X-linked Dilated Cardiomyopathy

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X-linked dilated cardiomyopathy is a form of heart disease.

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1. Introduction

X-linked dilated cardiomyopathy is a form of heart disease. Dilated cardiomyopathy enlarges and weakens the heart (cardiac) muscle, preventing the heart from pumping blood efficiently. Signs and symptoms of this condition can include an irregular heartbeat (arrhythmia), shortness of breath, extreme tiredness (fatigue), and swelling of the legs and feet. In males with X-linked dilated cardiomyopathy, heart problems usually develop early in life and worsen quickly, leading to heart failure in adolescence or early adulthood. In affected females, the condition appears later in life and worsens more slowly.

X-linked dilated cardiomyopathy is part of a spectrum of related conditions caused by mutations in the *DMD* gene. The other conditions in the spectrum, Duchenne and Becker muscular dystrophy, are characterized by progressive weakness and wasting of muscles used for movement (skeletal muscles) in addition to heart disease. People with X-linked dilated cardiomyopathy typically do not have any skeletal muscle weakness or wasting, although they may have subtle changes in their skeletal muscle cells that are detectable through laboratory testing. Based on these skeletal muscle changes, X-linked dilated cardiomyopathy is sometimes classified as subclinical Becker muscular dystrophy.

2. Frequency

X-linked dilated cardiomyopathy appears to be an uncommon condition, although its prevalence is unknown.

3. Causes

X-linked dilated cardiomyopathy results from mutations in the *DMD* gene. This gene provides instructions for making a protein called dystrophin, which helps stabilize and protect muscle fibers and may play a role in chemical signaling within cells. The mutations responsible for X-linked dilated cardiomyopathy preferentially affect the activity of dystrophin in cardiac muscle cells. As a result of these mutations, affected individuals typically have little or no functional dystrophin in the heart. Without enough of this protein, cardiac muscle cells become damaged as the heart muscle repeatedly contracts and relaxes. The damaged muscle cells weaken and die over time, leading to the heart problems characteristic of X-linked dilated cardiomyopathy.

The mutations that cause X-linked dilated cardiomyopathy often lead to reduced amounts of dystrophin in skeletal muscle cells. However, enough of this protein is present to prevent weakness and wasting of the skeletal muscles.

Because X-linked dilated cardiomyopathy results from a shortage of dystrophin, it is classified as a dystrophinopathy.

3.1 The gene associated with X-linked dilated cardiomyopathy

- DMD

4. Inheritance

As its name suggests, X-linked dilated cardiomyopathy has an X-linked pattern of inheritance. The *DMD* gene is located on the X chromosome, which is one of the two sex chromosomes. In females (who have two X chromosomes), a mutation in one of the two copies of the gene in each cell usually leads to relatively mild heart disease that appears later in life. In

males (who have only one X chromosome), a mutation in the only copy of the gene in each cell causes more severe signs and symptoms that occur earlier in life. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

5. Other Names for This Condition

- CMD3B
- dilated cardiomyopathy 3B
- DMD-associated dilated cardiomyopathy
- DMD-related dilated cardiomyopathy
- XLCM
- XLDC

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