Buschke-Ollendorff Syndrome

Subjects: Genetics & Heredity
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Buschke-Ollendorff syndrome is a hereditary disorder that primarily affects the skin and bones. Specifically, the condition is characterized by skin growths called connective tissue nevi and bone abnormalities, most commonly a pattern of increased bone density called osteopoikilosis. Buschke-Ollendorff syndrome is classified as a disorder of connective tissues, which provide support, strength, and flexibility to organs and tissues throughout the body.

genetic conditions

1. Introduction

Connective tissue nevi are small, noncancerous lumps on the skin. They tend to appear in childhood and are widespread in people with Buschke-Ollendorff syndrome. In some cases, the nevi are subtle and hard to feel. The most common form of these nevi are elastomas, which are made up of a type of stretchy connective tissue called elastic fibers. Less commonly, affected individuals have nevi called collagenomas, which are made up of another type of connective tissue called collagen.

Osteopoikilosis, which is from the Greek words for "spotted bones," refers to small, round areas of increased bone density that appear as bright spots on x-rays. Osteopoikilosis usually occurs near the ends of the long bones of the arms and legs, and in the bones of the hands, feet, and pelvis. The areas of increased bone density appear during childhood. They do not cause pain or other health problems.

Other bone abnormalities can also occur with Buschke-Ollendorff syndrome, although they are less common. For example, a small percentage of affected individuals have melorheostosis, which is characterized by excess bone growth on the surface of existing bones in a pattern resembling dripping candle wax. Melorheostosis usually affects the bones in one arm or leg, although it can also affect bones in other areas of the body. This abnormality can cause long-lasting (chronic) pain, permanent joint deformities (contractures), and a limited range of motion of the affected body part.

2. Frequency

Buschke-Ollendorff syndrome has an estimated incidence of 1 in 20,000 people worldwide.

3. Causes

Buschke-Ollendorff syndrome results from mutations in the *LEMD3* gene. This gene provides instructions for making a protein that helps control signaling through two chemical pathways known as the bone morphogenic protein (BMP) and transforming growth factor-beta (TGF-β) pathways. These signaling pathways regulate various cell functions and are involved in the growth of cells, including new bone cells.

Mutations in the *LEMD3* gene reduce the amount of functional LEMD3 protein that is produced. A shortage of this protein increases signaling through the BMP and TGF-β pathways. Studies suggest that the enhanced signaling increases the formation of bone tissue, resulting in areas of overly dense bone or excess bone growth. It is unclear how the increased signaling is related to the development of connective tissue nevi in people with Buschke-Ollendorff syndrome.

3.1. The Gene Associated with Buschke-Ollendorff Syndrome

- LEMD3

4. 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. In many cases, an affected person has a parent and other family members with the condition. While most people with Buschke-Ollendorff syndrome have both skin and bone abnormalities, some affected families include individuals who have the skin abnormalities alone or the bone abnormalities alone.

5. Other Names for This Condition

- BOS
- dermatofibrosis disseminata lenticularis
- dermatofibrosis lenticularis disseminata
- dermatofibrosis lenticularis disseminata with osteopoikilosis
- dermatofibrosis, disseminated, with osteopoikilosis
- dermatoosteopoikilosis
- osteopathia condensans disseminata

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