Mr. and Ms. X tragically lost their 16-month-old son in 2011. He was a healthy, active child without any known medical problems. The day of his death, his parents noticed that he was fussier than usual but was otherwise fine. In the morning, his parents found him unresponsive and called 911. Emergency services arrived promptly but it was too late. No cause of death could be made on autopsy but the medical examiner saved blood in a lavender tube for future DNA testing.

Months later, the family sought medical evaluation and was referred to a cardiovascular genetics program to help determine the cause of their son’s death and whether they may be at risk. A cardiologist performed EKGs and echocardiograms on the parents and their 4-year-old daughter. A genetic counselor assessed the family history of signs of an inherited condition. Cardiac testing for each family member was normal and there were no signs or symptoms of a cardiac disorder in the family. The team suggested their son’s death could have been due to a genetic arrhythmia syndrome such as long QT or Brugada syndrome and recommended genetic testing. Fortunately, the medical examiner saved blood in a manner that allowed for DNA testing. Clinical genetic testing was ordered on the deceased child and a mutation in the SCN5A gene was identified consistent with Brugada syndrome.

Brugada syndrome is a genetic arrhythmia causing increased risk for sudden death, especially at night. The identification of a mutation in the deceased allowed other family members to undergo genetic testing to determine if they were at increased risk. Family testing revealed that the father also carried the mutation but was asymptomatic and at low risk for a cardiac event. The daughter tested negative for the SCN5A mutation and the family was reassured that she was not at increased risk for sudden death.

Having an explanation for their son’s sudden death helped the parents in their grieving process. Learning their daughter wasn’t at increased risk provided enormous relief. They had been so worried since the death of their son that they took turns watching their daughter sleep to make sure she was safe. Genetic testing gave them information that allowed them to move forward with their lives.