



Monogenic Diabetes

WHAT IS MONOGENIC DIABETES?

Monogenic diabetes is a rare condition resulting from mutations (changes) in a single gene. In contrast, the most common types of diabetes—type 1 and type 2—are caused by multiple genes (and in type 2 diabetes, lifestyle factors such as obesity). Most cases of monogenic diabetes are inherited.

Monogenic diabetes appears in several forms and most often affects young people. In most forms of the disease, the body is less able to make insulin, a hormone that helps the body use glucose (sugar) for energy. Rarely, the problem is severe insulin resistance, a condition in which the body cannot use insulin properly.

A correct diagnosis can help people get the right treatment. For example, some children with monogenic diabetes are misdiagnosed with type 1 diabetes and are given insulin. When correctly diagnosed, some of these children can take diabetes pills instead, with even better glucose control. A correct diagnosis may also benefit family members, who might have monogenic diabetes themselves without knowing it.

DID YOU KNOW?

Many people with monogenic diabetes are mistakenly diagnosed with type 1 or type 2 diabetes and may not receive the best treatment.

WHAT ARE THE TWO MAIN FORMS OF MONOGENIC DIABETES?

Maturity-onset Diabetes of the Young (MODY)

MODY is the most common form of monogenic diabetes. It usually first occurs in children or teenagers but sometimes is not found until adulthood. MODY can be mild or severe, depending on which gene is involved. Researchers have found at least nine different genes responsible for MODY, and new genetic causes are still being discovered.

Neonatal diabetes

This rare condition occurs in the first 6 months of life. Many infants with neonatal diabetes don't grow well before birth and are born small for their age. The two types are

- Permanent neonatal diabetes, a lifelong condition
- Transient neonatal diabetes, which goes away during infancy but can return later in life

HOW ARE MODY AND NEONATAL DIABETES DIAGNOSED?

Because monogenic diabetes is rare, this diagnosis is often not considered in people with diabetes. However, certain factors can make doctors suspect that a diagnosis of type 1 or type 2 diabetes is not correct. A combination of tests and clinical factors help rule out type 1 or type 2 diabetes and identify MODY or neonatal diabetes.

Blood tests

Blood tests of glucose levels, and sometimes tests that show how much insulin the body is making, help with diagnosis. Doctors might also check for the presence of certain auto-antibodies (substances made by the body that work against one's own healthy tissues), which suggests type 1 diabetes.

Clinical factors

Doctors consider factors that may suggest monogenic diabetes:

- Being diagnosed in the first 6 months of life
- Having other conditions caused by a specific gene mutation, such as cysts in the kidneys
- Not being obese, or having diabetic family members who are normal weight
- Having a family history of diabetes, especially when a parent is affected
- Belonging to certain ethnic groups (with European Caucasians less affected by type 2 diabetes)

None of these factors alone mean someone might have monogenic diabetes. Instead, they are considered together, along with blood test results.

Genetic testing

A health care provider can best determine whether genetic testing is needed. Testing of the genetic information in a blood sample can determine whether a person has a gene causing MODY or neonatal diabetes. Doctors also can check family members of the person with MODY or neonatal diabetes for the presence or risk of diabetes.

WHAT IS THE TREATMENT FOR MODY?

Treatment depends on the type of MODY. Some people do not need any treatment besides diet and exercise. Others need diabetes medicines. These include insulin or a sulfonylurea—a type of diabetes pill that helps the body make more insulin. In one type of MODY, patients also may need treatment for related conditions such as kidney cysts and gout.

WHAT IS THE TREATMENT FOR NEONATAL DIABETES?

Doctors choose treatment based on the cause. Some types of neonatal diabetes can be treated with a sulfonylurea but others require insulin. Infants with transient neonatal diabetes may require insulin at first but the condition may disappear at about the age of 12 weeks. If the diabetes returns later in life, insulin might be needed.

WHAT SHOULD YOU DO IF YOU THINK YOU OR A FAMILY MEMBER MIGHT HAVE MONOGENIC DIABETES?

If you or a family member has been diagnosed with type 1 or type 2 diabetes, but you suspect it might be monogenic diabetes, talk with your doctor. Only a specialist, such as an endocrinologist specializing in diabetes, can diagnosis monogenic diabetes.

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The Hormone Health Network offers free, online resources based on the most advanced clinical and scientific knowledge from The Endocrine Society (www.endo-society.org). The Network's goal is to move patients from educated to engaged, from informed to active partners in their health care. This fact sheet is also available in Spanish at www.hormone.org/Spanish.

Monogenic Diabetes Fact Sheet

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Questions to ask your doctor

- Does my child (or do I) have a form of monogenic diabetes?
- What type of monogenic diabetes does my child (or do I) have?
- What are the treatment options?
- What are the advantages and disadvantages of each treatment option?
- Should I see a diabetes educator?
- Should I see an endocrinologist?

RESOURCES

- Find-an-Endocrinologist: www.hormone.org or call 1-800-HORMONE (1-800-467-6663)
- Find a diabetes educator (American Association of Diabetes Educators): www.diabeteseducator.org/DiabetesEducation/Find.html
- Hormone Health Network information about diabetes: www.hormone.org/diabetes
- National Diabetes Information Clearinghouse (National Institutes of Health): <http://diabetes.niddk.nih.gov/dm/pubs/mody/index.aspx>
- The American Diabetes Association: www.diabetes.org
- Juvenile Diabetes Research Foundation: www.jdrf.org



www.hormone.org