disabilities in children.

Down Syndrome

Down syndrome is the most common chromosomal abnormality of a generalized syndrome, occurring in 1 in 691 live births in the United States (National Down Syndrome Society, 2012c; Summar and Lee, 2011; Weijerman and de Winter, 2010). It occurs in people of all races and economic levels.

Etiology

The cause of Down syndrome is not known, but evidence from cytogenetic and epidemiologic studies supports the concept of multiple causality. Although the cause is unclear, the cytogenetics of the disorder is well established. Approximately 95% of all cases of Down syndrome are attributable to an extra chromosome 21 (group G), hence the name **nonfamilial trisomy 21**. Although children with trisomy 21 are born to parents of all ages, there is a statistically greater risk in older women, particularly those older than 35 years of age. For example, in women 35 years old, the chance of conceiving a child with Down syndrome is about 1 in 350 live births; but in women 40 years old, it is about 1 in 100. However, the majority (≈80%) of infants with Down syndrome are born to women younger than 35 years old, because younger women have higher fertility rates (National Down Syndrome Society, 2012c; Summar and Lee, 2011). About 4% of the cases may be caused by **translocation** of chromosomes 15 and 21 or 22. This type of genetic aberration is usually hereditary and is not associated with advanced parental age. About 1% of affected persons demonstrate **mosaicism**, which refers to a mixture of normal and abnormal chromosomes in the cells. The degree of cognitive and physical impairment is related to the percentage of cells with the abnormal chromosome makeup.

Diagnostic Evaluation

Down syndrome can usually be diagnosed by the clinical manifestations alone (Box 18-2 and Fig. 18-6), but a chromosome analysis should be done to confirm the genetic abnormality.