

Williams syndrome, also known as Williams-Beuren syndrome, is a rare genetic disorder characterized by growth delays before and after birth (prenatal and postnatal growth retardation), short stature, a varying degree of mental deficiency, and distinctive facial features that typically become more pronounced with age. Such characteristic facial features may include a round face, full cheeks, thick lips, a large mouth that is usually held open, and a broad nasal bridge with nostrils that flare forward (anteverted nares). Affected individuals may also have unusually short eyelid folds (palpebral fissures), flared eyebrows, a small lower jaw (mandible), and prominent ears. Dental abnormalities may also occur including abnormally small, underdeveloped teeth (hypodontia) with small, slender roots. Williams syndrome is a rare disorder that affects males and females in equal numbers and infants of any race may be affected. The prevalence of this disorder is approximately one in 10,000-20,000 births in the United States. The diagnosis of Williams syndrome may be confirmed by a thorough clinical evaluation that includes a detailed patient history and specialized blood tests that may detect elevated levels of calcium in the blood. Another test, known as fluorescent in situ hybridization [FISH], may be used to determine whether a deletion of one elastin gene on chromosome 7 is present. This deletion is believed to occur in the majority of individuals with Williams Syndrome.