Researchers link gene mutations to Ebstein's anomaly

February 16 2011

Ebstein's anomaly is a rare congenital valvular heart disease. Now, in patients with this disease, researchers of the Academic Medical Center Amsterdam in the Netherlands, the University of Newcastle, UK and the Max Delbrück Center for Molecular Medicine (MDC) Berlin-Buch have identified mutations in a gene which plays an important role in the structure of the heart. The researchers hope that these findings will lead to faster diagnosis and novel, more specifically targeted treatment methods (*Circulation Cardiovascular Genetics*).

Ebstein's anomaly is a heart defect in which the valve between the right ventricle and the right atrium is abnormally formed. Since the heart valve cannot close properly, heart function is compromised. Some patients with Ebstein's anomaly additionally suffer from a myocardial disease called left ventricular noncompaction (LVNC). This disease is associated with increased risk for sudden cardiac death or inadequate functioning of the heart muscle (myocardial insufficiency).

A few years ago in a study of LVNC patients, Prof. Ludwig Thierfelder and Dr. Sabine Klaassen (both MDC) discovered mutations in three different genes that encode muscle structural proteins. These proteins are important for heart contraction and for enabling the blood to be pumped through the body. One gene in which the MDC researchers identified mutations is the gene MYH7. Mutations in this gene in LVNC patients cause sponge-like muscle tissue to protrude into the left ventricle, thus impairing the contractile performance of the heart.

As a consequence of these findings, Dr. Alex V. Postma from Amsterdam, Professor Judith Goodship from Newcastle and PD Dr. Klaassen from the MDC sought to determine whether an association exists between Ebstein's anomaly, LVNC and mutations in the gene MYH7. In a multicenter study of cohorts from the Netherlands, Germany and the UK, they studied 141 Ebstein's patients who were not related to each other for mutations in MYH7. In eight of the study participants, the researchers identified mutations in this gene. Six of these patients also suffered from the myocardial disease LVNC in addition to Ebstein's anomaly.

"From these results we conclude that one mutation can lead to different congenital heart diseases. These can even occur concurrently, as here with Ebstein's anomaly and LVNC," said Dr. Klaassen. "In these cases we recommend that other family members also undergo cardiac examinations and genetic testing, since the risk for heart arrhythmia or heart failure is increased in mutation carriers even if they are not known to have a congenital heart defect. The earlier the mutations encoding the structural proteins of the heart are recognized, the better: close monitoring, long-term ECG recording and drug treatment can be conducted at an early stage. This means that physicians can advise and treat their patients more effectively."

More information: Mutations in the Sarcomere Gene MYH7 in Ebstein's Anomaly, DOI:10.1161/CIRCGENETICS.110.957985

Provided by Helmholtz Association of German Research Centres

Citation: Researchers link gene mutations to Ebstein's anomaly (2011, February 16) retrieved 1 May 2024 from https://medicalxpress.com/news/2011-02-link-gene-mutations-ebstein-anomaly.html

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.