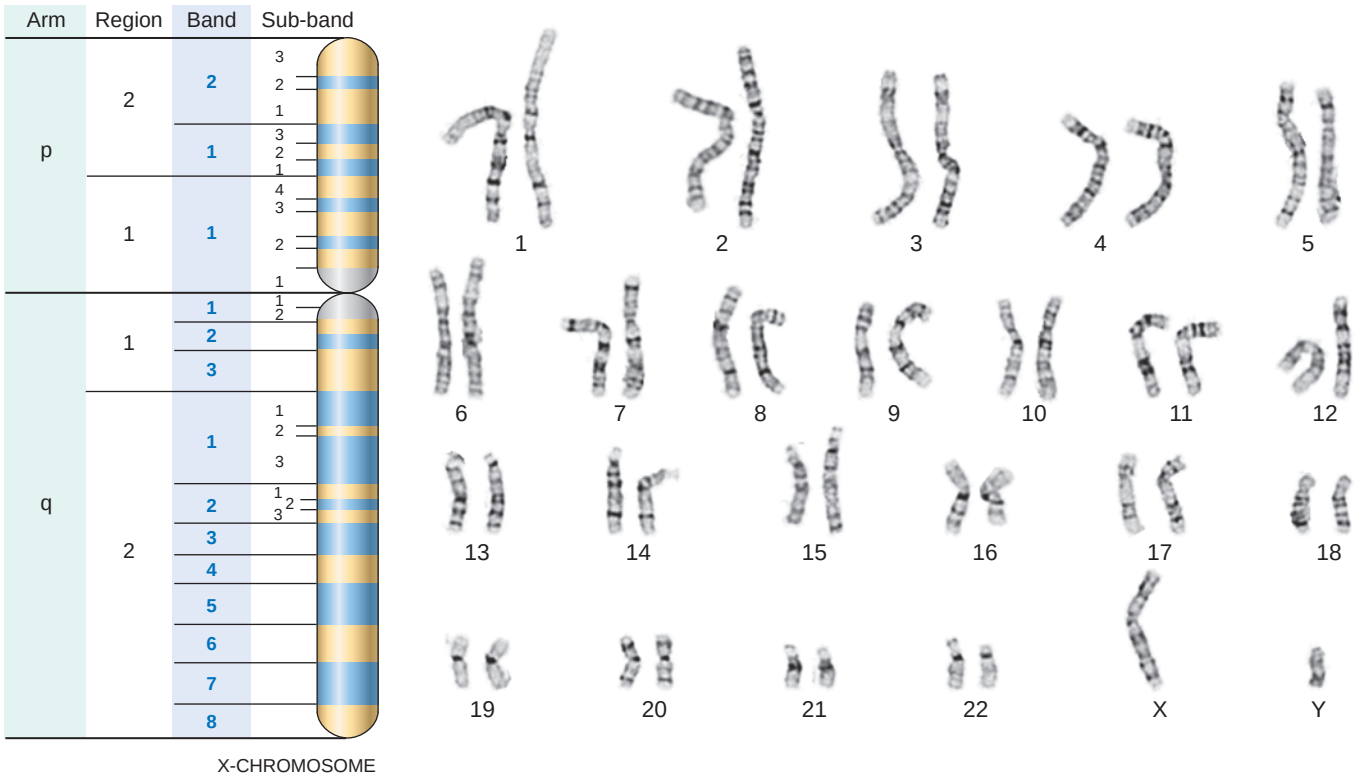

Figure 5-17 G-banded karyotype from a normal male (46,XY). Also shown is the banding pattern of the X-chromosome with nomenclature of arms, regions, bands, and sub-bands. (Courtesy of Dr. Stuart Schwartz, Department of Pathology, University of Chicago, Chicago, IL.)

per karyotype can be recognized. The use of these banding techniques permits certain identification of each chromosome and roughly delineates breakpoints and other gross alterations (described later).

Commonly Used Cytogenetic Terminology

Karyotypes are usually described using a shorthand system of notations in the following order: total number of chromosomes is given first, followed by the sex chromosome complement, and finally the description of abnormalities in ascending numerical order. For example, a male with trisomy 21 is designated *47,XY,+21*. Notations denoting structural alterations of chromosomes and their corresponding abnormalities are described later.

The short arm of a chromosome is designated *p* (for petit), and the long arm is referred to as *q* (the next letter of the alphabet). In a banded karyotype, each arm of the chromosome is divided into two or more regions bordered by prominent bands. The regions are numbered (e.g., 1, 2, 3) from the centromere outward. Each region is further subdivided into bands and sub-bands, and these are ordered numerically as well (Fig. 5-17). Thus, the notation *Xp21.2* refers to a chromosomal segment located on the short arm of the X chromosome, in region 2, band 1, and sub-band 2.

Structural Abnormalities of Chromosomes

The aberrations underlying cytogenetic disorders may take the form of an abnormal number of chromosomes or alterations in the structure of one or more chromosomes. The normal chromosome complement is expressed as 46,XX for the female and 46,XY for the male. Any exact multiple of the haploid number of chromosomes (23) is called *euploid*. If an error occurs in meiosis or mitosis and a cell acquires a chromosome complement that is not an exact multiple of 23, it is referred to as *aneuploidy*. The usual causes for aneuploidy are *nondisjunction* and *anaphase lag*. When nondisjunction occurs during gametogenesis, the gametes formed have either an extra chromosome (n + 1) or one less chromosome (n – 1). Fertilization of such gametes by normal gametes results in two types of zygotes—trisomic (2n + 1) or monosomic (2n – 1). In anaphase lag, one homologous chromosome in meiosis or one chromatid in mitosis lags behind and is left out of the cell nucleus. The result is one normal cell and one cell with monosomy. As seen subsequently, monosomy or trisomy involving the sex chromosomes, or even more bizarre aberrations, are compatible with life and are usually associated with variable degrees of phenotypic abnormalities. **Monosomy involving an autosome generally causes loss of too much genetic information to permit live birth or even embryogenesis, but several autosomal trisomies do permit survival.** With the exception of trisomy 21, all yield severely handicapped infants who almost invariably die at an early age.

Occasionally, *mitotic errors in early development give rise to two or more populations of cells with different chromosomal complement, in the same individual,* a condition referred to as *mosaicism*. Mosaicism can result from mitotic errors during the cleavage of the fertilized ovum or in somatic cells. Mosaicism affecting the sex chromosomes is relatively common. In the division of the fertilized ovum, an error may lead to one of the daughter cells receiving three sex