The story is a familiar one: a young person drowns, but members of his or her family insist that the deceased was a “good swimmer.” Now, researchers from the Mayo Clinic think they may have found a genetic component to this unlikely tragedy. The researchers identified a new mutation in a gene associated with long-QT syndrome (LQTS) in a young female drowning victim (Ackerman MJ, Tester DJ, Coburn JP, Edwards WD. Molecular diagnosis of the inherited long-QT syndrome in a woman who died after near-drowning. N Engl J Med. 1999;341:1121–1125).

Approximately 50% of LQTS is associated with mutations in 5 known genes that encode cardiac ion channels or auxiliary ion-channel subunits, according to Michael Ackerman, MD, who led the research team. In their report, the group described a new mutation in the KVLQT1 gene of a 19-year-old woman who was found face down in 4 feet of water in her health club swimming pool. Although she was resuscitated, she died 12 days later. Researchers conducted the genetic testing because of the unexplained death in the strong swimmer, the fact that she had a prolonged QT interval in an electrocardiogram test done during her hospitalization after her electrolyte balances were normal, and the signs of moderate myocardial ischemia at autopsy.

Researchers were able to use the information derived from studies of her DNA to find the same syndrome in her maternal grandfather, her mother, and her 18-year-old sister. Her sister is now receiving treatment to prevent a similar problem. After investigating this case, researchers reviewed 35 cases of LQTS seen at the Mayo Clinic and identified 6 patients who had a person or family history of drowning or near-drowning. They found mutations of the KVLQT1 gene in all 6 of the patients with such a history but in only 3 of the 29 patients with no family history.


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Genetic Drowning Trigger
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