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## NEW GENETIC DEAFNESS SYNDROME IDENTIFIED

German Researchers Describe Rare Genetic Disease that Causes Deafness and Irregular Heart Beat

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EMBARGOED for release until 10:30 a.m. Eastern time on Wednesday, March 9, 2011

For more information, please contact:

Ellen R. Weiss,  
eweiss@biophysics.org  
240-290-5606

Philip Schewe,  
pschewe@aip.org  
301-209-3092

WASHINGTON, D.C. (March 9, 2011) -- 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."

This work was funded by The Geers-Stiftung, Bonn; Imhoff-Stiftung, Köln; Köln Fortune, University Hospital of Cologne, Deutsche Forschungsgemeinschaft; Forschung contra Blindheit; Initiative Usher Syndrome.V.; the Austrian Science Fund; the Agence Nationale pour la Recherche; the Fondation de France; the Marie Curie Research Training Network CavNET; and the University of Innsbruck.

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: <http://tinyurl.com/4h4y5lk>

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#### MORE MEETING INFORMATION

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Each year, the Biophysical Society Annual Meeting brings together more than 6,000 scientists and hosts more than 4,000 poster presentations, 200 exhibits, and more than 20 symposia. The largest meeting of its type in the world, the Biophysical Society Annual Meeting retains its small-meeting flavor through its subgroup meetings, platform sessions, social activities, and committee programs.

#### QUICK LINKS

Meeting Home Page:

<http://www.biophysics.org/2011meeting>

General Meeting Information:

<http://www.biophysics.org/GeneralInfo/Overview/tabid/2062/Default.aspx>

Search abstracts:

<http://www.abstractsonline.com/plan/start.aspx?mkey={FEA830A5-24AD-47F3-8E61-FCA29F5FEF34}>

#### PRESS REGISTRATION

The Biophysical Society invites credentialed journalists, freelance reporters working on assignment, and public information officers to attend its Annual Meeting for free. For more information on registering as a member of the press, please contact Ellen Weiss at [eweiss@biophysics.org](mailto:eweiss@biophysics.org) or 240-290-5606. Also see:

<http://www.biophysics.org/Registration/Press/tabid/2148/Default.aspx>

#### ABOUT THE BIOPHYSICALSOCIETY

The Biophysical Society, founded in 1956, is a professional, scientific society established to encourage development and dissemination of knowledge in biophysics. The society promotes growth in this expanding field through its annual meeting, monthly journal, and committee and outreach activities. Its over 9,000 members are located throughout the U.S. and the world, where they teach and conduct research in colleges, universities, laboratories, government agencies, and industry. For more information on the society or the 2011 Annual Meeting, visit [www.biophysics.org](http://www.biophysics.org)

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