# New genetic deafness syndrome identified

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Ten years ago, scientists seeking to understand how a certain type of feature on a cell called an L-type calcium channel worked created a knockout mouse missing both copies of the CACNA1D gene.

The CACNA1D gene makes a protein that lets calcium flow into a cell, transmitting important instructions from other cells. The knockout mice lived a normal life span, but their hearts beat slowly and arrhythmically. They were also completely deaf.

Today at the 55th Annual Biophysical Society Meeting in Baltimore, an international team lead by Hanno Bolz of the University of Cologne in Germany has identified a mutation on the CACNA1D gene affecting two families in Pakistan. The altered gene adds one extra amino acid to the middle of the protein, which is more than 2,000 amino acids in length.

The result: family members with two copies of the mutated gene are not only deaf but also have an irregular heart beat. "Their heart beats slowly, dropping below 30 beats a minute during sleep," says Joerg Striessnig, professor at the University of Innsbruck in Austria and one of the senior study authors.

The researchers analyzed the family's mutation and determined that it does not destroy the protein, says Striessnig. "Normally, part of the protein acts like a hinge to open the calcium channel once the cell gets stimulated. The mutated protein still sits in the cell's surface membrane where it should be, but the hinge does not open the channel," he says.

"It's not only interesting for medicine but also for understanding how these channels work as molecular machines ."

**More information:** The presentation, "Biophysical Properties of a Human Disease-Causing Mutation in Cav1.3 L-type Calcium Channels" by Andreas Lieb et al is at 10:30 a.m. on Wednesday, March 9, 2011 in the Baltimore Convention Center, Hall C. Abstract: tinyurl.com/4h4y5lk

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