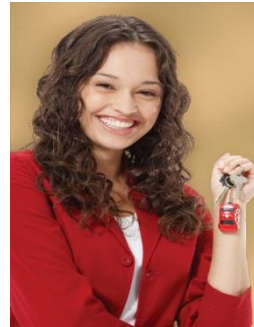
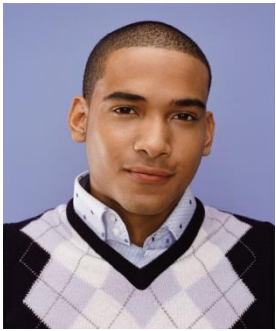


7.4 Human Genetics and Pedigrees

KEY CONCEPT

A combination of methods is used to study human genetics.



7.4 Human Genetics and Pedigrees

▶ A combination of methods is used to study human genetics

- The basic principles of genetics are the same in all sexually reproducing organisms.
 - Inheritance of many human traits is complex.
 - Single-gene traits are important in understanding human genetics.



7.4 Human Genetics and Pedigrees

▶ Females can carry sex-linked genetic disorders.

- Males (XY) express all of their sex linked genes.
- Males tend to show sex-linked disorders more than females

Chromosome X



Examples of known genes

- DMD
Duchenne's muscular dystrophy
- RP2
Retinitis pigmentosa
- DFN2
X-linked deafness
- FMRI
Fragile X syndrome
- OPN1MW
Deuteranopia (red-green colorblindness)
- F8
Hemophilia A

Chromosome Y



Examples of known genes

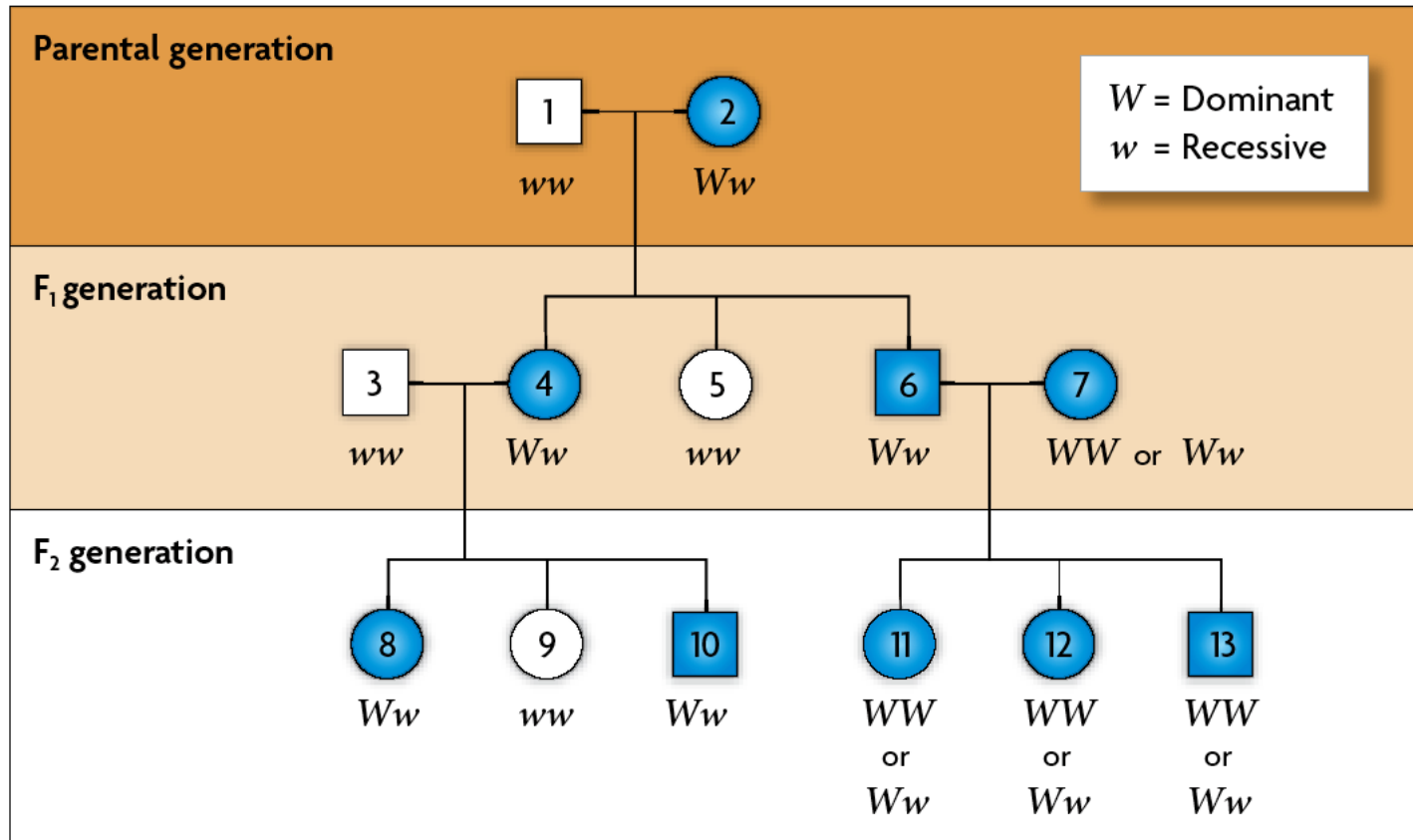
- SRY
Testes-determining factor
- TTY5
Testes-specific transcript



7.4 Human Genetics and Pedigrees

▶ A pedigree is a chart for tracing genes in a family.

- Phenotypes are used to infer genotypes on a pedigree.
- Autosomal genes show different patterns on a pedigree than sex-linked genes.



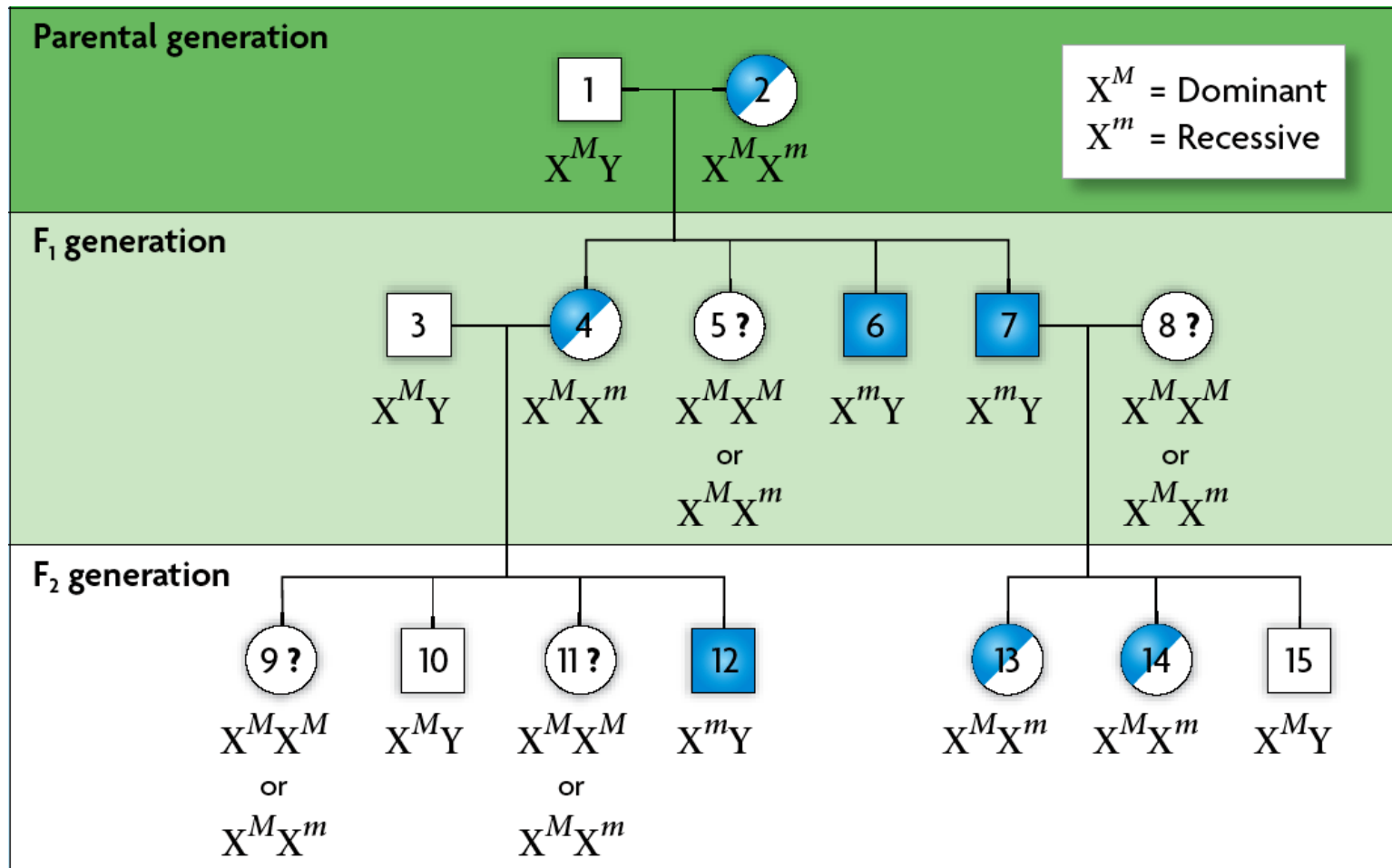
7.4 Human Genetics and Pedigrees

▶ How do we read a pedigree chart?

- □ = Males
- ○ = Females
- Horizontal lines between a male and female signify marriage
- Horizontal lines above males and females signify offspring
- Vertical lines signify children of a couple
- Colored in shapes mean the person has the genetic disorder
- Half colored in shape means the person is a carrier

7.4 Human Genetics and Pedigrees

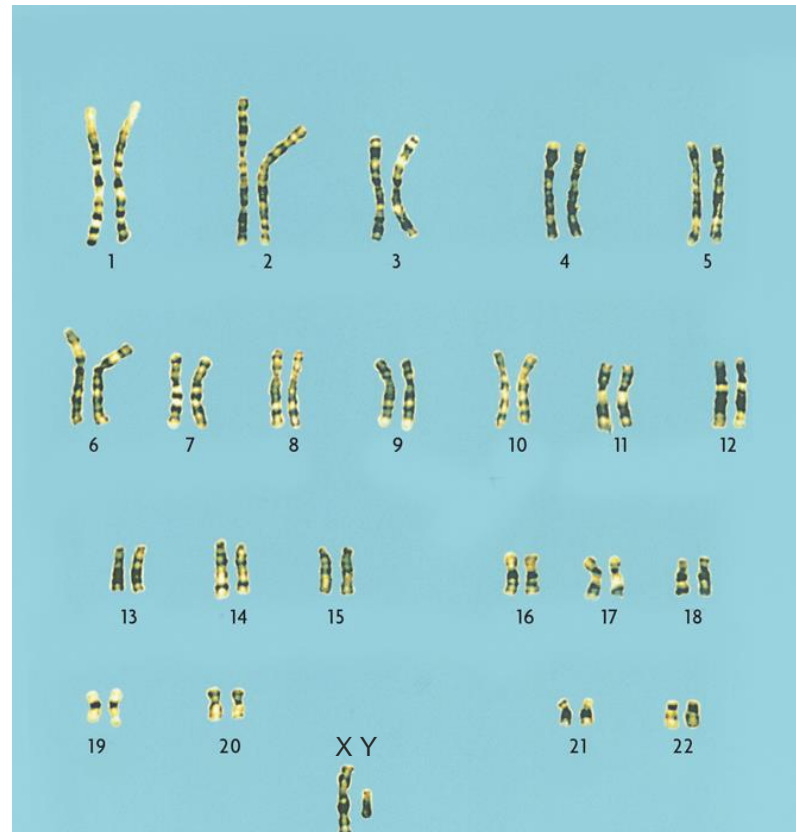
- If the phenotype is more common in males, the gene is likely sex-linked.



7.4 Human Genetics and Pedigrees

▶ Several methods help map human chromosomes.

- A karyotype is a picture of all chromosomes in a cell.



7.4 Human Genetics and Pedigrees

- Karyotypes can show changes in chromosomes.
 - deletion of part of a chromosome or loss of a chromosome
 - large changes in chromosomes
 - extra chromosomes or duplication of part of a chromosome

