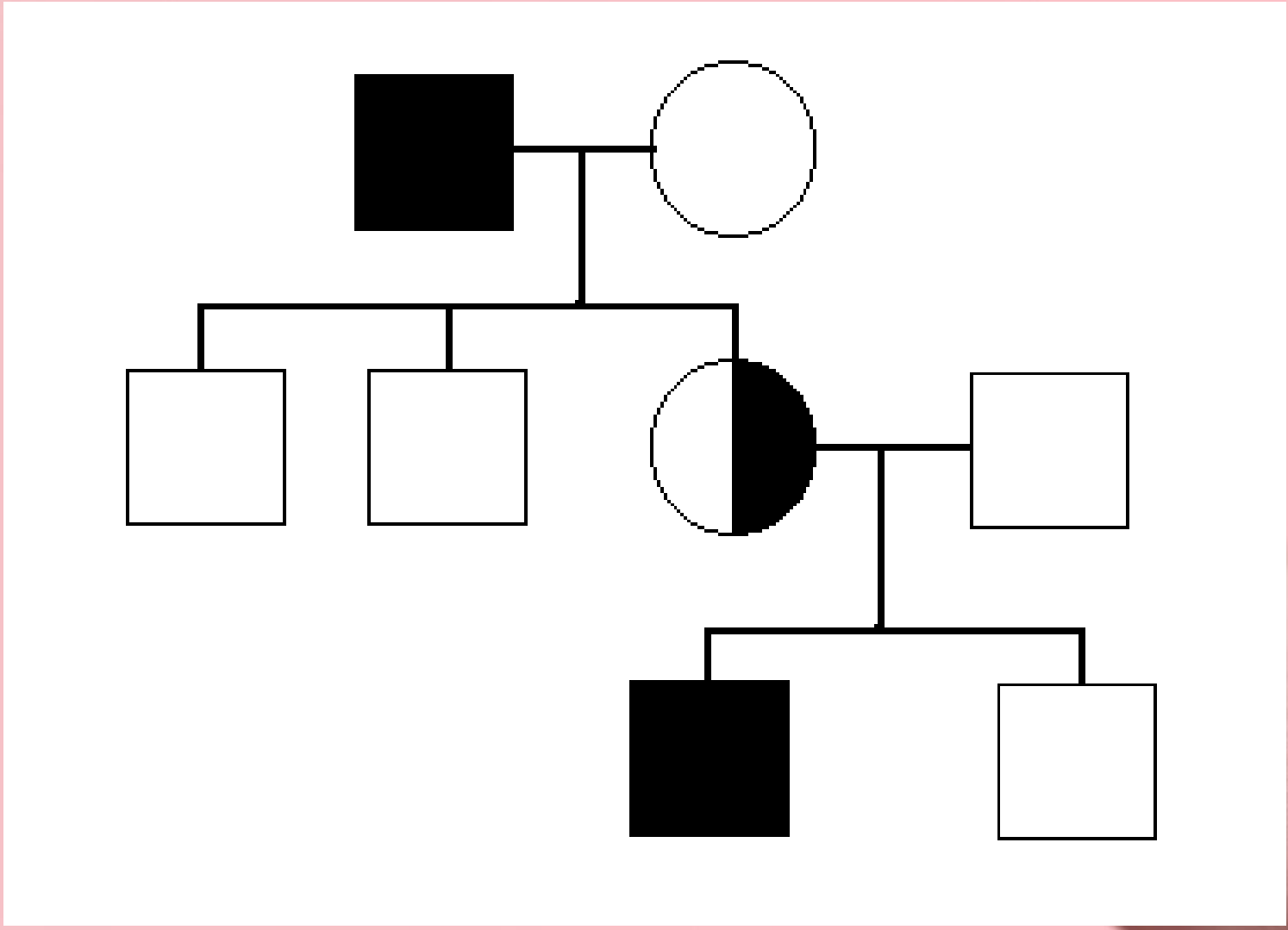
- put genes in bacteria and they make things humans need
- Ex. Growth hormone

- Curing genetic diseases – 5% of babies in USA born with one
- Decoding the human genome (determine the nucleotide sequence of about 3 billion nucleotides or about100,000 genes and to map their location on every chromosome)
- Personal Id
- Diagnosis of disease – 4,000 human genetic disorders

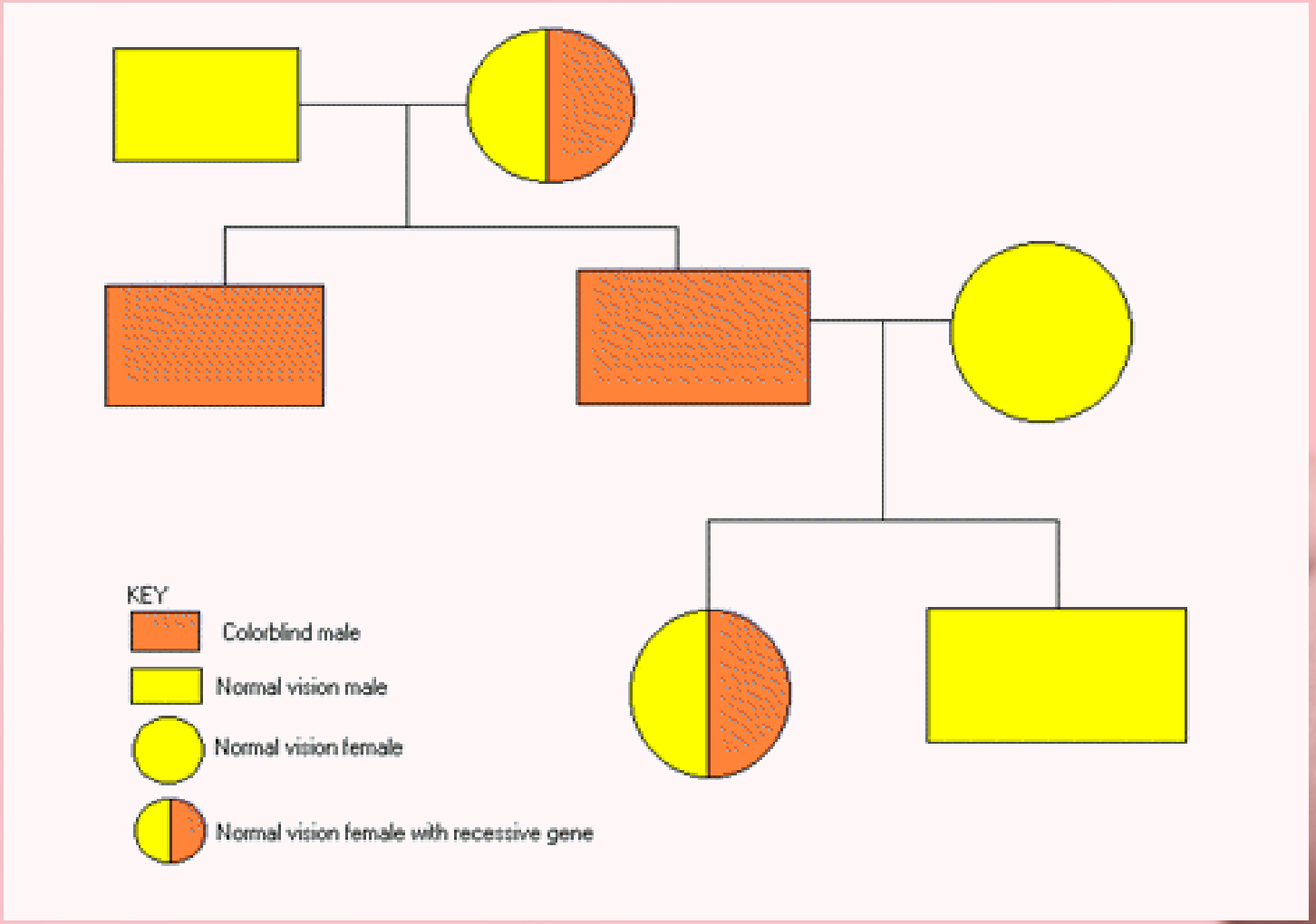
Genetic Counseling

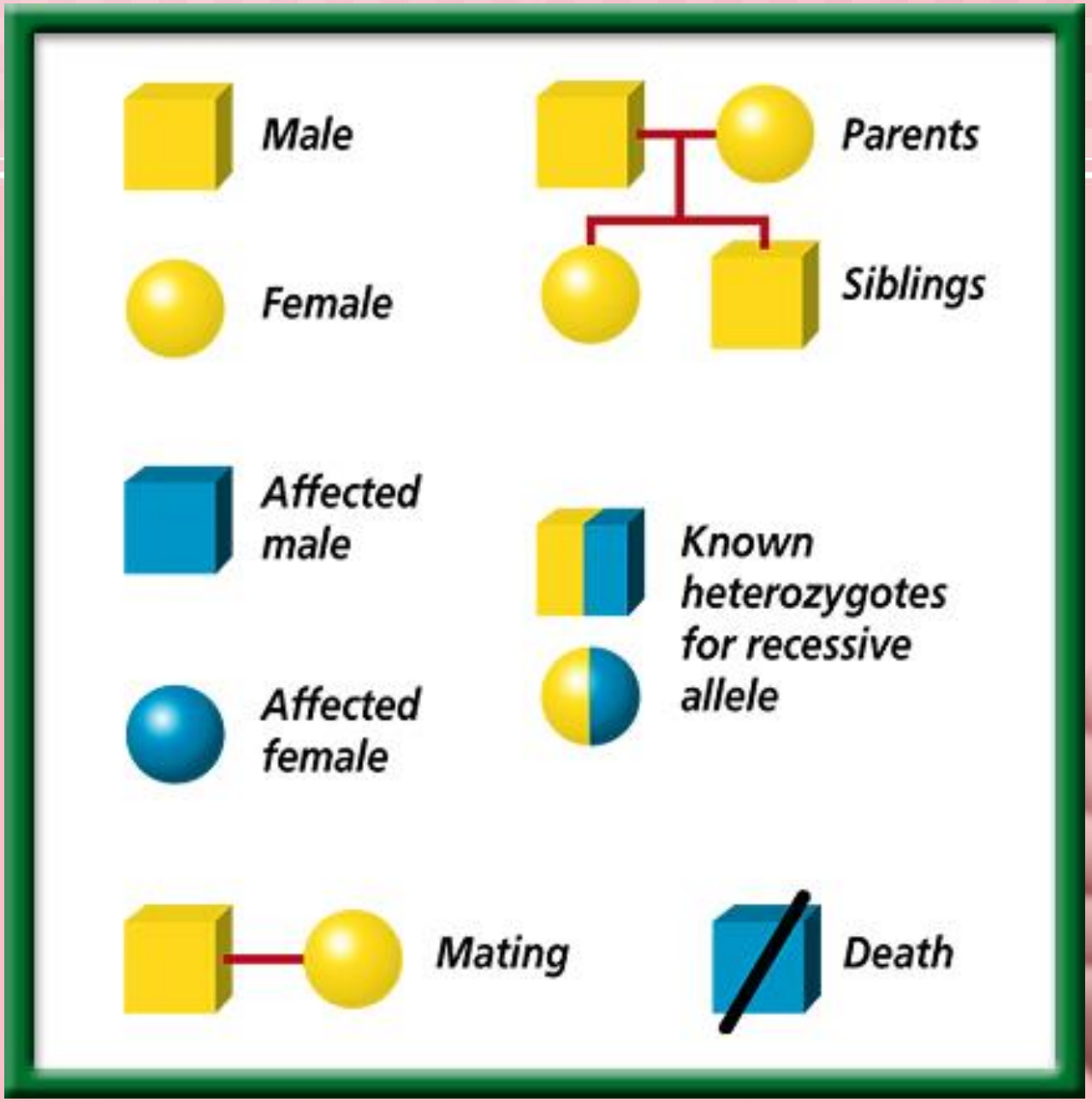
- Karyotype
- DNA Fingerprint
- CVB
- Amniocentesis
- Family History

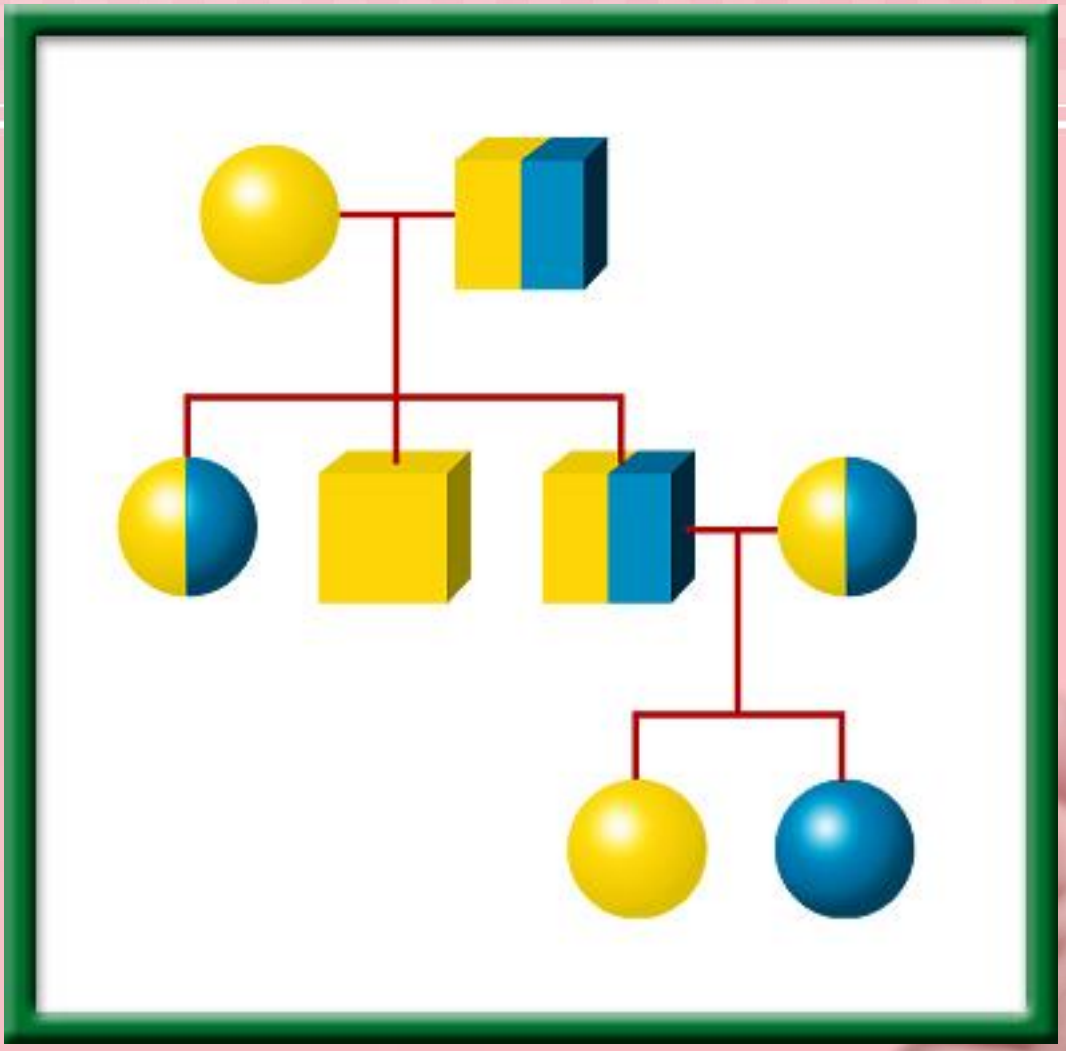


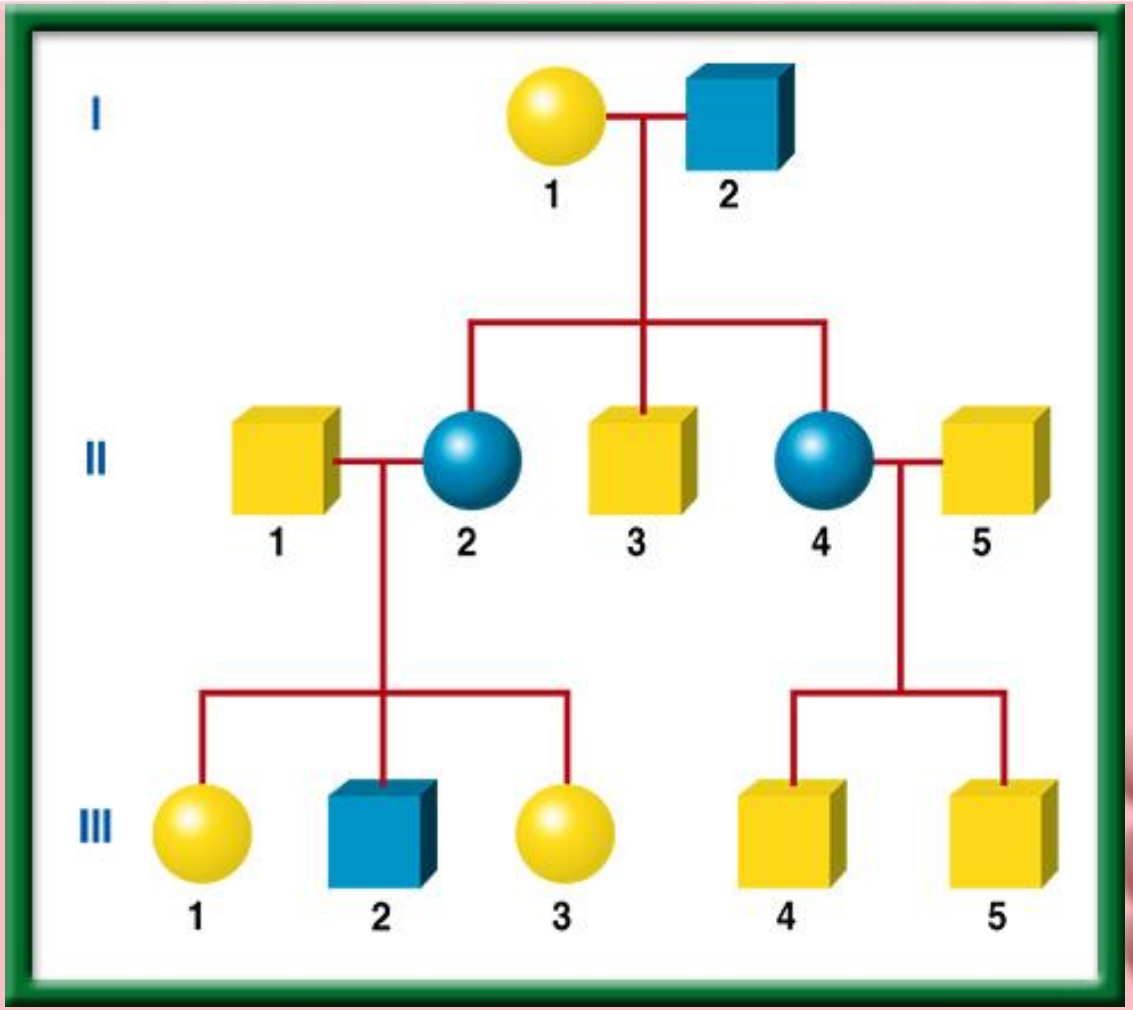


Pedigrees

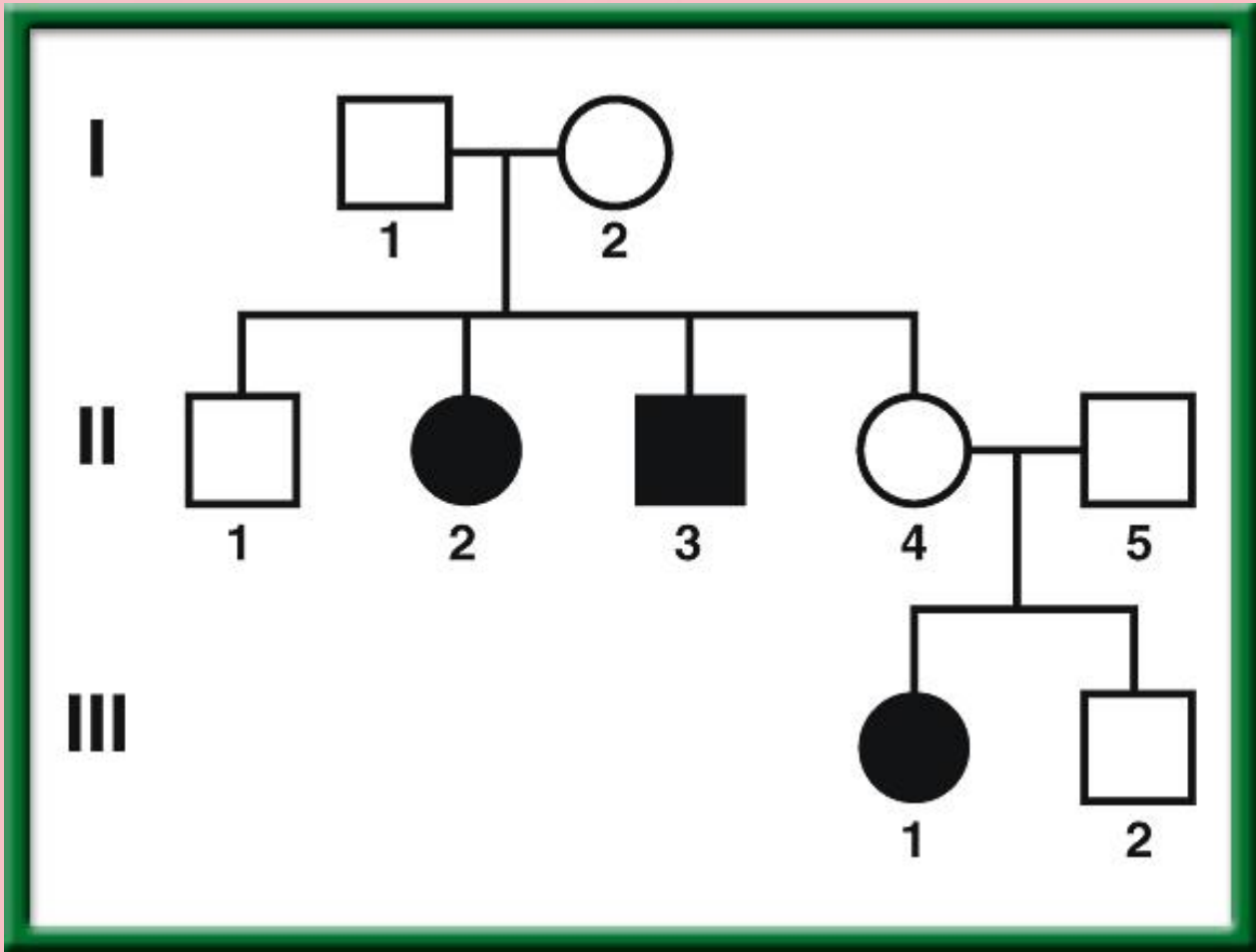

KEY
Colorblind male
Normal vision male
Normal vision female
Normal vision female with recessive gene


Male
Female
Affected male
Affected female
Mating
Parents
Siblings
Known heterozygotes for recessive allele
Death

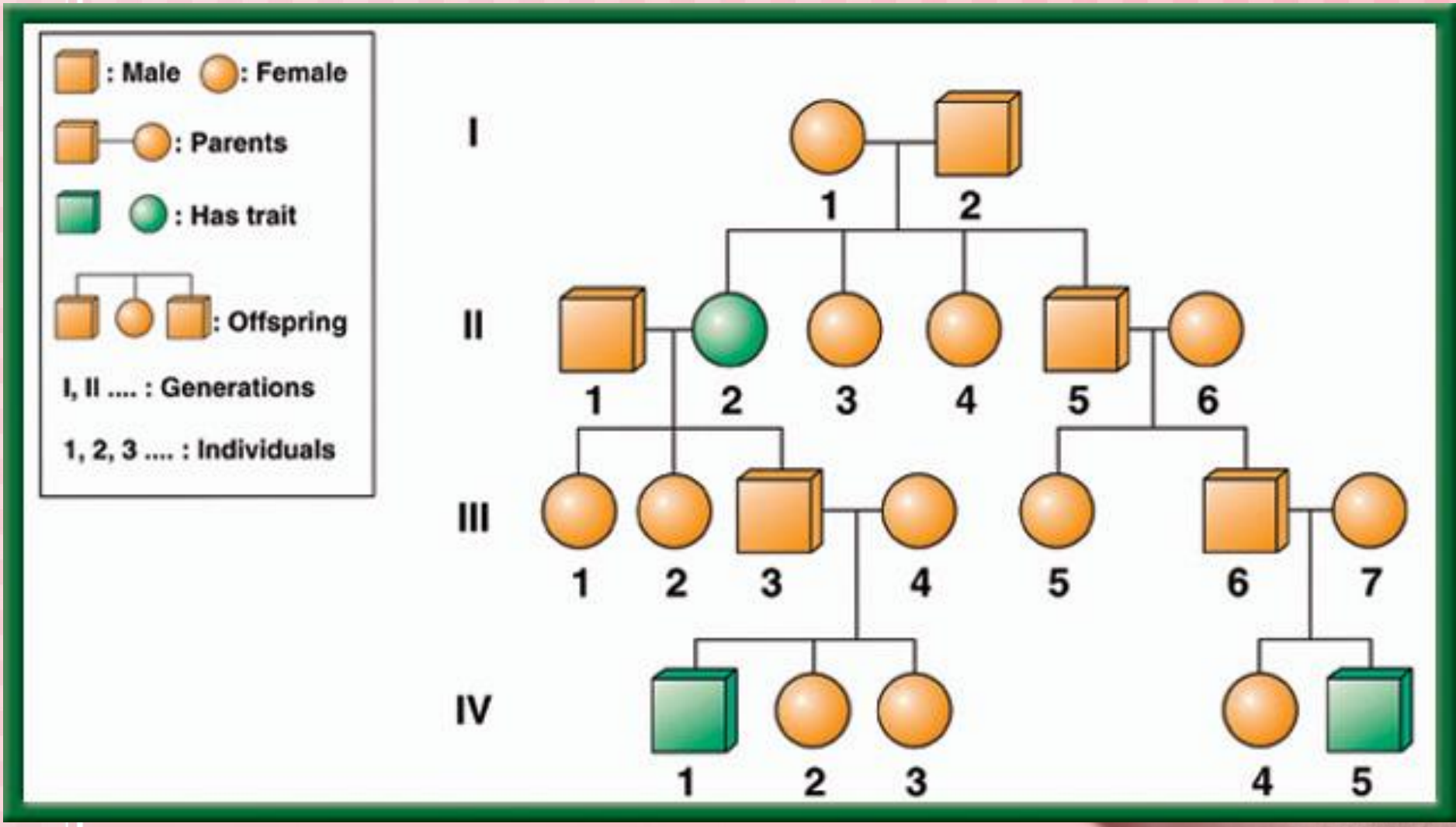



I
1
2
II
1
2
3
4
5
III
1
2
3
4
5

- Which type of inheritance pattern is shown in the above pedigree?
- What is individual II-4's genotype?
- How many different genotypes are possible for individual III-2?



In notes


: Male
: Female
: Parents
: Has trait
: Offspring
I, II .... : Generations
1, 2, 3 .... : Individuals
I
II
III
IV
1
2
3
4
5
6
7