

"Freeman-Sheldon syndrome is a rare inherited disorder characterized by multiple contractures (i.e., restricted movement around two or more body areas) at birth (congenital), abnormalities of the head and face (craniofacial) area, defects of the hands and feet, and skeletal malformations. Craniofacial abnormalities may consist of characteristic facial features that cause the individual to appear to be whistling. These features include an extremely small puckered mouth (microstomia); a "full" forehead appearance, unusually prominent cheeks; and thin, pursed lips. Affected infants may also have an unusually flat middle portion of the face, a high roof of the mouth (palate), an unusually small jaw (micrognathia), an abnormally small tongue (microglossia), and/or a raised, scar-like mark in the shape of an "H" or a "V" extending from the lower lip to the chin. Affected infants often have abnormalities affecting the eyes including widely-spaced deep-set eyes, crossed eyes (strabismus), and/or downslanting eyelid folds (palpebral fissures). Malformations of the hands and feet are also characteristic of Freeman-Sheldon syndrome. Children with Freeman-Sheldon syndrome may also exhibit speech impairment; swallowing and eating difficulties; vomiting; failure to grow and gain weight at the expected rate (failure to thrive); and/or respiratory problems that may result in life-threatening complications. Freeman-Sheldon syndrome can be inherited as an autosomal dominant genetic trait. However, most cases occur randomly with no apparent cause (sporadically). Freeman-Sheldon syndrome is a rare disorder that affects males and females in equal numbers. Approximately 100 cases have been reported in the medical literature since the disorder was first described in 1938 by Drs. Freeman and Sheldon. Some cases occurred within families (kindreds) over several generations. Some symptoms and physical findings associated with the disorder are apparent at birth (congenital). Freeman-Sheldon syndrome is one of a group of disorders that are associated with multiple congenital contractures (MCCs). MCCs occur in approximately 1 in 3,000 children. Because the disorder shares features with other congenital contracture syndromes it sometimes is misdiagnosed making it difficult to determine its true frequency in the general population."