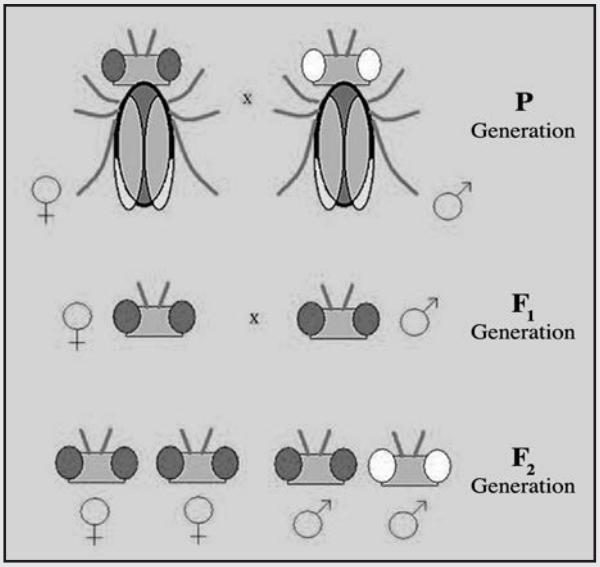
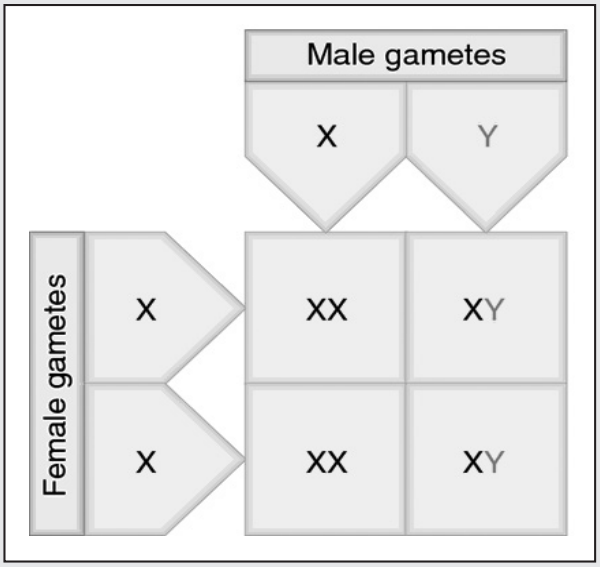
## Figure 34




Thomas Morgan's *Drosophila* sex-linked eye color experiment.

## Figure 35




Punnett square showing sex chromosome combinations for male and female gametes.



cross, the percentage of recombinant offspring was correlated with the distance between the two genes. Sturtevant used vast amounts of recombination data from different crosses to predict the relative position of the genes on each chromosome and produced the first chromosome map in *Drosophila.* By working out the number of recombinants, it is possible to obtain a measurement for the distance between the genes. This distance is called a genetic map unit. Linkage mapping has been used to identify the location of many genes that cause genetic diseases.

After Morgan's results were published, Punnett used this information to identify linkage groups in his pea studies. Morgan, meanwhile, continued his productive research career and identified more than two dozen mutants. For this work, he was awarded the Nobel Prize in 1933.[13]

## How Is Sex Determined?

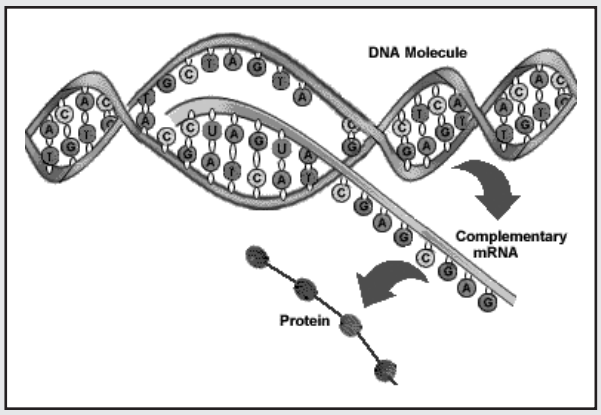
The factors involved in sex determination in animals are rather diverse. In most cases, sex determination is genetic, but in many species temperature, environmental chemicals, or social structure determines the sex of the embryo. Even the combinations of sex chromosomes are different in different species. In the fruit fly species that Morgan worked with, the number of X chromosomes determines the sex of the offspring: flies with two Xs are female and those with one X and one Y are male. Although the Y chromosome is present in male flies, the Y chromosome alone does not determine the sex of the individual.

In contrast, the presence of the Y chromosome in human beings determines the male sex because of an essential gene called SRY (sex determining region Y) that controls the male traits. The undifferentiated gonad in humans is destined to become the ovary (the default pathway) unless told otherwise. This command is stored in the SRY gene, which is located on the Y chromosome. SRY turns on other genes that will reverse the female-committed pathway to a male-bound pathway. Before sexual differentiation (around two months in gestation), the human embryo has a pair of gonads that will become the ovaries plus both sets (male and female) of reproductive tracts. In the absence of a Y chromosome, the ovaries develop, and the female sex hormones promote the development of the female reproductive tract and kill off the male tract. However, in the presence of SRY, which is found only on the Y chromosome, a set of genes that are essential for the development of the testes are turned on, and a cascade of events causes the maturation of the testes as well as the male internal and external genitalia. Sex determination in humans can be worked out using the Punnett square (Figure 35).[14]

## An X-linked Trait in Humans—Colorblindness

Human eye color, as indicated earlier, is a polygenic type of inheritance. However, color vision in humans

## Figure 53




**In DNA transcription, one strand of DNA is used as a template for transcription, using the DNA–RNA complimentary base-pairing rule and RNA polymerase as the builder.**

plementary base-pairing rule and RNA polymerase as the builder. An A in the DNA strand places a U on the growing RNA strand, and a T in DNA places an A on the RNA strand.

An example of transcription from DNA to mRNA is shown in Table 3. The top row represents the DNA sequence of a gene. The bottom row represents the transcriptional product mRNA.

Notice that RNA does not have T, so U is transcribed from A. The three major steps in the process of transcription are as follows:

1. **Initiation –** The promoter region upstream of the DNA sequence to be transcribed is bound by the RNA polymerase.
2. **Elongation –** A, U, G, or C is added based on the DNA sequence.
3. **Termination –** As the RNA grows, the completed segment separates from the DNA template.

Once the entire DNA sequence has been transcribed, RNA polymerase encounters the terminal sequence of the gene and dissociates from both the newly transcribed RNA and DNA. The entire RNA thus detaches, and in eukaryotes it is further modified before it matures and moves to the cytoplasm (Figure 54). The DNA then restores its double-stranded structure.

After transcription, several modifications must occur before the "mature" mRNA leaves the nucleus. To facilitate the transport of mRNA to its final destination and prevent it from degradation while in transit, extra nucleotides, called a cap, and a poly A tail (a long nucleotide consisting of A's) are added to the beginning and end of the mRNA, respectively. Another modification is the removal of the noncoding region that has been transcribed from the gene. Recall that a typical gene in the eukaryotic genomic structure has intervening noncoding regions called introns and coding regions called exons. The intron portion transcribed to mRNA must be excised, and the exon portions must be stitched together before the final mRNA product is usable (Figure 55). In addition, different exons can recombine in a variety of ways to create different mRNA transcripts, and ultimately different proteins. This is one reason that our cells can make many more proteins than there are genes in our genome.

## Translation and Post-translational Modification

Protein synthesis occurs on the ribosomes. Once mature mRNA reaches the ribosome, a translator that can read the genetic code and translate the DNA/RNA language into the language of protein is needed. This process of translation is carried out by **transfer RNA (tRNA)**. Translation can be divided into three stages (Figure 56):

- **Initiation –** The initiation of translation begins when mRNA, the first tRNA carrying the first amino acid, and the small and large subunits of the ribosome come together to form a complex. An mRNA first binds to a small subunit. This allows the first initiator tRNA carrying methionine with the anticodon UAC to bind to the start codon AUG on the mRNA. Next, a large ribosomal subunit binds to the smaller ribosomal subunit and creates a "docking" site (P site) on the ribosome for the first tRNA (with a methionine) to dock.
- **Elongation –** On the large ribosomal subunit, another docking site, the A site, is available for the binding of the next tRNA with the anticodon that matches the next codon on the mRNA molecule. The new amino acid carried by this tRNA forms a peptide bond with the first amino acid. The first tRNA then exits the large ribosome, and the second tRNA moves from the A site to the P site, making room for the next tRNA to enter. The elongation process continues, and a growing peptide chain starts to form.
- **Termination –** The elongation step continues until a stop codon is encountered, and the translation is terminated. The completed chain then dissociates from the ribosome, and the entire complex disassembles.

Proteins made in this way may or may not be ready to perform their job—in some cases, they are further modified before being delivered to their final destination. Most of these modifications occur as the protein travels through the endomembrane system of the endoplasmic reticulum, Golgi apparatus, or