Freeman-Sheldon syndrome: report of 6 cases
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Abstract
Freeman-Sheldon syndrome is a morphologically well-defined syndrome that results in a dysmorphic status combining bone anomalies and joint contractures with characteristic facies. FSS (Freeman-Sheldon Syndrome) is also known as craniocarpotarsal dysplasia (or dystrophy), distal arthrogryposis type IIA (DAIIA), whistling face syndrome, and whistling face-windmill vane hand syndrome. The syndrome is an autosomal dominant trait and characterized by flattened, mask-like facies, microstomia, protruding lips (as in whistling), deep-set eyes with hypertelorism, and camptodactyly with ulnar deviation of the fingers and talipes equinovarus. We are reporting 6 cases with this syndrome that were referred to our genetic center from 2000 to 2006 for cytogenetic study and clinical genetic counseling.

Key words: Freeman-Sheldon syndrome; Distal arthrogryposis; Whistling face

Introduction
Craniocarpotarsal dysplasia was first described by Ernest Arthur Freeman (British orthopedic surgeon, 1900-1975) and Joseph Harold Sheldon (British physician, 1920-1964) in 1938 (1), including certain skeletal malformations with facial characteristics. At first it was understood that the syndrome was very rare, but up until 1990, sixty-five cases have been reported (2). Both sexes are affected equally. The intelligence of patients is usually normal, though there are occasional reports of association with mental retardation, especially in cases with combined prominent structural anomalies of the central nervous system.

Freeman-Sheldon Syndrome is considered a form of distal arthrogryposis (DA) with additional features. Distal arthrogryposis without additional features is known as DA Type 1 (DA1). Extended classification of distal arthrogryposis syndromes are listed in table 1.

DA2A or Freeman-Sheldon syndrome is a rare