Freeman-Sheldon Syndrome

National Cancer Institute

Source

National Cancer Institute. Freeman-Sheldon Syndrome. NCI Thesaurus. Code C98931.

A rare syndrome that is inherited in an autosomal dominant or recessive pattern and caused by mutations in the MYH3 gene. It is a severe form of arthrogryposis. It is characterized by the presence of distinctive facial features (small mouth, midface hypoplasia, short nose, drooping of the eyelids, deep folds in the area between the nose and the lips, and strabismus), joint deformities that lead to permanently bent fingers and toes, club foot, scoliosis, and walking difficulties.