

Clinical Manifestations of Down Syndrome The clinical diagnosis of Down syndrome is not difficult for experienced physicians due to the characteristic gestalt of these patients. Patients are usually identified at birth or shortly thereafter. However, the diagnosis may be challenging in premature babies, some older patients, certain ethnic groups, and in mosaicism. Each individual with Down syndrome has different healthcare needs. Key diagnostic features are the distinctive physical appearance, poor growth and developmental delay. The signs and symptoms may be variable.

Physical Appearance Individuals with Down syndrome can have the following physical features– & Brachycephaly with flat occiput, wide open fontanel & Flat facial profile*, flat nasal bridge & Protruding tongue, small mouth & Dysplastic*, small, low set ears & Upward slant of palpebral fissures*, epicanthic folds, squint, speckled iris, palebrae ‘purse’ on laughing or crying & Short and broad neck, abundant neck skin* & Short and broad hands, short and broad fingers, small middle phalanx of 5th finger* (clinodactyly), simian crease (single palmer crease) & Increased space between 1 and 2 toes (sandal gap) & Hypotonia*, hyper-extensibility/hyper-flexibility*, lack of Moro reflex* Features marked with asterisks (*) are useful for making a diagnosis in the newborn. Figure 1 shows the important features of Down syndrome.

Growth Children with Down syndrome generally have low birth weight, and poor growth velocity especially during the initial years, partly contributed by feeding problems due to hypotonia and a small oral cavity or due to the co-morbid conditions such as cardiovascular problems and/or other gastrointestinal problems. Thereafter, the tendency towards development of obesity increases with age and is quite common amongst adults with Down syndrome. The factors responsible for obesity include associated hypothyroidism, high leptin levels, and poor basal metabolic rate. Though the Indian growth charts are not available, growth monitoring can be done through the currently available Western Down syndrome growth charts [1]. Regular growth monitoring in the initial years and then annually throughout childhood would be helpful in early identification of under nutrition and obesity in these children.

Characteristic facial features in children with Down syndrome at different ages. Upper panel shows the characteristic flat facies with upslant of eyes, open mouth appearance and protruded tongue. Bottom panel shows the short hand and fingers, clinodactyly (solid arrow), simian crease (arrow head) and sandal gap (dotted arrow)