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Mutations Induce Severe Cardiomyopathy

Mutations in three genes that are important for heart contraction can induce left ventricular noncompaction (LVNC), a special form of cardiomyopathy. This was a key finding from current research conducted by Dr. Sabine Klaassen, Susanne Probst, and Prof. Ludwig Thierfelder of the Max Delbrück Center for Molecular Medicine (MDC) Berlin-Buch, Prof. Erwin Oechslin (Adult Congenital Cardiac Centre, Toronto, Canada) and Prof. Rolf Jenni (Cardiovascular Center, Zürich, Switzerland). In LVNC, the myocardial tissue of the left ventricle takes on a sponge-like appearance and protrudes into the ventricle which can greatly impair the pumping performance of the heart. Of the 63 LVNC patients studied, the scientists found 11 patients (17 percent) with several myocardial gene mutations. The researchers suspect that these genetic mutations can trigger severe cardiomyopathy. In the future, genetic testing can determine whether individual family members of the affected patients also carry this mutation and are, thus, predisposed to LVNC. The results of the study have just been published in the journal Circulation (2008, Vol. 117, pp. 2893-2901)*.

The heart muscle makes the heart beat about seventy times per minute, thus providing the entire body with oxygen and nutrients. Dysfunction of the heart muscle may lead to cardiac arrhythmia, cardiac insufficiency, and even heart failure.

In LVNC, a disease which was just discovered a few years ago, the left ventricle of the heart resembles that of an embryo. Since the disease can also occur in small children, scientists assumed it was a developmental disorder of the heart muscle tissue.

Now, Dr. Klaassen and her colleagues have been able to show that the disease is due to a genetic defect and is thus a familial disease. It affects genes whose proteins are responsible for contraction and, thus, for the pumping function of the heart muscle, i.e. genes encoding beta-myosin heavy chain, alpha-cardiac actin, and troponin T.

Genetic testing on individual families showed that the probability of an affected parent passing on the gene mutations to his or her children is 50 percent. "That is why gene testing of these families is so important," Dr. Klaassen said.

If a gene test turns out to be negative, the tested person can be certain that he or she will not get LVNC. But if the test is positive, the implication is not so clear. "As a consequence of the altered heart muscle tissue, the affected person may develop functional myocardial impairment later in life," Dr. Klaassen explained.

However, a mutation in these genes need not inevitably lead to myocardial insufficiency. "We examined a 70-year-old patient who did not show any symptoms of the disease although she had the mutation," the cardiologist added. "Apparently, other genetic factors, as well as environmental factors, like a healthy lifestyle, influence the manifestation of the disease."

*Mutations in Sarcomere Protein Genes in Left Ventricular Noncompaction

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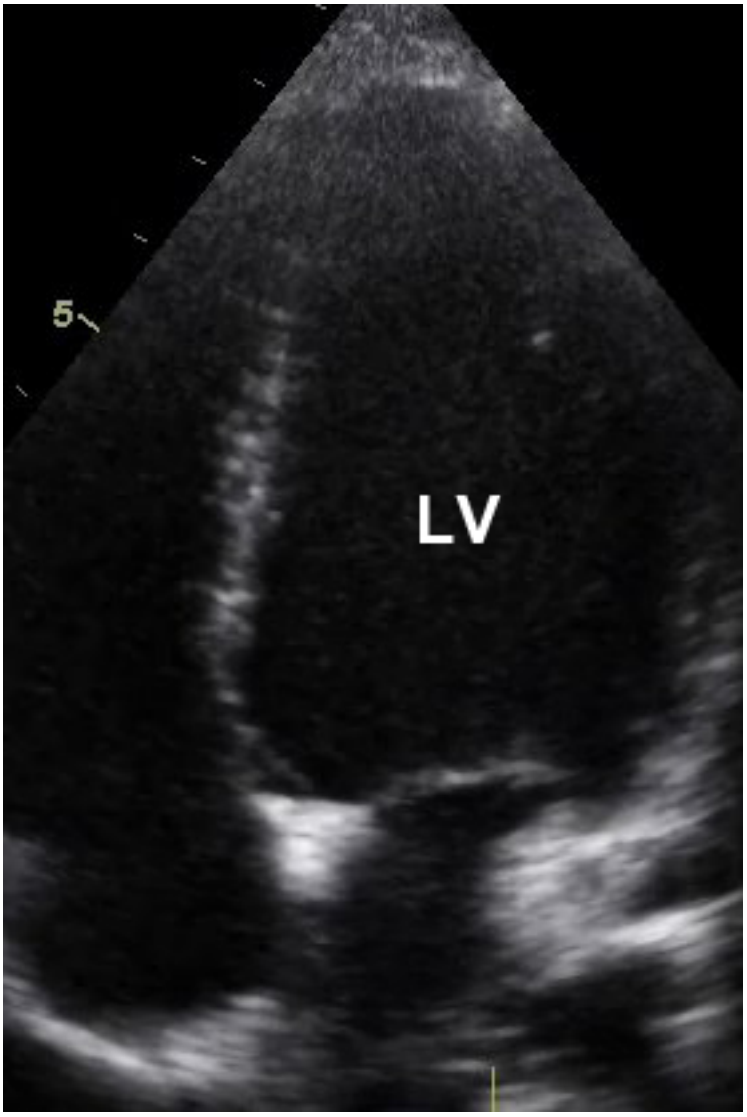
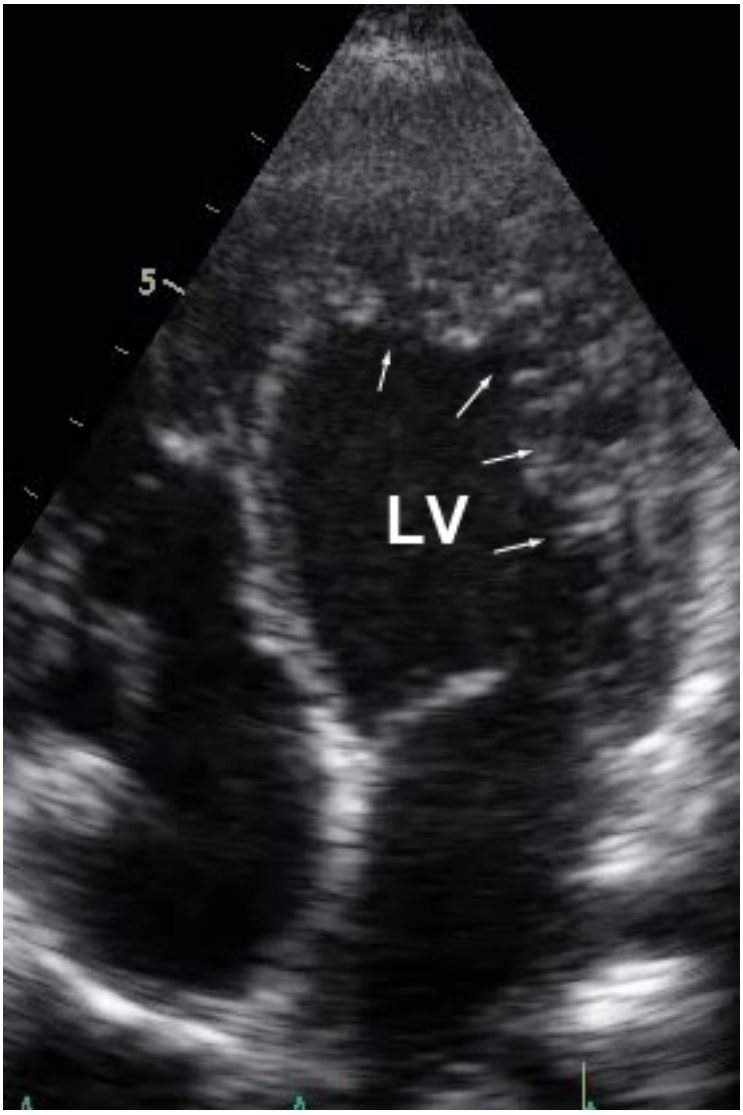


Ultrasound image of a healthy heart - In a healthy heart, the left ventricle, LV (above right), the right ventricle (above left), and their atriums are surrounded by compact muscle tissue.
(Picture: Sabine Klaassen/ Copyright: MDC)



Ultrasound image of a heart with the myocardial disorder LVNC - In a patient with LVNC, sponge-like muscle tissue protrudes into the left ventricle (above right as indicated by arrows) and impairs the heart's ability to pump blood. (Picture: Sabine Klaassen/ Copyright: MDC)