Cantu Syndrome

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ABCC9 gene

ABCC9 gene

ATP binding cassette subfamily C member 9

Normal Function

The ABCC9 gene provides instructions for making the sulfonylurea receptor 2 (SUR2) protein. This protein forms one part (subunit) of a channel that transports charged atoms of potassium (potassium ions) across cell membranes. Each of these channels consists of eight subunits: four SUR2 proteins and four proteins produced from either the KCNJ8 or KCNJ11 gene. The SUR2 subunits regulate the activity of the channel, determining whether it is open or closed.Channels made with the SUR2 protein are known as ATP-sensitive potassium (K-ATP) channels. The channels open and close in response to the amount of ATP, the cell's main energy source, inside the cell. The resulting transport of potassium ions is part of a complex network of signals that relay chemical messages into and out of cells.Although K-ATP channels are present in cells and tissues throughout the body, the highest levels of SUR2-containing channels are found in skeletal and heart (cardiac) muscle. These channels indirectly help regulate the concentration of calcium ions in cells. This regulation is essential for normal heart function. The function of these channels in other tissues is unclear.

Health Conditions Related to Genetic Changes

CantÃº syndrome

At least 14 mutations in the ABCC9 gene have been found to cause CantÃº syndrome, a rare condition characterized by excess hair growth (hypertrichosis), a distinctive facial appearance, and heart defects. Each of the mutations changes a single protein building block (amino acid) in the SUR2 protein. These changes likely alter the structure of the protein and its ability to regulate the activity of K-ATP channels. Studies suggest that the abnormal channels are open when they should be closed. However, it is unknown how this problem with potassium channel function leads to excess hair growth, heart defects, and the other features of CantÃº syndrome.

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Familial atrial fibrillation

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Familial dilated cardiomyopathy

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Other disorders

At least two mutations in the ABCC9 gene have been identified in people with dilated cardiomyopathy, a form of heart disease that enlarges and weakens the 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. Research suggests that each mutation changes the structure of the SUR2 protein and disrupts the regulation of the K-ATP channel. Although K-ATP channels appear to play an important role in cardiac muscle, little is known about how malfunctioning channels are related to dilated cardiomyopathy.

Other Names for This Gene

ABC37 ABCC9_HUMAN ATFB12 ATP-binding cassette sub-family C member 9 ATP-binding cassette sub-family C member 9 isoform SUR2A ATP-binding cassette sub-family C member 9 isoform SUR2B ATP-binding cassette transporter sub-family C member 9 ATP-binding cassette, sub-family C (CFTR/MRP), member 9 CANTU CMD1O sulfonylurea receptor 2 SUR2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

Tests of ABCC9

Scientific Articles on PubMed

PubMed

Catalog of Genes and Diseases from OMIM

ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 9; ABCC9

CARDIOMYOPATHY, DILATED, 1O; CMD1O

Gene and Variant Databases

NCBI Gene

ClinVar

References

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Genomic LocationThe ABCC9 gene is found on chromosome 12.

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