Chapter 7

DNA Detective: Extensions of Mendelism, Sex Linkage, Pedigree Analysis, and DNA Fingerprinting

1

7.1 Extension of Mendelian Genetics

- Patterns of inheritance that are not straightforward are called extensions of Mendelian genetics
- When the offspring of two different parents has a phenotype that is intermediate to either parent, the trait is said to display _____

2

Extension	Example
Incomplete dominance Heterozygote is intermediate to either homozygote.	Flower color in snapdragons  Red = *RR* × White = *rr* = Pink = *Rr*

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Extension of Mendelian Genetics

- Predicting inheritance is more difficult for traits that are controlled by many genes, or are _____ – especially when the environment can also influence the trait

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Extension of Mendelian Genetics

Two other extensions are:

- _____ – both alleles are expressed
- _____ – there are more than two allele options for the gene

- These extensions are used in the **ABO blood system** of blood typing

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Extension of Mendelian Genetics/Essay 7.1

- Codominance:
  - If you are blood type AB, you have one allele for A and one allele for B
  - Both are seen in the codominant phenotype

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Extension of Mendelian Genetics/Essay 7.1

- Multiple allelism:
  - There are 3 options for alleles in ABO blood types (I^A, I^B, and i)
  - Even though there are 3 options, each person only has 2 alleles

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Red blood cell phenotype	Red blood cell genotype
Type O	ii
Type A (Sugar A)	I^AI^A or I^Ai
Type B (Sugar B)	I^BI^B or I^Bi
Type AB (Sugars A and B)	I^AI^B

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Extension	Example
Multiple allelism One gene has many alleles.	Coat color in rabbits is controlled by four different alleles. **Genotype** / **Phenotype** cc — Albino C^hC^h — Himalayan $C^{ch}C^{ch}$ — Chinchilla C^+C^+ — Darkly pigmented rabbit

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Extension of Mendelian Genetics/Essay 7.1

- While the ABO blood system involves extensions of Mendelian genetics, inheritance of the **Rh factor** of the red blood cells (+ or -) is determined by straightforward two-allele, completely dominant manner with RH^+ dominant to RH^-

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Extension of Mendelian Genetics

- **Hemophilia** is a genetic blood-clotting disorder and an example of _____, the ability of one gene to affect many different functions
  - Hemophilia can lead to excessive bruising, pain and swelling in the joints, vision loss from bleeding into the eye, and low red blood cell counts (anemia), resulting in fatigue
  - Neurological problems may occur if bleeding or blood loss occurs in the brain

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Extension	Example
Pleiotropy One gene has many effects.	The singed gene in fruit flies controls both bristle formation and fertility. **Genotype** / **Phenotype** *BB* and *Bb* — Normal bristles and fertile *bb* — Singed bristles, infertile

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7.2 Sex Determination and Sex Linkage

- _____ – non-sex chromosomes (22 pairs)
- **Sex chromosomes** – X and Y (1 pair)
- In humans, _____ involves the X and Y chromosomes

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Sex Determination and Sex Linkage

- There are two types of sex chromosomes in humans: X and Y
- If you have _____ you are female
- If you have _____ you are male

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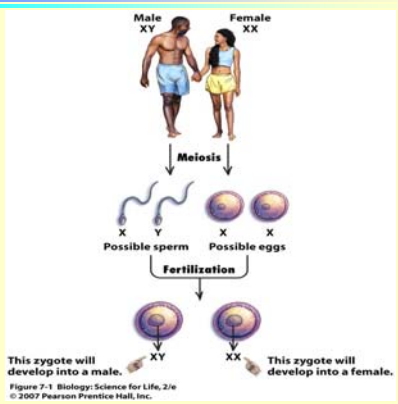

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Sex Determination and Sex Linkage

- All egg cells contain an ____ chromosome
- Sperm cells can either contain an _____ or a ____ chromosome
- Fathers determine the sex of the offspring
- Sex determination occurs through different mechanisms in some nonhumans species

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Type of Organism	Mechanism of Sex Determination
Vertebrates (fish, amphibians, reptiles, birds, and mammals)	In most vertebrates, sex is determined at fertilization by the suite of chromosomes present but differs from human sex determination in that the male may have two of the same chromosomes and the female two different chromosomes. Organisms with two of the same sex chromosomes are called homogametic; organisms with two different sex chromosomes are called heterogametic. Females are the homogametic sex in most mammal and fly species, but the reverse is true in butterflies, fish, and birds. For these species, the female determines the sex of the offspring.
Egg-laying reptiles	In many egg-laying species, two organisms with the same suite of sex chromosomes could become different sexes. In these organisms, sex depends on which genes are activated during embryonic development. For example, the sex of some reptiles is determined by the incubation temperature of the egg. Incubation temperature modifies the number and placement of several enzymes and hormone receptors in the egg. Some researchers have found that, applying a drop of the female hormone estrogen to the shell of an incubating egg will produce female offspring in temperature conditions that would normally yield all male hatchlings.
Wasps, ants, bees	In these species, sex is determined by the presence or absence of fertilization. In bees, males (drones) develop from unfertilized eggs. Females (workers or queens) develop from fertilized eggs.
Bony fishes	Some species of bony fishes change their sex after maturation. This mode of sex determination is one in which all individuals will become females unless they are deflected from that pathway due to social signals such as dominance interactions.
Caenorhabditis elegans	The nematode *C. elegans* can either be male or have both male and female reproductive organs. Organisms with both male and female reproductive organs are called hermaphrodites.

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Meiosis and Sex Chromosomes

- Sometimes the homologous pairs do not separate during meiosis
- This is called _____
- Nondisjunction results in gametes with incorrect number of chromosomes
- If fertilized, the offspring has an incorrect number of chromosomes

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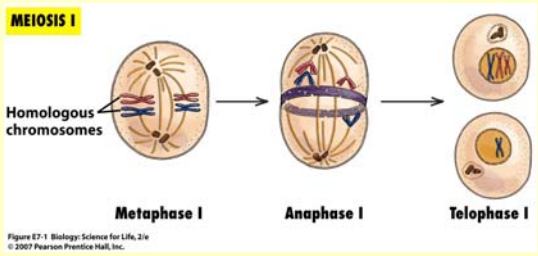

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Meiosis and Sex Chromosomes/Essay 7.2

- Having an incorrect number of chromosomes is usually detrimental to the organism
- One example is trisomy 21 (Down Syndrome)
  - One extra chromosome #21

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Conditions Caused by Nondisjunction of Autosomes	Approximate Frequency Among Live Births	Comments
Trisomy 21—Down syndrome	The probability that a woman will have a child with Down syndrome increases with age. In mothers younger than age 35, Down Syndrome occurs in approximately 1 per 1000 births and at age 45, around 4 per 1000 births.	People with Down syndrome tend to be mentally retarded, have abnormal skeletal development, and have heart defects.
Trisomy 13—Patau syndrome	1 in 5000	Affected individuals are mentally retarded, deaf, and have a cleft lip and palate.
Trisomy 18—Edwards syndrome	1 in 6000	Babies with Edwards syndrome have malformed organs, ears, mouth, and nose, leading to an elfin appearance. They are mentally retarded and usually die within 6 months of birth.

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Conditions Caused by Nondisjunction of Sex Chromosomes	Approximate Frequency Among Live Births	Comments
XO—Turner syndrome	1 in 5000 females	People with only one X chromosome are females with retarded sexual development. They are usually sterile since their ovaries often fail to develop. They can have webbing of the neck, shorter stature, and some hearing impairment. Since they are missing an X chromosome, affected females have 45 chromosomes.
Trisomy X—Meta Female	1 in 1000 females	Meta females have three X chromosomes. Two of the X chromosomes are condensed to Barr bodies and most XXX females develop normally. Since these women have an extra X chromosome, the cells of their bodies have 47 chromosomes.
XXY—Kleinfelter syndrome	1 in 1000 males	Males with the XXY genotype are less fertile than XY males, have small testes, sparse body hair, some breast enlargement, and may have mental retardation. Testosterone injections can reverse some of the anatomical abnormalities but not the mental retardation and lowered fertility.
XXY—Kleinfelter syndrome	1 in 1000 males	Males with two Y chromosomes tend to be taller than average but have a normal male phenotype.

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Sex Linkage

- The genes on the X or Y chromosomes are called _____
- Genes on X are called "_____," while those on Y are called "_____"
- The X chromosome is much larger and carries far more genetic information
  - Y chromosome only contains ~20 genes that mostly determine maleness or male fertility
  - X chromosome contains ~1500 genes that have no alleles on the Y chromosome, including color vision, blood clotting, and certain structural proteins in muscles

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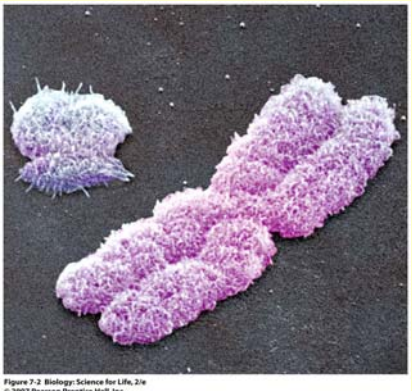
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X-Linked Genes

- **X-liked genes** are located on the X chromosome
- Males inherit their X chromosomes from their mothers
- Males get the Y chromosome from their fathers
- Since males only have one X chromosome, they are more likely to suffer from X-linked diseases, like hemophilia, caused by recessive alleles

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X-Linked Genes

- Since females get one X chromosome from each parent, and have two copies, they are less likely to suffer from X-linked diseases because they have a greater likelihood of carrying at least one functional version of each X-linked gene
- Females can be _____ and pass the disease on to offspring, especially sons
- Many female organisms actually shut off or inactivate one of their 2 X chromosomes early in development – this process is called **X inactivation** (Essay 7.3, p. 178)

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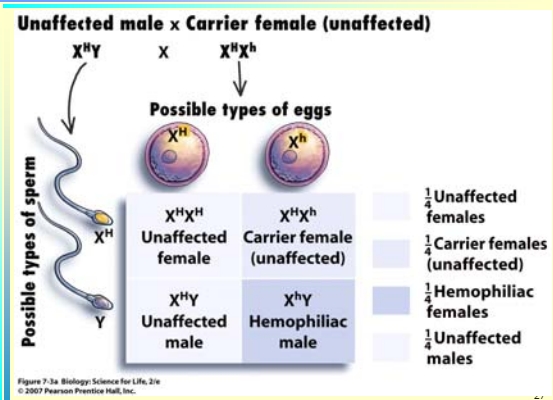

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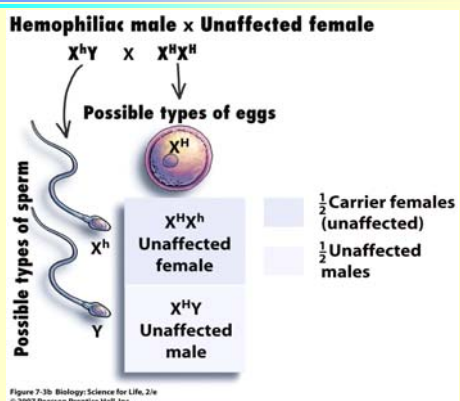

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X-Linked Genes

- There are several X-linked traits in humans and other organisms

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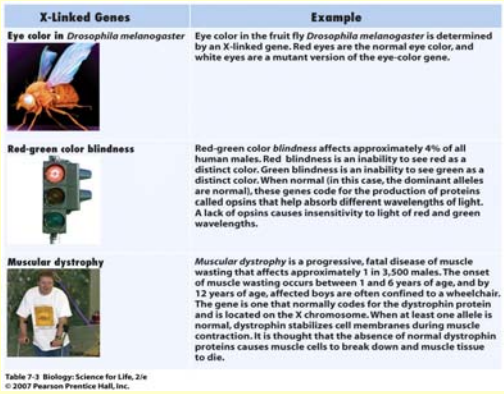

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Y-Linked Genes

- Located on the Y chromosome
- Passed from fathers to sons
- Very few genes on the Y chromosome
- SRY gene (sex-determining region of the Y chromosome)
  - The expression of this gene triggers a series of events leading to development of the testes and some of the specialized cells required for male sexual characteristics

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7.3 Pedigrees

- A **pedigree** is a chart showing inheritance patterns in a family (a family tree that follows the inheritance of a genetic trait for many generations of relatives
- Often used in studying human genetics since it is impossible and unethical to set up controlled matings between humans
- Allow scientists to study inheritance by analyzing matings that have already occurred

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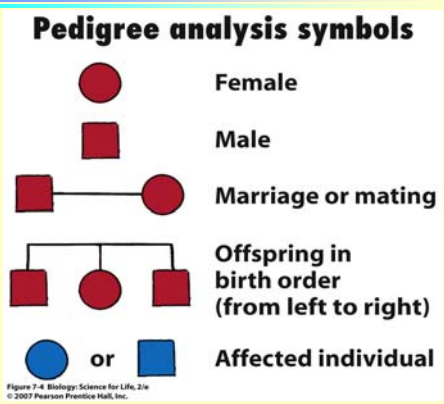
Pedigree analysis symbols



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Pedigrees

- Pedigrees can be used for different types of inheritance patterns

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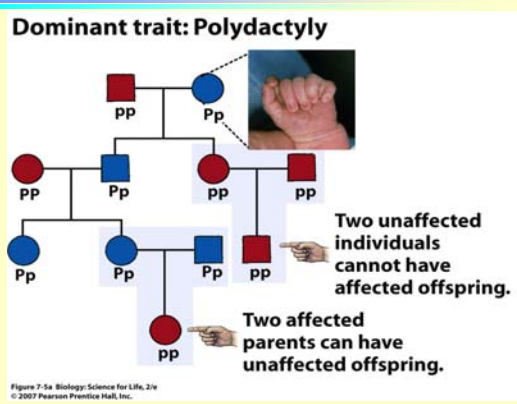
Dominant trait: Polydactyly



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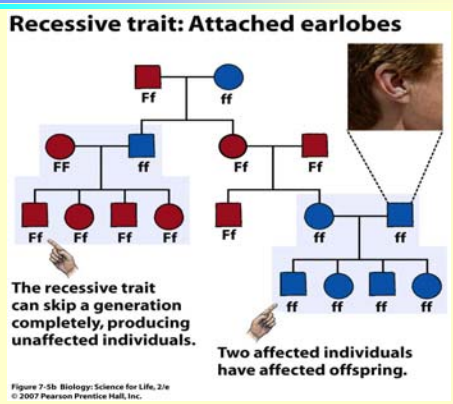
Recessive trait: Attached earlobes



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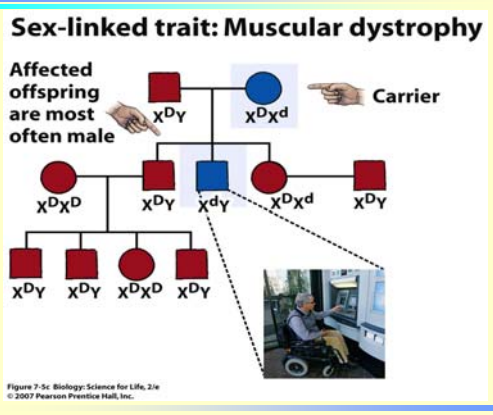

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Pedigrees

- Pedigrees involving the Romanov family (the former Russian royal family) are useful in showing inheritance of the hemophilia allele

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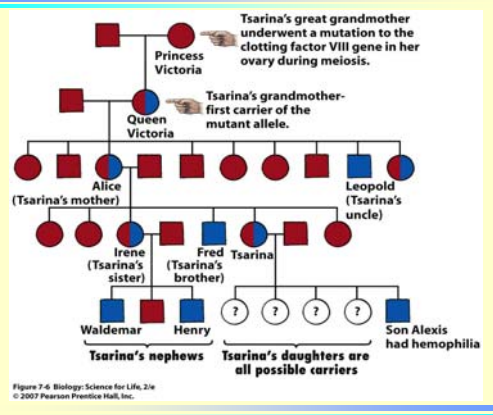

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7.4 DNA Fingerprinting

- Unless they are identical twins, individuals have unique DNA
- **DNA fingerprinting** is an unambiguous identifying technique that takes advantage of differences in DNA sequence
- The process of DNA fingerprinting begins by isolating DNA from blood, semen, vaginal fluids, hair roots, skin, skeletal remains, or elsewhere

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Polymerase Chain Reaction (PCR)

- If there is only a small amount of DNA available, scientists can augment the amount using a technique called **PCR** (_____)
  - Used to amplify or produce large quantities of DNA

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PCR

- Put DNA to be amplified in a test tube
- Add in nucleotides (A, T, C, G)
- Add in ***Taq* polymerase**
  - From *Thermus aquaticus*
  - Can make DNA at high temperatures

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PCR

- Heat tube to _____ DNA (split up the middle of the double helix to produce single strands) and then cool
- *Taq* polymerase builds new DNA strands
- Keep heating and cooling to make millions of DNA copies

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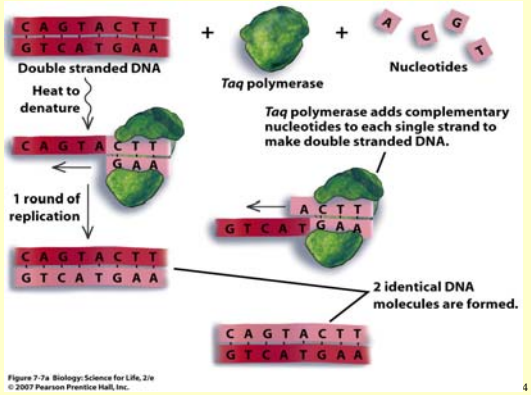

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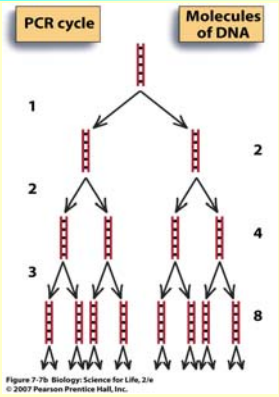

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DNA Fingerprinting

- Treat the DNA with _____
- These enzymes cut DNA at specific nucleotide sequences
- Because each individual has distinct nucleotide sequences, cutting different people's DNA with the same enzymes produces fragments of different sizes

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RFLP Analysis

- The different sized fragments are called _____, or **RFLPs**
- The RFLP patterns can be analyzed

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RFLP Analysis

- Everyone has genetic sequences that vary in number called _____, or **VNTRs**
- Everyone has different amounts of VNTRs
- The VNTRs make the different sized RFLPs

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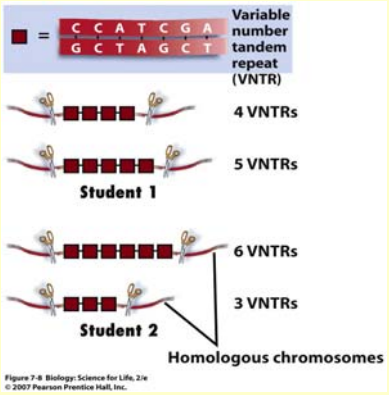

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Gel Electrophoresis

- Fragments of DNA from restriction enzyme cleavage are separated from each other when they migrate through a support called **agarose gel**
- An electric current is applied, and the gel impedes the larger DNA fragments more than the smaller ones
- The current pulls the negatively charged DNA molecules toward the bottom (positively charged) edge of the gel, further facilitating the size-based separation

Gel Electrophoresis

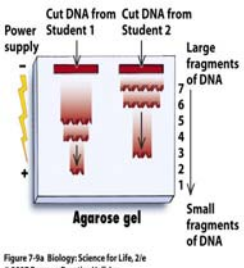
- The size-based separation of molecules of DNA when an electric current is applied to a gel is called **gel electrophoresis**
- Segments of DNA with more repeats would be heavier than those with fewer repeats
- Heavier DNA segments will not migrate as far in the gel as lighter DNA fragments
- Thus, agarose gel electrophoresis separates the DNA fragments on the basis of their size

Gel Electrophoresis

- The separated DNA fragments are then drawn out of the gel using filter paper
- The filter paper is treated with chemicals that break the hydrogen bonds in DNA and separate the strands
- The single stranded DNA is stuck to the filter paper
- The filter paper is washed with a radioactive **probe** of single stranded DNA
- The probe is designed to be complementary to the specific VNTR sequences present

Gel Electrophoresis

- Once it is bound to the labeled probe, the presence of radioactive DNA can be detected on photographic film, which records the location of DNA when the radiation emitted from radioactive DNA molecules bombards the film
- A piece of X-ray film placed over the filter paper shows the locations of the radioactive DNA as a series of bands
- Different individuals have different patterns of bands – which make up the fingerprint



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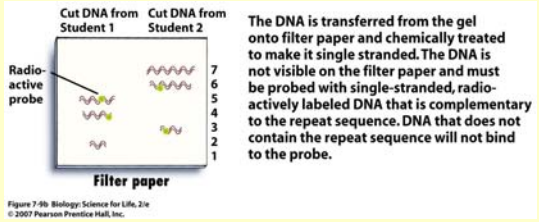
DNA from two different individuals is cut with restriction enzymes and loaded on an agarose gel. When these fragments are subjected to an electric current, shorter fragments migrate through the gel faster than do larger fragments.

Student 1 has DNA sequences that carry 4 and 5 repeat sequences. Student 2 has 3 and 6 repeats. The remaining DNA is DNA that does not carry repeat sequences. Even though the DNA is visible in this figure, DNA is not visible with the unaided eye.



The DNA is transferred from the gel onto filter paper and chemically treated to make it single stranded. The DNA is not visible on the filter paper and must be probed with single-stranded, radio-actively labeled DNA that is complementary to the repeat sequence. DNA that does not contain the repeat sequence will not bind to the probe.

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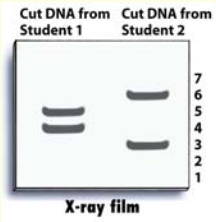

When the filter is exposed to X-ray film, radioactive DNA sequences (where the probe is bound) produce a characteristic banding pattern, or DNA fingerprint. Student 1's DNA has 4 and 5 VNTRs, and Student 2's DNA has 3 and 6 VNTRs. The two bands represent the number of VNTR repeats.

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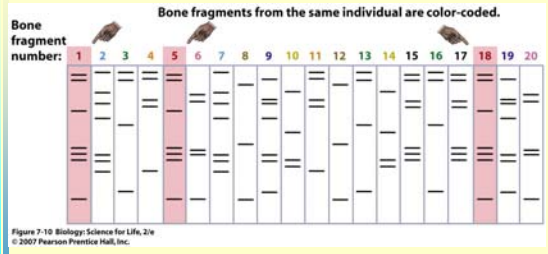

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Meiosis and DNA Fingerprinting

- DNA fingerprints of children should be similar to the DNA fingerprints of parents
- The VNTRs occur on chromosomes and the chromosomes are put into gametes and inherited by offspring

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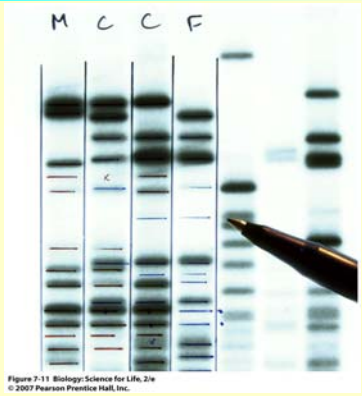

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Meiosis and DNA Fingerprinting

- DNA fingerprinting can show which individuals are the parents of specific children

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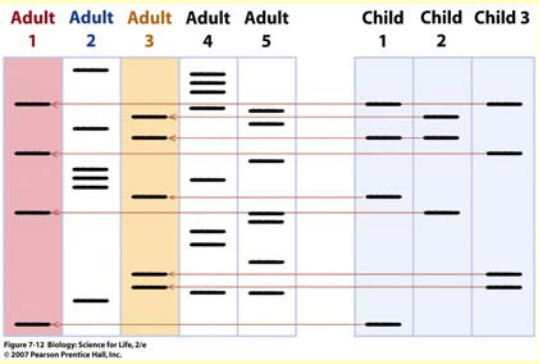

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DNA Fingerprinting

- Anna Anderson was a young woman who claimed to be Anastasia Romanov, the supposedly executed Russian princess
- A DNA fingerprint showed she was not related to the family, based on DNA analyses of the family remains found in their grave

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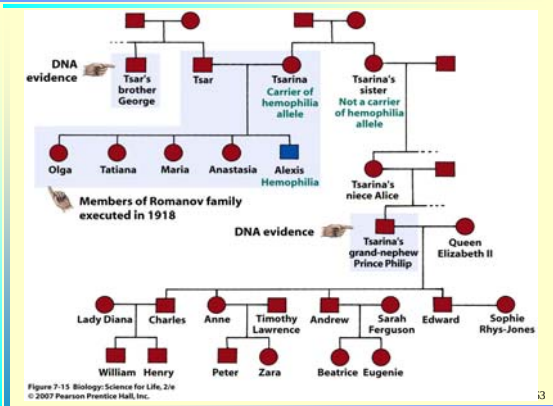

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