

# New study uses DNA to solve mystery of sudden unexplained death

December 10 2014

---

Researchers at the Scripps Translational Science Institute (STSI) have launched a clinical trial aimed at cracking one of the toughest mysteries in forensic science—sudden unexplained death.

The institute has teamed with the San Diego County Medical Examiner to sequence the genomes of adults, children and infants in the region whose death, most likely from heart failure, can't be explained using traditional medical investigative methods.

A dozen or more of these deaths occur annually in San Diego County, according to the medical examiner.

While large-scale DNA sequencing is increasingly being used in medicine to identify diseases, such as specific types of cancers, and to guide therapies, medical examiners have rarely employed this powerful scientific tool in their investigations.

The Molecular Autopsy Study being conducted by STSI could introduce a new paradigm in the field.

Researchers hope to identify genes associated with sudden death and discover previously unrecognized genetic mechanisms for these types of cases. The findings will be used to develop preventative screening programs and potentially life-saving interventions for relatives of [study participants](#) and others with the same DNA signature.

## Findings could help save lives

"Using state-of-the-art DNA sequencing and powerful analytics tools we can, for the first time, determine the root cause of [sudden unexpected death](#) in many families," said STSI Director Eric Topol, M.D., who also is chief academic officer of Scripps Health. "This may turn out to be important not only for [family members](#) to know if they are at risk and need to take preventive steps, but also for the public to avoid such catastrophes in young people."

STSI is a National Institutes of Health sponsored consortium led by Scripps Health in collaboration with The Scripps Research Institute (TSRI).

In almost each instance of sudden unexplained death, family members are left wondering if their relative's passing was a harbinger of some potential threat to them, said San Diego County Chief Deputy Medical Examiner Jon Lucas, M.D.

"If genetic sequencing and careful analysis of DNA data can help establish a cause in some of these cases, that could answer important questions for surviving family members and provide a sense of closure from a painful loss," he said.

Study participants must be 45 years old or younger, and not have any history of excessive drug use, alcohol abuse, morbid obesity, heart disease or any other serious medical conditions. Potential participants are initially identified by the Medical Examiner's staff and then reviewed by the STSI research team. Once the family or other next of kin gives consent, and the participant is accepted into the study, blood and heart muscle samples are collected for analysis. Researchers also plan to sequence the DNA of participant parents or other biological family members for comparison.

Researchers hope to enroll up to 100 primary study participants. They also are exploring the possibility of expanding the project to other county medical examiner departments in California and other regions of the country.

"In individuals with early unexplained sudden death, genome sequencing has the ability to uncover rare conditions with difficult-to-detect symptoms that are invisible to standard physical autopsy," said Ali Torkamani, director of genomic informatics and drug discovery at STSI. "We hope that if a rare life-threatening genetic condition is detected, life-saving interventions may be indicated in living relatives."

"This is a great opportunity for the county to work with cutting-edge researchers to solve this mystery, and we're happy to be part of a study that could make a real difference in people's lives," said Dianne Jacob, chairwoman of the San Diego County Board of Supervisors.

## **Mother seeks reason for son's death**

One of the study's first participants was Jason Lappies, 31, of San Diego, who died June 26. Jason's roommate found his body on the couch of their apartment a few hours after Jason laid down to watch a World Cup soccer match.

"He was very active and very healthy," his mother Mary Lappies said. "He didn't show any signs of being sick or not feeling good. It came with absolutely no warning."

Jason left behind friends across the globe, partly from the time he spent in South Korea teaching English to children, she said.

"His Facebook profile included the phrase 'No stoppin curiosity,' which perfectly described the way he lived. He had an amazing energy. He

could just pick people up and light up a room," she said. "By participating in the Molecular Autopsy Study, Jason's gift for helping others can live on."

Provided by The Scripps Research Institute

Citation: New study uses DNA to solve mystery of sudden unexplained death (2014, December 10) retrieved 11 May 2024 from

<https://medicalxpress.com/news/2014-12-dna-mystery-sudden-unexplained-death.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.