The reunion of the extended Slone family in Kentucky was an unusual event. Not only did ninety relatives gather, but medical researchers also attended, sampling blood from everyone. The reason—the family is very rare in that many members suffer from hereditary pancreatitis, locally known as Slone's disease. In this painful and untreatable condition, the pancreas digests itself. This organ produces digestive enzymes and hormones that regulate the blood glucose level. The researchers were looking for biochemical instructions, in the form of genes, that might explain how the disease arises. This information may also help the many thousands of people who suffer from nonhereditary pancreatitis.

Kevin Slone, who organized the reunion, knew well the ravages of his family's illness. In 1989, as a teenager, he was hospitalized for severe abdominal pain. When he was hospitalized again five years later, three-quarters of his pancreas had become scar tissue. Because many relatives also complained of frequent and severe abdominal pain, Kevin's father, Bobby, began assembling a family tree. Using a computer, he traced more than 700 relatives through nine generations. Although he didn't realize it, Bobby Slone was conducting sophisticated and invaluable genetic research.

David Whitcomb and Garth Ehrlich, geneticists at the University of Pittsburgh, had become interested in hereditary pancreatitis and put the word out that they were looking for a large family in which to hunt for a causative gene. A colleague at a new pancreatitis clinic at the University of Kentucky put them in touch with the Slones and their enormous family tree. Soon after the blood sampling at the family reunion, the researchers identified the biochemical cause of hereditary pancreatitis.

Affected family members have a mutation that blocks normal control of the manufacture of trypsin, a digestive enzyme that breaks down protein. When the powerful enzyme accumulates, it digests the pancreas. A disorder felt painfully at the whole-body level is caused by a problem at the biochemical level. Researchers are using the information provided by the Slone family to develop a diagnostic test and treatments for this debilitating disorder. ■