ANK2 Gene

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ankyrin 2. The ANK2 gene provides instructions for making a protein called ankyrin-B.

Keywords: genes

1. Normal Function

Ankyrin-B is part of a family of ankyrin proteins, which interact with many other types of proteins in cells throughout the body. Ankyrins help organize the cell's structural framework (the cytoskeleton) and link certain proteins that span the cell membrane to this framework. Additionally, ankyrins play key roles in important functions including cell movement (migration) and cell growth and division (proliferation).

The ankyrin-B protein is active in many cell types, particularly in the brain and in heart (cardiac) muscle. This protein mainly interacts with ion channels and ion transporters, which are complexes of proteins that move charged atoms (ions) across cell membranes. In the heart, the flow of ions (such as sodium, potassium, and calcium) through ion channels and ion transporters generates the electrical signals that control the heartbeat and maintain a normal heart rhythm. Ankyrin-B ensures these channels and transporters are in their proper locations in the cell membrane so they can regulate the flow of ions into and out of cardiac muscle cells. In addition, ankyrin-B helps ensure that signaling molecules that regulate the activity of ion channels and ion transporters are in the proper location.

2. Health Conditions Related to Genetic Changes

2.1. Ankyrin-B syndrome

At least ten mutations in the *ANK2* gene have been found to cause ankyrin-B syndrome, a condition characterized by a variety of heart problems. Most often, mutations in the *ANK2* gene lead to abnormalities of the heart's natural pacemaker (the sinoatrial node), a heart rate that is slower than normal (bradycardia), a disruption in the rhythm of the heart (arrhythmia), and an increased risk of fainting (syncope) and sudden death.

Each of the identified mutations in the *ANK2* gene changes a single protein building block (amino acid) in the ankyrin-B protein. Most of these mutations alter a region of the ankyrin-B protein important for its function. At least one *ANK2* gene mutation prevents ankyrin-B from getting to the cell membrane where it is needed to function. As a result of these genetic changes, the ankyrin-B protein cannot target ion channels and ion transporters to their correct locations in cardiac muscle cells. Although the channels and transporters are produced normally by the cell, they are unable to function if they are not inserted correctly into the cell membrane. This loss of functional channels and transporters in the heart disrupts the normal flow of ions, which alters the heart's normal rhythm and leads to the heart problems that can be a part of ankyrin-B syndrome.

2.2. Autism spectrum disorder

Autism spectrum disorder

3. Other Names for This Gene

- ANK2_HUMAN
- ankyrin 2, neuronal
- ankyrin B
- ankyrin, brain
- ankyrin, nonerythroid
- ankyrin-2, nonerythrocytic

- brank-2
- LQT4

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