Congenital contractural arachnodactly (Beals syndrome)

Genetics

-FBN2 (Fibrillin 2) is only gene known

-AD; mostly inherited; some de novo

Clinical findings/Dysmorphic features

-Marfanoid appearance; long slender fingers/toes; crumpled ears; contractures of major joints (knees and ankles) at birth; muscle hypoplasia; kyphosis/scoliosis; severe/lethal: aortic dilation

Etiology

-Prevalence lower than Marfan syndrome

Pathogenesis

-Fibrillin 2 is a glycoprotein of the extracellular matrix microfibrils --> co-distributed with fibrillin 1 in many tissues; precise function is not known

Genetic testing/diagnosis

-FBN2 sequencing (75%)