Turner syndrome is a random mutation of chromosomes that affects only females. There is an absence of all or part of the second X chromosomes in some or all of the cells in the body. If a child does not exhibit classic TS characteristics, but is short and falling below the curve on the growth chart, has delayed pubertal development, presents verbally gifted, or expresses academic or social challenges, she may have TS. Diagnosis is made with a Karyotype blood test. An endocrinologist will monitor the patient, screen for other conditions and provide specialized medical care, and treatments. Early interventions can promote a positive self-esteem and improved prognosis.

Medical evidence indicates that early diagnosis and treatment is essential.