Congenital contractural arachnodactyly

Description

Congenital contractural arachnodactyly is a disorder that affects many parts of the body. People with this condition typically are tall with long limbs (dolichostenomelia) and long, slender fingers and toes (arachnodactyly). They often have permanently bent joints (contractures) that can restrict movement in their hips, knees, ankles, or elbows. Additional features of congenital contractural arachnodactyly include underdeveloped muscles, a rounded upper back that also curves to the side (kyphoscoliosis), permanently bent fingers and toes (camptodactyly), ears that look "crumpled," and a protruding chest (pectus carinatum). Rarely, people with congenital contractural arachnodactyly have heart defects such as an enlargement of the blood vessel that distributes blood from the heart to the rest of the body (aortic root dilatation) or a leak in one of the valves that control blood flow through the heart (mitral valve prolapse). The life expectancy of individuals with congenital contractural arachnodactyly varies depending on the severity of symptoms but is typically not shortened.

A rare, severe form of congenital contractural arachnodactyly involves both heart and digestive system abnormalities in addition to the skeletal features described above; individuals with this severe form of the condition usually do not live past infancy.

Frequency

The prevalence of congenital contractural arachnodactyly is estimated to be less than 1 in 10,000 worldwide.

Causes

Mutations in the *FBN2* gene cause congenital contractural arachnodactyly. The *FBN2* gene provides instructions for producing the fibrillin-2 protein. Fibrillin-2 binds to other proteins and molecules to form threadlike filaments called microfibrils. Microfibrils become part of the fibers that provide strength and flexibility to connective tissue that supports the body's joints and organs. Additionally, microfibrils regulate the activity of molecules called growth factors. Growth factors enable the growth and repair of tissues throughout the body.

Mutations in the *FBN2* gene can decrease fibrillin-2 production or result in the production of a protein with impaired function. As a result, microfibril formation is

reduced, which probably weakens the structure of connective tissue and disrupts regulation of growth factor activity. The resulting abnormalities of connective tissue underlie the signs and symptoms of congenital contractural arachnodactyly.

Learn more about the gene associated with Congenital contractural arachnodactyly

- FBN2

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Arthrogyroposis, distal, type 9
- Beals syndrome
- Beals-Hecht syndrome
- CCA
- Contractural arachnodactyly, congenital
- DA9
- Distal arthrogyropsis type 9

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Congenital contractural arachnodactyly (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0220668/)

Genetic and Rare Diseases Information Center

- Congenital contractural arachnodactyly (https://rarediseases.info.nih.gov/diseases/5899/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Congenital contractural

arachnodactyly%22)

Catalog of Genes and Diseases from OMIM

- CONTRACTURAL ARACHNODACTYLY, CONGENITAL; CCA (https://omim.org/entry/121050)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28congenital+contractural+arachnodactyly%5BTIAB%5D%29+OR+%28beals+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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