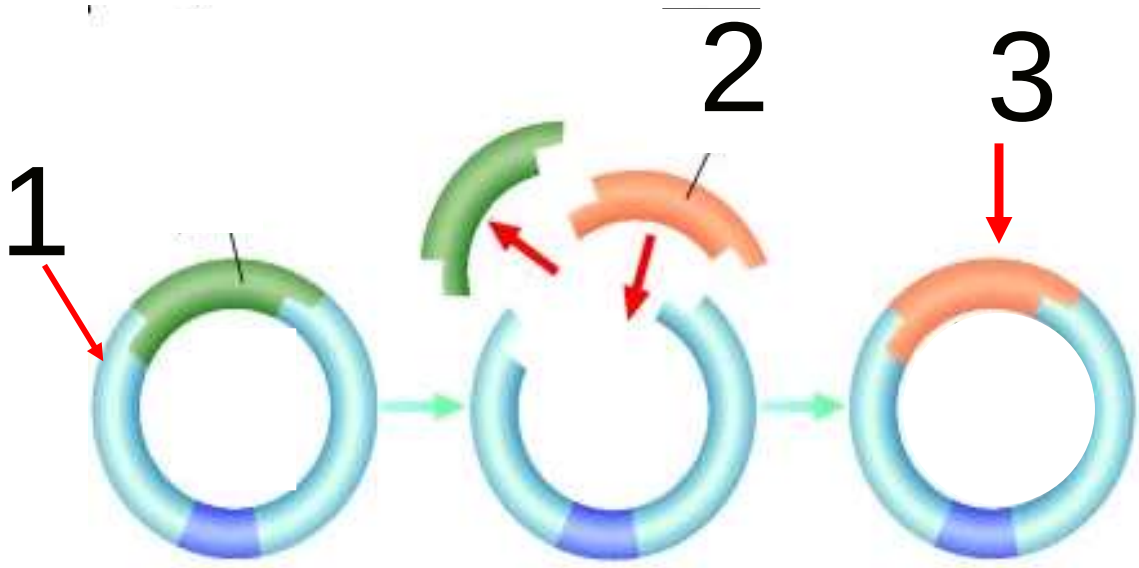
9.6 Genetic Screening and Gene Therapy

Today's Warm-up

- On your paper, write the vocabulary term that matches each numbered item below.



9.6 Genetic Screening and Gene Therapy

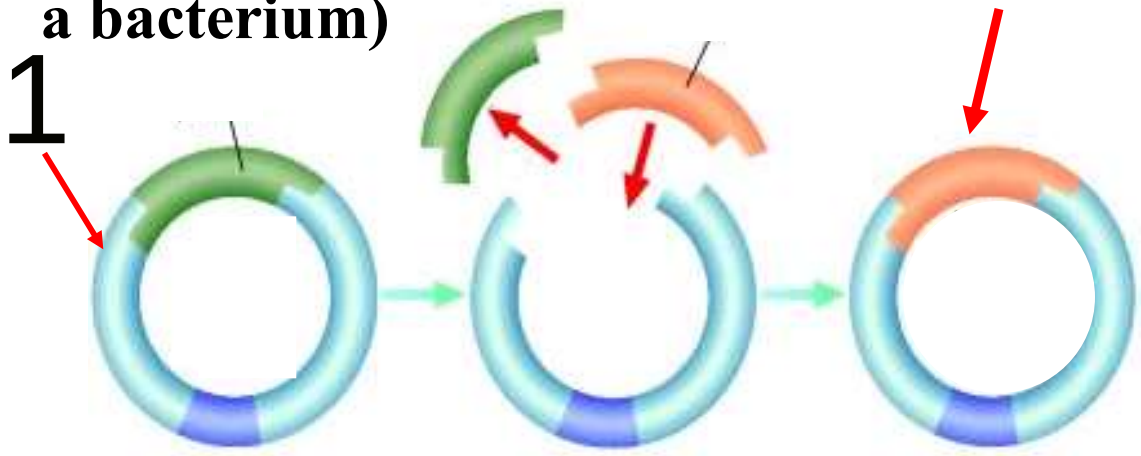
- Today's Warm-up

Vector (plasmid from a bacterium)

Donor DNA from another organism

Recombinant DNA (DNA composed from more than one source

Transgenic organism (has recombinant DNA



9.6 Genetic Screening and Gene Therapy

EQ

How can our knowledge of genetics improve human quality of life?

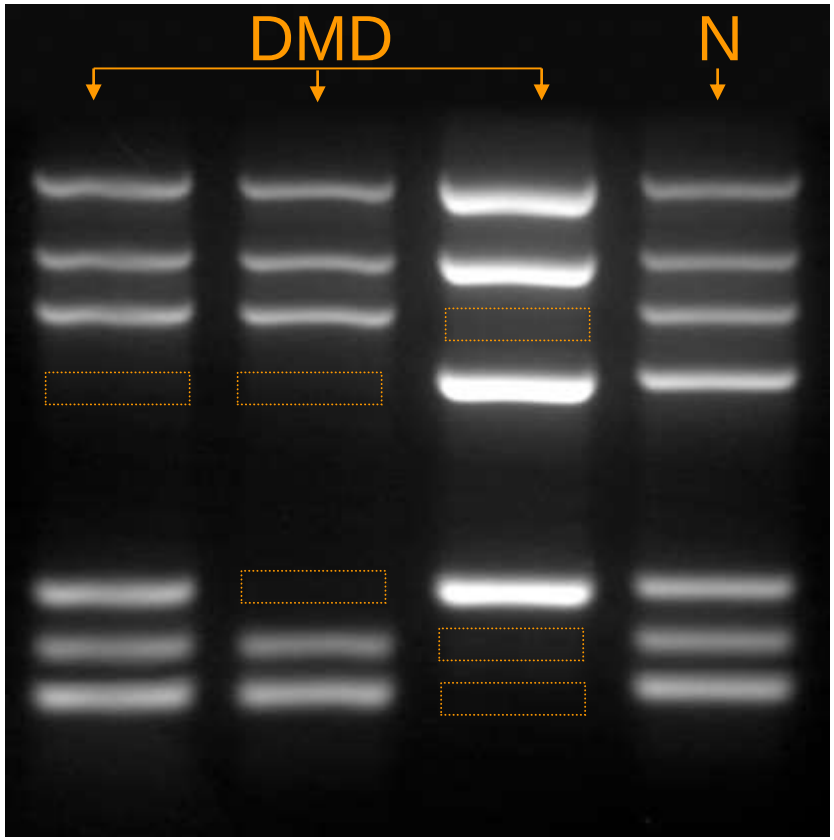
KEY CONCEPT

Genetics provides a basis for new medical treatments.

9.6 Genetic Screening and Gene Therapy

Genetic screening can detect genetic disorders.

- Genetic screening involves the testing of DNA.
  - determines risk of having or passing on a genetic disorder
  - used to detect specific genes or proteins
  - can detect some genes related to an increased risk of cancer
  - can detect some genes known to cause genetic disorders



9.6 Genetic Screening and Gene Therapy

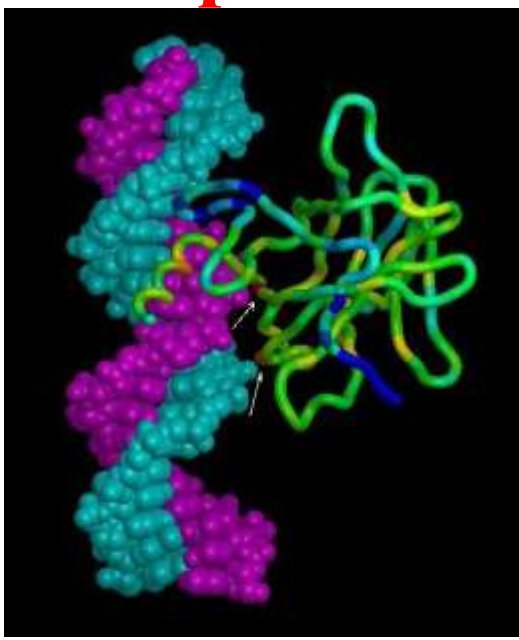
Gene therapy is the replacement of faulty genes.

- Gene therapy replaces defective or missing genes, or adds new genes, to treat a disease.
- Adding a corrected copy of the gene may help the affected cells, tissues and organs work properly.
- Gene therapy differs from traditional drug-based approaches, which may treat the problem, but which do not repair the underlying genetic flaw.

9.6 Genetic Screening and Gene Therapy

Why use gene therapy?

1. **To replace missing or mutated genes.** (most common gene therapy approach)
   - Some cells become diseased because certain genes have been permanently turned off. Other cells may be missing certain genes.
   - Researchers hope that replacing missing or defective genes can help treat certain diseases.
   - For instance, a common tumor suppressor gene called p53 normally prevents tumor growth in your body. Several types of cancer have been linked to a missing or inactive p53 gene. If doctors could replace p53 where it's missing, that might trigger the cancer cells to die.



9.6 Genetic Screening and Gene Therapy

Why use gene therapy?

1. **To replace missing or mutated genes.**
2. **To change the regulation of a gene.**
   - Mutated genes that cause disease could be turned off so that they no longer promote disease, or healthy genes that help prevent disease could be turned on so that they can inhibit the disease.
3. **To make diseased cells more evident to the immune system.**
   - In some cases, the immune system doesn't attack diseased cells because it doesn't recognize them as intruders. Using gene therapy, physicians could potentially infuse mutated cells with genes that make them more recognizable to your immune system.
   - Or, enhancements could be made to immune cells to make it easier for them to recognize mutated cells.

9.6 Genetic Screening and Gene Therapy

- Gene therapy has many technical challenges.
  - inserting gene to correct faulty cells
  - controlling gene expression
  - determining effect on other genes

9.6 Genetic Screening and Gene Therapy

First Approved Gene Therapy – Adenosine Deaminase

- The first disease approved for gene therapy treatment was **adenosine deaminase** (*ADA*) deficiency, a rare genetic disorder.
- The *ADA* gene encodes an enzyme called adenosine deaminase, which is needed for immune system function.
- Children with this disorder have severe immunodeficiency and are prone to serious, sometimes life-threatening, infections.
- Although *ADA* deficiency can be treated with a drug called PEG-ADA, the drug costs more than $100,000 per year and it must be taken by injection for life.

9.6 Genetic Screening and Gene Therapy

First Approved Gene Therapy – Adenosine Deaminase

- ADA deficiency was selected for the first approved human gene therapy trial for several reasons:
  - The disease is caused by a defect in a single gene, which increases the likelihood that gene therapy will succeed.
  - The gene is regulated in a simple, "always on" fashion, unlike many genes whose regulation is more complex.
  - The amount of *ADA* used in therapy does not need to be precisely regulated. Even small amounts of the enzyme are known to be beneficial, and larger amounts are well tolerated by patients.

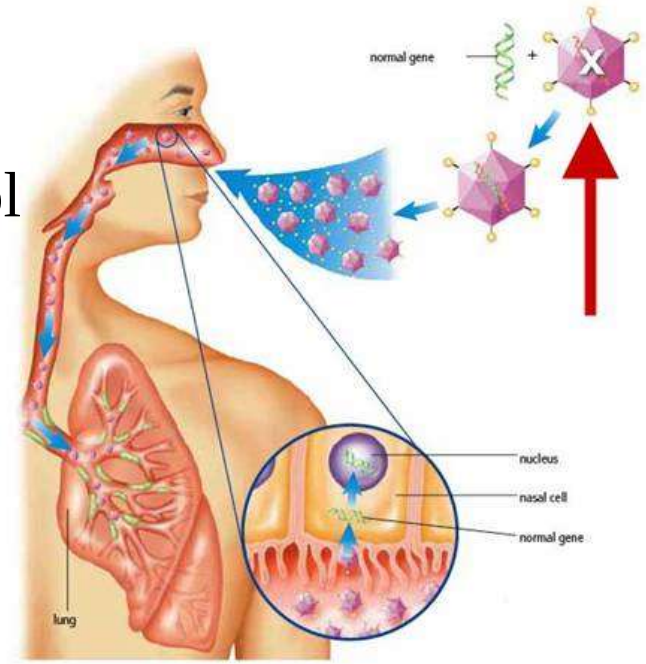
In 1990 Ashanti de Silva became the first patient to receive gene therapy for ADA deficiency. Shown here at age 13, she continues to lead a healthy, active life.

Photo: Courtesy of Van de Silva

9.6 Genetic Screening and Gene Therapy

- **Gene Therapy is Promising for Cystic Fibrosis**
  - Patients would be able to inhale the gene therapy.

1. Gene therapy trials in CF patients introduced the normal CFTR gene into liposomes and sprayed these liposomes into the nose of CF patients with an aerosol or hose.
2. The liposomes fuse with lipids in cell membranes of trachea cells, release the normal CFTR gene into the cytoplasm of these cells and is copied into mRNA.
3. The normal protein is then translated from this mRNA, enters the cell membranes, and starts to transport chloride ions out of cells.



9.6 Genetic Screening and Gene Therapy

- **Sickle Cell Anemia: Hope from Gene Therapy**

1. Complete the Punnett Squares provided using the following genotypes:
   - AA (normal hemoglobin)
   - AS (carrier for sickle cell trait – heterozygous)
   - SS (has sickle cell trait)
2. Analyze the results:
   1) Possible phenotypes in offspring
   2) Probability of being normal, carrier, having sickle cell trait in the offspring

Gene Therapy Discussion

- Who decides what is normal and what is a disability?
- How does this view affect an individual with a disability or disorder? Will they be viewed or treated differently by society than "normal" individuals?
- Are disabilities diseases? Do they need to be cured or prevented?
- How does this view affect an individual with a disability or disorder?

Gene Therapy Discussion

- Gene therapy in somatic (body) cells (*such as what was done to the mice in the video*) does not alter the genetic code of eggs and sperm and only affects the individual being treated. How do you feel about 'germline' gene therapy in which the eggs and sperm are altered, thus determining how the trait will be passed to future generations?

- Currently gene therapy is very costly. Who will determine which individuals will be offered treatment with gene therapy? Who will pay for the therapy? Should only people with life-threatening conditions have access to gene therapy?