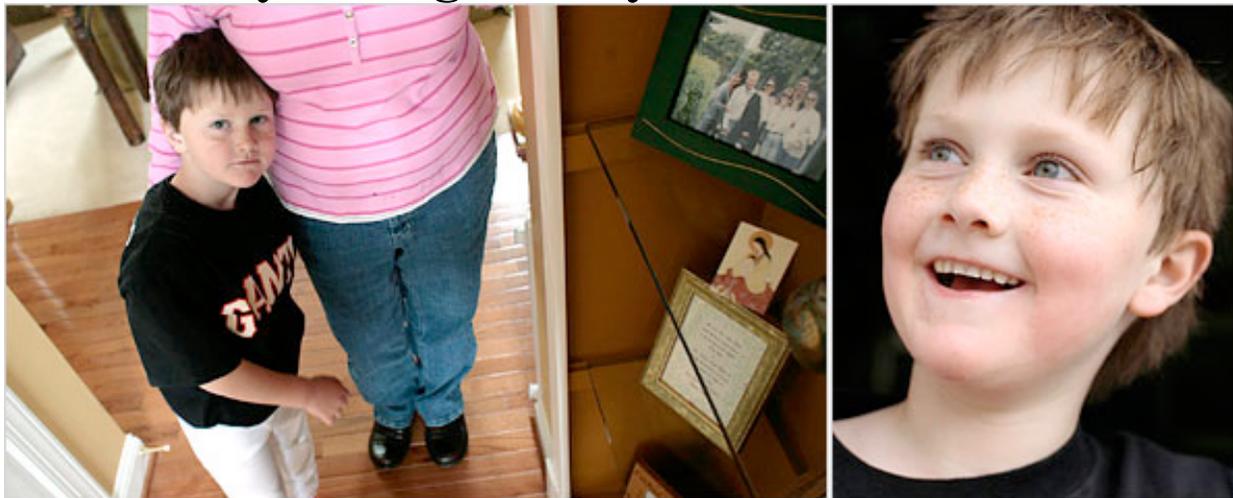


Some Diabetics Don't Have What They Thought They Had



Ryan Collins, 6, of Aldie, Va., was given a diagnosis of Type 1 diabetes shortly after birth. Doctors now know he has a rare form of diabetes that can be treated with pills instead of insulin.

ANDREW POLLACK
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Ryan Collins of Aldie, Va., was only 10 weeks old when doctors made the diagnosis: [Type 1 diabetes](#). That meant up to eight insulin shots per day, a big burden on him and his family.

“He couldn’t be anywhere unless there was someone around to give a shot,” said his mother, Dana Collins. “Everything had to be planned. There was no impromptu anything.”

Until last month, that is, when Ryan, now almost 7, stopped needing shots.

Ryan, it turns out, does not have Type 1 diabetes after all. He has a rare form of [diabetes](#), not yet discovered when he was born, that is caused by a genetic mutation. And it is treatable by a pill.

Ryan’s story is the latest case of how research is changing doctors’ understanding of diabetes.

Diabetes is named, from the Greek, for its symptoms of heavy thirst and frequent urination. But for hundreds of years, it has been possible to make a further breakdown. Diabetes mellitus, from the Latin for honeyed, was the form with sweet urine. [Diabetes insipidus](#) was marked by bland or insipid urine.

This may have been the first time two diseases with similar symptoms were distinguished by a “molecular” test. “We had receptors for one molecule and could figure it out,” said **Dr. Yves A. Lussier**, **director of the center for biomedical informatics at the [University of Chicago](#)**, referring to taste buds.

Today these are considered completely different diseases. Mellitus is the one commonly called diabetes. It involves high blood sugar and insulin. Insipidus is related to a different hormone, [vasopressin](#).

In the 1930s, after it became possible to treat diabetes with insulin derived from animals, diabetes mellitus itself began to be subdivided into what would come to be called Type 1, marked by lack of insulin, and Type 2, marked by insensitivity to insulin. The distinction became stronger in the 1970s with the development of blood tests to tell them apart better, though there is still no definitive test.

Now, in just the last few years, scientists have found about 35 genes that influence the risk of getting diabetes, said Dr. David Altshuler, a professor of medicine and [genetics](#) at Harvard. But so far, there is no overlap at all between the genes that help cause Type 1 and those that help cause Type 2, he said.

“In that regard, it’s held up that they are separate diseases at the genetic level,” Dr. Altshuler said.

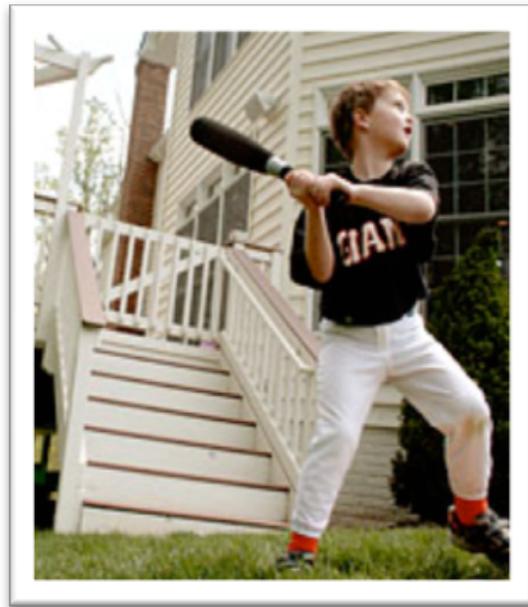
Some scientists think genetic analysis will further subdivide Type 2. While there is yet no major new class (a Type 3 or Type 4), scientists are discovering the genetic causes of several rare types of diabetes that are caused by single mutations.

The most common such “monogenic” diabetes is maturity-onset diabetes of the young, or MODY. Dr. Andrew Hattersley of the University of Exeter in England, a leading researcher, said 1 percent to 2 percent of diabetics might have MODY. But most do not know it. “There were a whole lot of people misdiagnosed as Type 1 or Type 2,” Dr. Hattersley said. But now there are genetic tests for MODY, and people with MODY might need less treatment, he said.

Dr. Hattersley has also helped show that diabetes diagnosed in the first six months of

life is monogenic. And about half those cases are caused by a particular mutation that can be overcome by sulfonylurea pills, an old and inexpensive class of diabetes drugs.

After he tested positive for that mutation, Ryan Collins spent five days last month in the [University of Chicago Medical Center](#), where doctors gradually substituted the pills for the insulin.



Andrew Council for The New York Times
Ryan Collins, at home in Virginia, was weaned off the insulin he had been taking since infancy after doctors changed his diagnosis.

Thirty-five or so American children with that mutation have now been weaned off insulin, according to the [Chicago medical center](#), which has been involved in many of the cases. It says there may be 1,000 to 2,000 such children who have not been identified.

Ryan now takes three small pills with breakfast and three with dinner. His body is making insulin again, and his blood sugar is

better controlled than when he was taking all those shots.

“He’s ecstatic,” Ms. Collins said. “For this summer, I’m signing him up for summer camp, which he’s never been able to do before.”