

children, however, these features may be less obvious, and behavioral manifestations may initially suggest the diagnosis (Box 18-3). In carrier females, the clinical manifestations are extremely varied.

Box 18-3

Clinical Manifestations of Fragile X Syndrome

Physical Features

Increased head circumference

Long, wide, or protruding ears

Long, narrow face with prominent jaw

Strabismus

Mitral valve prolapse, aortic root dilation

Hypotonia

In postpubertal males, enlarged testicles

Behavioral Features

Mild to severe cognitive impairment (CI)

Speech delay; may be rapid speech with stuttering and word repetition

Short attention span, hyperactivity

Hypersensitivity to taste, sounds, touch

Intolerance to change in routine

Autistic-like behaviors, such as social anxiety and gaze aversion

Possible aggressive behavior