Opinion
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Benign Scapuloperoneal Muscular Dystrophy with Cardiomyopathy

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Opinion

Benign scapuloperoneal muscular dystrophy with cardiomyopathy which is also known as Emery-Dreifuss Muscular Dystrophy Type 2 (EDMD 2) is a genetic condition characterized by joint contractures, muscle weakness, and heart issues. These symptoms usually begin early in childhood. The joint contractures restrict the movement of the joints, most often in the elbows, ankles, and neck. Muscle weakness usually occurs in the arms, shoulders, and hips. Problems with the heart typically begin to occur in adulthood when an individual is over 20 years old. Problems may include poor exercise tolerance, palpitations, and eventual heart failure.

EDMD 2 is a genetic condition caused by changes in the *LMNA* gene. We inherit our genes in pairs, one from each parent typically. EDMD 2 is inherited in an autosomal dominant manner. Autosomal dominant means an individual only needs one copy of the changed gene that causes the condition.

EDMD2 can be diagnosed when all three symptoms are present with the addition of genetic testing and muscular imaging performed. There is no way to prevent EDMD2, but there are ways to help symptoms. Treatment varies depending on how severe the symptoms are, but it can involve physical therapy to strengthen muscles, surgeries of the joints, and medication for heart problems.

Emery-Dreifuss muscular dystrophy is a condition that primarily affects muscles used for movement and the heart. Among the earliest features of this disorder are joint deformities called contractures. Contractures restrict the movement of certain joints, ankles, and neck, most often the elbows, and usually become noticeable in early childhood. Most affected individuals also experience muscle weakness and wasting that worsen slowly over time, beginning in muscles of the upper arms and lower legs and later also affecting muscles in the shoulders and hips.

People with Emery-Dreifuss muscular dystrophy develop heart problems by adulthood. In many cases, these heart problems are abnormalities of the electrical signals that control the heartbeat and abnormal heart rhythms. If untreated, these abnormalities can lead to a sensation of fluttering or pounding in the chest an unusually slow heartbeat, fainting, heart failure, and an increased risk of sudden death. The overall prevalence of Emery-Dreifuss muscular dystrophy is unknown. The prevalence of the autosomal dominant type is unknown, although it appears to be more common than the X-linked type. The autosomal recessive type appears to be very rare; only a few cases have been reported worldwide. Mutations in several genes, including *FHL1, EMD* and *LMNA*, can cause Emery-Dreifuss muscular dystrophy. Mutations in the *EMD* gene or, less commonly, in the *FHL1* gene cause the X-linked type of the condition. Mutations in the *LMNA* gene cause both the autosomal dominant and autosomal recessive types of the condition.

The genes associated with Emery-Dreifuss muscular dystrophy appear to be essential for the normal function of skeletal and cardiac muscle. The *EMD* and *LMNA* genes provide instructions for making proteins that are components of the nuclear envelope, which surrounds the nucleus in cells. The nuclear envelope regulates the movement of molecules into and out of the nucleus, and researchers believe it may play a role in regulating the activity of certain genes. The protein produced from the *FHL1* gene appears to be involved in other muscle cell functions, including maintaining the structure of these cells, chemical signaling and influencing muscle growth and size.

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