Frequently Asked Questions on Monogenic Diabetes

What is monogenic diabetes?
The most common types of diabetes are type 1 and type 2 diabetes. In both of these common forms of diabetes, a large number of different genes are involved in the development and progression of the disease. In other words, these forms of diabetes are “polygenic”, with multiple contributing genetic factors (in addition to environmental factors). No single mutation in any one of the genes alone could cause type 1 or type 2 diabetes.

However, in very rare cases, diabetes can be caused by a mutation in a single gene, resulting in a different type of diabetes called “monogenic diabetes”. To date, about 20 different single gene mutations are known to cause monogenic diabetes. Monogenic diabetes can result in either neonatal diabetes (diagnosed in newborns and infants) or in maturity onset diabetes of the young (MODY) which affects adults.

Because monogenic diabetes is so rare, and has only relatively recently been recognized and characterized, it is often misdiagnosed as either type 1 or type 2 diabetes.

How common are monogenic forms of diabetes?
Monogenic forms of diabetes are very rare. They account for an estimated 1 – 5 percent of all cases of diabetes in young people.

Who should be tested for monogenic diabetes?
Genetic testing can diagnose most forms of monogenic diabetes. People who were diagnosed with diabetes before the age of six-to-nine months should consider undergoing genetic testing for monogenic forms of neonatal diabetes. For a list of other considerations regarding testing for monogenic diabetes see [http://monogenicdiabetes.uchicago.edu/who-should-be-tested/](http://monogenicdiabetes.uchicago.edu/who-should-be-tested/).

What steps should I take to find out if I have monogenic diabetes?
If you think that you may have a monogenic form of diabetes, you should contact your physician for a consultation. Your physician can evaluate whether you should be tested for monogenic diabetes. You should not change or discontinue your current treatment regimen until you are tested and your care can be directed by a medical specialist trained in monogenic diabetes.

What is the treatment for monogenic diabetes?
There are many forms of monogenic diabetes, each caused by a different gene mutation. Some forms may be treated with oral medication, while others are treated with insulin.

If appropriate, any transition off insulin therapy must be pursued under the careful supervision of a physician. Such a transition is appropriate only in cases where genetic testing reveals a very specific, rare mutation causing one of the forms of monogenic diabetes treatable with oral medication.

Insulin remains a life-saving therapy for people with diabetes. People with diabetes can live long, healthy lives on insulin therapy.
What has the American Diabetes Association done to advance the understanding of monogenic diabetes?

The American Diabetes Association recently funded an expansion of the Monogenic Diabetes Registry at the University of Chicago, which now includes more than 2,000 participants and provides educational and clinical support to patients, families and clinicians. Through this funding, Dr. Louis Philipson discovered new gene mutations that cause monogenic forms of diabetes, demonstrated cost-efficiency of targeted genetic testing for the most common causes of monogenic diabetes, and characterized complications and appropriate therapy for specific gene mutations.

This registry serves as a national resource for interpretation of genetic results and guidance on appropriate therapy for various types of monogenic disease. For more information on the Monogenic Diabetes Registry, and how people with monogenic diabetes may participate, please see http://monogenicdiabetes.uchicago.edu/registry/.