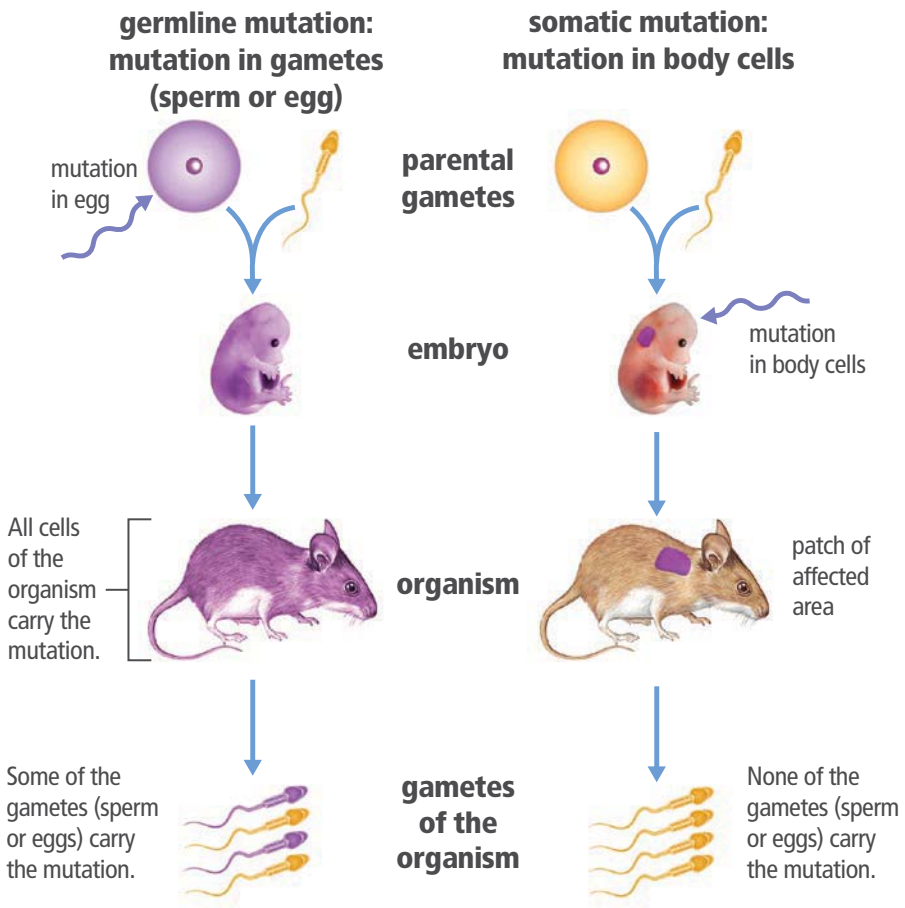
Effects of Mutations

You have seen how HbS, the sickle cell anemia allele, can be passed on from parent to offspring. Whether or not a mutation such as this gets inherited depends on the type of cell in which the mutation occurs. If a mutation is transmitted, it may or may not affect the phenotype, or the physical expression of a trait, in the organism.

Impacts on Offspring

There are two major types of cells in the body: body cells and germ cells. Germ cells are involved in the formation of gametes. Body cells, or somatic cells, include all other cells of the body. Mutations happen in both of these cell types, but only mutations in germ cells may be passed from parent to offspring. Mutations in the germ line affect the phenotype of offspring. Often, this effect is so harmful that offspring do not develop properly or die before they can reproduce. Other mutations, though less severe, often still result in less adaptive phenotypes. More rarely, a mutation results in a more beneficial phenotype.

FIGURE 15: Mutations can occur in gametes and in body cells.



Explain Would a mutation in one of your muscle cells be passed down to your offspring? Use evidence to support your explanation.

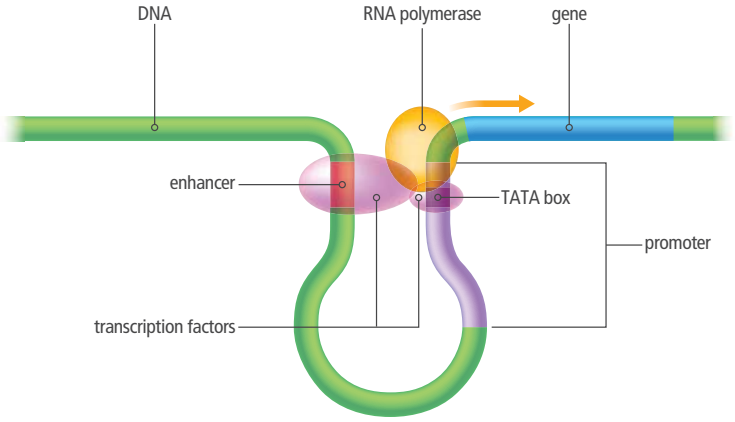
Impacts on Phenotype

Chromosomal mutations affect many genes and have a major impact on the organism. A mutation may break up a gene, inactivating it, or make a new hybrid gene with a new function. Translocated genes may also come under the control of new promoters.

Gene mutations, though smaller in scale than chromosomal mutations, can also have a big effect on an organism. Even a mutation in a noncoding region can cause problems. Recall that DNA sequences such as promoters and enhancers interact with transcription factors and RNA polymerase to start transcription. Therefore, a mutation that affects any one of these elements could also affect the expression of one or more genes.

Collaborate How might a mutation that affects a regulatory element, such as a promoter, transcription factor, or enhancer, affect the expression of a gene? Discuss possible outcomes of mutations affecting each of these elements. Would the gene be expressed? If so, how might its expression change?

FIGURE 16: A promoter is a segment of DNA that binds to proteins that help initiate the transcription of a gene.

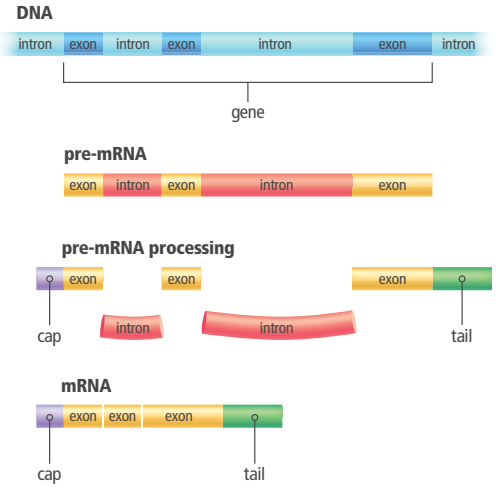


Many gene mutations, however, do not affect an organism's phenotype. Remember that many codons code for the same amino acid. Therefore, some substitutions have no effect, especially those occurring in the third nucleotide of a codon. If AAG changes to AAA, the resulting protein still has the correct amino acid, lysine. Similarly, an incorrect amino acid might have little effect on a protein if it has about the same size or polarity as the original amino acid or if it is far from an active site.

Cause and Effect

Would a mutation in an intron affect the structure and function of the resulting protein? Explain your answer.

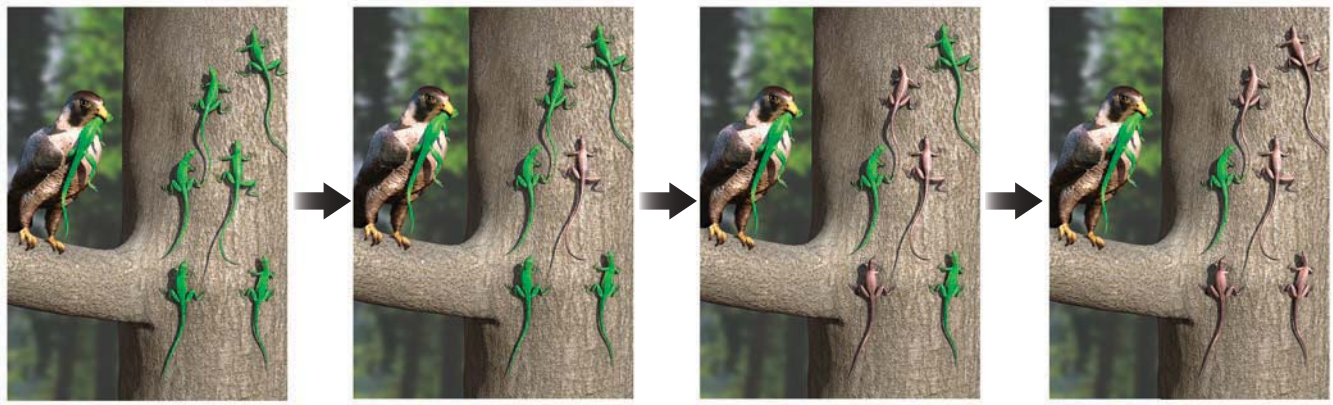
FIGURE 17: In mRNA processing, introns are removed from the pre-mRNA strand.



Impacts on Genetic Diversity

Genetic variation, or genetic diversity, is the variety of genes within a population. While genetic recombination via sexual reproduction is a major source of genetic diversity, mutations in germ cells are the ultimate source of genetic diversity in an organism's genome. Genetic diversity is the basis of a process called natural selection. In natural selection, environmental factors "select for" phenotypes that allow organisms to better survive and reproduce. For example, an individual might have a phenotype that allows the organism to attract more mates than other individuals. This individual would have more opportunities to pass down their genes, and over the course of many generations, this phenotype could become more prevalent in the population.

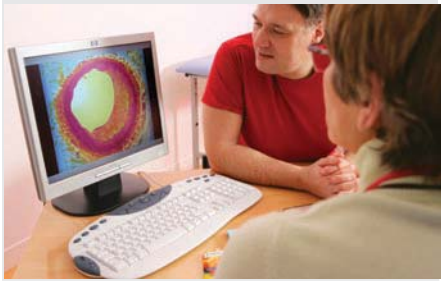
FIGURE 18: Mutations increase genetic diversity, which is the basis of natural selection.



Analyze Assume that in these lizards, the brown phenotype results from a mutation. Why does this phenotype become more common in the population over time?

When less adaptive phenotypes result from mutations, natural selection typically removes these mutant alleles from the population. Less adaptive phenotypes may make it difficult for organisms to survive or reproduce. These traits are "selected against" by environmental factors and tend to become less prevalent in a population over time.

FIGURE 19: A mutation in humans has been shown to protect against coronary artery disease.



Sometimes, a mutation results in a more beneficial phenotype. These mutations are favored by natural selection. For example, one type of deletion mutation in humans has been shown to protect against coronary artery disease, a condition characterized by the hardening and thickening of artery walls.

Explain Two species of rabbits occupy an area that experiences four seasons. The first type of rabbit has white fur in the winter and brown fur in the spring. The other species has brown fur all year round. Which of these types of rabbits has a more beneficial phenotype? Explain your answer.

Explain In some cases, mutations that have some harmful effects continue to persist in certain human populations. Why might a mutation with detrimental effects persist in a given population?