BEALS SYNDROME

Beals syndrome is a disorder of connective tissue. The syndrome was first explained by Beals and Hecht in 1971. Features of Beals syndrome are found throughout the body, especially in large joints.

The vast majority of people with Beals syndrome have isolated features in the musculoskeletal system that often improve with age.

What other names do people use for Beals syndrome?

Beals syndrome is also referred to as Congenital Contractural Arachnodactyly (CCA).

How prevalent is Beals syndrome?

There is no information on the exact prevalence of Beals syndrome; however, it is estimated that the incidence (number of new cases within a given time) of Beals syndrome is less than 1 in 10,000 people per year. Beals syndrome affects males and females of all ethnicities.

What are the characteristics of Beals syndrome?

Beals syndrome shares some features with Marfan syndrome. A person with Beals syndrome may have long, thin limbs and long fingers and toes. Most affected people have a folding of the upper ear, also known as crumpled ears, and permanent bending (flexion contractures) of major joints, such as the knees, hips, and elbows. Additionally, most people with Beals syndrome have bent fingers and/or toes (campodactyly). Kyphosis and scoliosis (front to back and side to side curvature of the spine, respectively) is present in about half of the people with Beals syndrome.

Some people with Beals syndrome have a thickening and leakage of the mitral valve of the heart. Very rarely, people with Beals syndrome have more significant cardiovascular (heart and blood vessel) issues, such as congenital (at birth) problems with the structure of the heart (congenital heart disease) or aortic root enlargement. The severity of symptoms of Beals syndrome vary, even among family members.

What causes Beals syndrome?

Beals syndrome is caused by a change (mutation) in the fibrillin-2 gene (FBN2). The fibrillin-2 gene provides instructions on how to make the fibrillin-2 protein. Fibrillin-2, which binds to other proteins, helps form microfibrils. Microfibrils are an important part of connective tissue that provides strength and flexibility. Microfibrils interact with growth factors. These growth factors regulate how cells perform in different parts of the body. When the fibrillin-2 gene has a defect, the fibrillin-2 protein cannot make good quality microfibrils, and sometimes cannot make the amount of microfibrils that the body needs. This defect is what causes many of the physical features of Beals syndrome.

People can inherit Beals syndrome, meaning they get the mutation from a parent who also has the disorder. Since it is an autosomal dominantly inherited disorder, only one parent needs to have a change in the gene that causes Beals syndrome in order to pass it on.

Other people have a spontaneous mutation, meaning that they are the first in their family to have Beals syndrome.

People with Beals syndrome have a 50 percent chance of passing the mutation on each time they have a child.

How is Beals syndrome diagnosed?

Beals syndrome is diagnosed based on the presence of a group of specific features:

- Long, thin limbs (dilichostenomelia), narrow head and body
- Permanent bending (flexion contractures) of multiple joints that restrict movement of the elbows, knees, hips, and fingers
- Abnormal bending of the spine that causes a bowing of the back (kyphoscoliosis)
- Underdeveloped muscles (muscular hypoplasia)
- Abnormalities of the ear (abnormal pinnae), such as crumpled ears

Other features include:

- Long, slender fingers and toes (arachnodactyly)
- Permanently bent fingers and toes that cannot completely straighten (camptodactyly)
- Protruding chest bone (pectus carinatum)
- Mitral valve prolapse

- Mitral valve regurgitation
- Aortic dilation

Genetic testing for FBN2 is available. When there is a known disease-causing gene mutation in the family, prenatal testing is available for pregnancies that are at risk for passing on the FBN2 gene mutation.

Genetic counseling is also available. Genetic counseling can be helpful in family planning, determining genetic risk, and discussing the availability of prenatal testing.

How is Beals syndrome managed?

Management of Beals syndrome depends on the severity of the features. Several exams can determine the severity of Beals syndrome:

- Echocardiogram by a cardiologist
- Musculoskeletal examination for the presence of contractures and kyphosis/scoliosis by an orthopedist
- Medical genetics consultation

An echocardiogram is recommended every two years until it is clear that the aorta is not involved. A physical exam, at least yearly, is also recommended to check for kyphosis and scoliosis.

The treatment for Beals syndrome is different for each person because treatment is dependent on the features. It is important that anyone diagnosed with Beals syndrome has a team of doctors to help monitor the syndrome. These doctors can include pediatricians, surgeons, cardiologists, orthopedists, ophthalmologists, and other healthcare professionals.

People with Beals syndrome benefit from physical therapy that can improve mobility of joints and strengthen the muscles.

Surgical procedures may be necessary to release joints and other parts of the body that are in a permanently bent position (flexion contractures). Sometimes, braces and surgical correction are used to provide stability when kyphoscoliosis or scoliosis is present.

What is the life expectancy for someone with Beals syndrome?

Beals syndrome does not impact life expectancy.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.