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Causes, Signs and Symptoms of Congenital Contractural Arachnodactyly

Sonat Cehn*
Department of Medicine, University of Chicago, Chicago, USA

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Description

Beals-Hecht syndrome, also known as Congenital Contractural Arachnodactyly (CCA), is a rare autosomal dominant connective tissue condition. Similar to Marfan syndrome, patients with CCA frequently have extraordinarily large fingers and toes and arms that are longer than they are tall. A disease called congenital contractural arachnodactyly affects several body parts. Individuals with this syndrome often have long arms, long fingers, and long slender toes (dolichostenomelia) (arachnodactyly). They frequently have contractures which are permanently bent joints which can limit movement in their hips, knees, ankles, or elbows. Underdeveloped muscles a rounded upper back that also curves to the side (kyphoscoliosis), permanently bent fingers and toes (camptodactyly), "crumpled"-looking ears, and a projecting chest (pectus) are further characteristics of congenital contractural arachnodactyly. Rarely, those born with congenital contractural arachnodactyly develop heart conditions such aortic root dilatation, which is an expansion of the blood vessel that transports blood from the heart to the rest of the body or a leak in one of the valves that regulate blood flow through the heart (mitral valve prolapse). Depending on the severity of their symptoms people with congenital contractural arachnodactyly can expect to have longer or shorter lives. In addition to the skeletal characteristics mentioned above a rare severe type of congenital contractural arachnodactyly also involves heart and digestive system problems in people with this severe version of the condition typically do not survive through infancy.

Causes

Congenital contractural arachnodactyly may originate from fresh mutations in the 5q23-located Fibrillin-2 (FBN2) gene or it may be inherited from a parent in an autosomal dominant pattern, meaning that just one copy of the changed gene in each cell is enough to cause the condition. Congenital contractural arachnodactyly is brought on by mutations in the FBN2 gene. The fibrillin-2 protein is made according to instructions from the FBN2 gene. Microfibrils are threadlike filaments that are created when fibrillin-2 interacts to additional proteins and chemicals. Microfibrils become a component of the fibres that give connective tissue, which supports the body's joints and organs, strength and flexibility. Microfibrils also control the activity of chemicals known as growth factors. Growth factors allow all physiological tissues to expand and repair themselves. A protein with a compromised function or less fibrillin-2 can be produced as a result of mutations in the FBN2 gene. As a result, there is a decrease in microfibril synthesis, which likely impairs connective tissue integrity and messes up growth factor activity control. Congenital contractural arachnodactyly has symptoms and signs that are caused by faulty connective tissue.

Signs and symptoms

Despite the two disorders having different causes, the signs and symptoms of CCA frequently match those of Marfan syndrome. The key big joint contractures that characterise CCA are present in all affected children at birth and range in severity. Even if the contractures are modest, they usually go better with time. However, camptodactyly, or permanently bent fingers and toes, is virtually always present. People with CCA generally have ears that look crumpled (a significant Marfan syndrome differentiating trait), joint stiffness, underdeveloped muscles (muscular hypoplasia), and they may have curved spines in addition to having long fingers and toes and a tall, slender physique (congenital kyphoscoliosis). If kyphoscoliosis is present, it frequently gets worse with time and can need surgery. The blood channel known as the aorta, which transports blood from the heart to the rest of the body, may occasionally become excessively enlarged (aortic root dilatation.

Contact: Sonat Cehn, E-mail: Sonatcehn@gmail.com