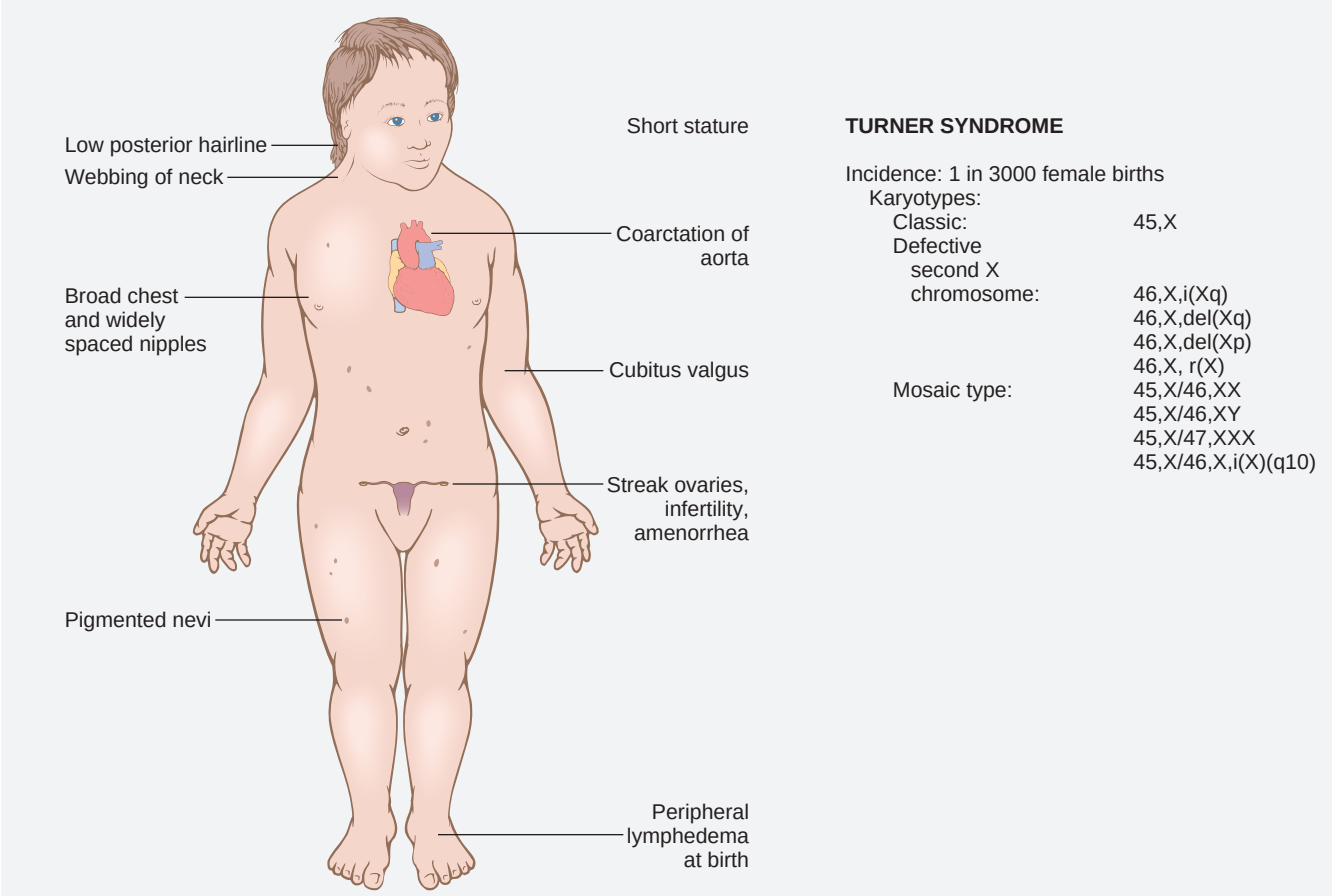

Figure 5-22 Clinical features and karyotypes of Turner syndrome.

features such as cardiac malformations and endocrine abnormalities. Clearly several other genes located on the X chromosome are also involved.

Hermaphroditism and Pseudohermaphroditism

The problem of sexual ambiguity is exceedingly complex, and only limited observations are possible here; for more details, reference should be made to specialized sources. It will be no surprise to medical students that the sex of an individual can be defined on several levels. *Genetic sex* is determined by the presence or absence of a Y chromosome. No matter how many X chromosomes are present, a single Y chromosome dictates testicular development and the genetic male gender. The initially indifferent gonads of both the male and the female embryos have an inherent tendency to feminize, unless influenced by Y chromosome-dependent masculinizing factors. *Gonadal sex* is based on the histologic characteristics of the gonads. *Ductal sex* depends on the presence of derivatives of the müllerian or wolffian ducts. *Phenotypic*, or *genital, sex* is based on the appearance of the external genitalia. Sexual ambiguity is present whenever there is disagreement among these various criteria for determining sex.

The term true *hermaphrodite* implies the presence of both ovarian and testicular tissue. In contrast, a pseudohermaphrodite represents a disagreement between the phenotypic and gonadal sex (i.e., a female pseudohermaphrodite has ovaries but male external genitalia; a male pseudohermaphrodite has testicular tissue but female-type genitalia). The genetic bases of these conditions are quite variable and beyond the scope of our discussion here.

KEY CONCEPTS

Cytogenetic Disorders Involving Sex Chromosomes

- In females, one X chromosome, maternal or paternal, is randomly inactivated during development (Lyon hypothesis).
- In **Klinefelter syndrome,** there are two or more X chromosomes with one Y chromosome as a result of nondisjunction of sex chromosomes. Patients have testicular atrophy, sterility, reduced body hair, gynecomastia, and eunuchoid body habitus. It is the most common cause of male sterility.
- In **Turner syndrome,** there is partial or complete monosomy of genes on the short arm of the X chromosome, most commonly due to absence of one X chromosome (45,X) and less commonly from mosaicism, or from deletions involving the short arm of the X chromosome. Short stature, webbing of the neck, cubitus valgus, cardiovascular malformations, amenorrhea, lack of secondary sex characteristics, and fibrotic ovaries are typical clinical features.