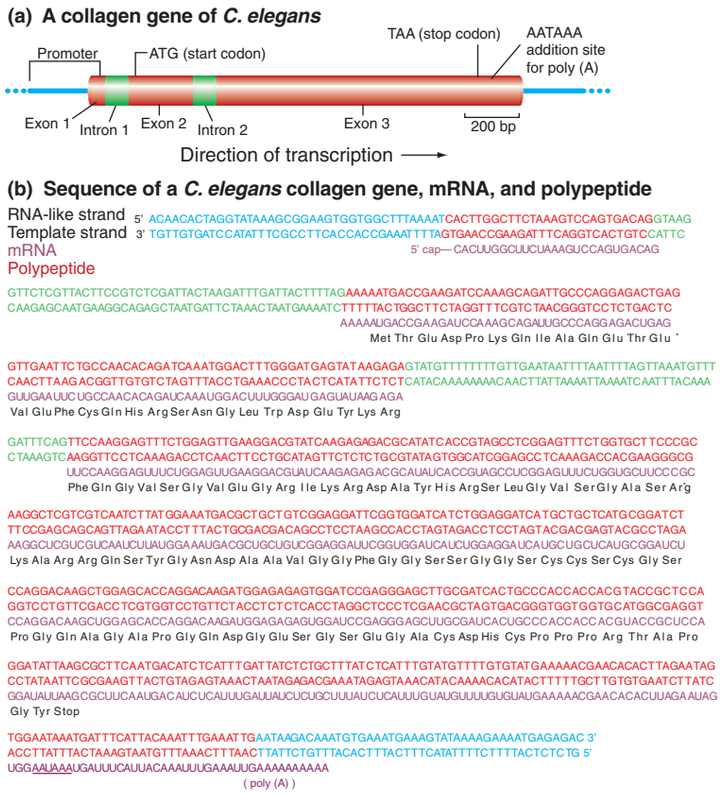

Figure 8.26 Expression of a *C. elegans* gene for collagen. (a) Landmarks in the collagen gene. **(b)** Comparison of the sequence of the collagen gene's DNA with the sequence of nucleotides in the mature mRNA (*purple*) pinpoints the start of transcription, the location of exons (*red*) and introns (*green*), and the position of the AAUAAA poly-A addition signal (*underlined in purple*). Translation of the mRNA according to the genetic code determines the amino acids of the protein product.

How Mutations Affect Gene Expression

We have seen that the information in DNA is the starting point of gene expression. The cell transcribes that information into mRNA and then translates the mRNA information into protein. Mutations that alter the nucleotide pairs of DNA may modify any of the steps or products of gene expression.

Mutations in a Gene's Coding Sequence Can Alter the Gene Product

Because of the nature of the genetic code, mutations in a gene's amino-acid-encoding exons generate a range of repercussions (Fig. 8.27a).

Silent Mutations Do Not Alter the Amino Acid Specified

One consequence of the code's degeneracy is that some mutations, known as **silent mutations,** direct the inclusion of the same amino acid as the unmutated DNA and therefore have no effect on the amino-acid composition of the encoded polypeptide or on phenotype. The majority of silent mutations change the third nucleotide of a codon, the position at which most codons for the same amino acid differ. For example, a change from GCA to GCC in a codon would still yield alanine in the protein product.

Missense Mutations Replace One Amino Acid with Another

Missense mutations that cause substitution in the polypeptide of an amino acid with chemical properties similar to the one it replaces may have little or no effect on protein function. Such substitutions are *conservative.* For example, a mutation that alters a GAC codon for aspartic acid to a GAG codon for glutamic acid is a conservative substitution because both amino acids have acidic R groups. By contrast, *nonconservative* missense mutations that cause substitution of an amino acid with very different properties are likely to have more noticeable consequences. A change of the same GAC codon for aspartic acid to GCC, a codon for alanine (an amino acid with an uncharged, nonpolar R group), is an example of a nonconservative substitution. The effect on phenotype of any missense mutation is