MedicalXpress

Mayo researchers discover overdiagnosis of long QT heart syndrome

May 31 2007

'Sudden death' disorder a challenge to diagnosis, but improper diagnosis has serious implications

ROCHESTER, Minn. -- Congenital long QT syndrome (LQTS) can be lethal if not diagnosed -- yet recent increased awareness of the disorder may lead to diagnosing patients when they don't have the syndrome and then prescribing treatments that restrict patients' lifestyles, a new Mayo Clinic study shows.

In their review of 176 patients referred to Mayo Clinic for a second opinion about a diagnosis of LQTS, Mayo specialists found that about 40 percent did not show sufficient evidence to merit that diagnosis. Most of these were dismissed as normal. Of the 176, most were taking drugs to correct a presumed LQTS problem and were restricted from strenuous physical activity, including competitive sports. About 10 percent had undergone surgery to have an implantable cardioverter defibrillator (ICD), a treatment for LQTS.

"Two of every five patients who were referred to us with the diagnosis of LQTS left Mayo Clinic without the diagnosis," explains Michael Ackerman, M.D., Ph.D., the pediatric cardiologist who directs Mayo's Long QT Syndrome Clinic and is the director of the Mayo Clinic Windland Smith Rice Sudden Death Genomics Laboratory. He is the senior author of a study that appeared in the May 22 issue of Circulation.

"This is dramatic evidence of just how challenging LQTS is to correctly diagnose because it can present with such a large variety of symptoms,"

Dr. Ackerman says. “There’s a real need for cautious, comprehensive evaluation by experienced LQTS specialists because there is a risk for overdiagnosis. And when that happens, some of the treatments that patients may receive when they carry this diagnosis -- such as taking medications for life, having a defibrillator implanted, or being restricted from competitive sports -- can have a profound effect on quality of life.”

Dr. Ackerman emphasizes that this subset of patients seen at Mayo Clinic may not be representative of all LQTS patients. Their cases may not have been clear in the first place, prompting the families to seek a second opinion at Mayo Clinic. He cautions against interpreting the results of this study to mean that 40 percent of all LQTS patients have errors in diagnosis. “Instead, this study has exposed the fact that sometimes the clues we use to suspect the diagnosis of LQTS can be misread, misinterpreted, mismeasured and mistaken,” he explains.

Significance of the Mayo Clinic Research

The risk of underdiagnosing LQTS has been well established in recent years. Early this year, this same investigative team published findings from a series of coroner/medical examiner cases showing that approximately half of the LQTS cases discovered postmortem had exhibited legitimate sudden death “warning signs" including sudden faints and a positive family history of sudden unexplained deaths. But the current Mayo Clinic study is the first report of the potential for overdiagnosis of LQTS. As such, it emphasizes the need to clarify methods for interpreting data obtained from heart tests used to evaluate LQTS patients, particularly data of borderline cases, Dr. Ackerman says.

“This is a diagnosis in which the stakes are extremely high,” he says. “On one hand, failing to recognize this disease could end in a tragic, premature death that could have been prevented easily with current treatments. On the other hand, the proverbial dark side of increased

awareness of LQTS and its potential lethality may prompt physicians to think, 'If I'm in doubt, I'm going to diagnose LQTS because it's the medically/legally safe thing to do.' However, what may not be appreciated is that a diagnosis of LQTS has a huge ripple effect on these individuals and their families. It is vital that we get this diagnosis right in both directions."

About Long QT Syndrome (LQTS)

Long QT syndrome is a genetic disorder of the heart's electrical system that occurs in about 1 in 3,000 people. Approximately 5 percent to 10 percent of the time, its first symptom is sudden death, often related to physical exertion or auditory triggers such as an alarm clock. However, most cases can be diagnosed following warning signs that suggest its potential presence and from objective data derived from an electrocardiogram (ECG), exercise or adrenalin stress testing, and genetic testing. Genetic testing for this condition has been available as a clinical diagnostic test for only three years. A number of treatments can then be prescribed.

Warning signs include:

- Sudden fainting spells or seizures -- especially during exercise or following an auditory trigger such as doorbells, alarms or ringing telephones.

- A family history of a sudden, unexplained death before 50 years of age -- including an unexplained drowning, unexplained motor vehicle accident and even sudden infant death syndrome (SIDS).

  "When LQTS is detected and treated properly, sudden death is now thankfully very rare for this disease," Dr. Ackerman says.

Source: Mayo Clinic