

Hormone Disorders

# Monogenic Diabetes

Patient Guide



Average readability





## Introduction

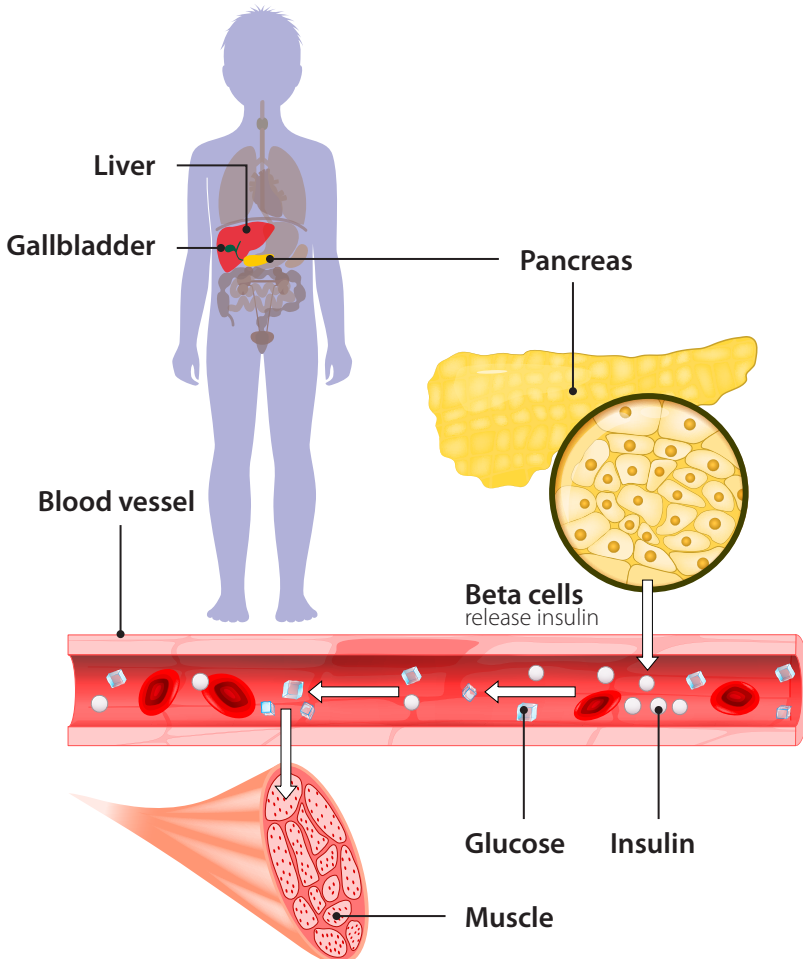
The aim of this leaflet is to describe a condition known as Monogenic Diabetes. It will discuss information on how it is diagnosed, treated and some of the problems it may cause. Hopefully, this leaflet will help you understand this condition and give you a basis for discussions with your General Practitioner or Specialist Team.

## What is Diabetes Mellitus?

Diabetes mellitus is a condition where blood glucose (“sugar”) level is too high. There are two main types, Type 1 and Type 2. There are some other rarer types of diabetes too. Some of them are called monogenic diabetes (controlled by a single gene).

# What is Monogenic Diabetes?

Monogenic diabetes is a disease that affects the ability to produce insulin. Insulin is a hormone (a “messenger”) created by the pancreas. It controls the amount of glucose in the bloodstream at any given moment and helps the body to turn food into energy.





Monogenic diabetes include Neonatal Diabetes Mellitus and MODY (maturity onset diabetes in the young).

## Neonatal diabetes mellitus

Neonatal diabetes mellitus (NDM) is a disease that occurs in the first 6 months of life. Infants do not produce enough insulin, leading to an increase in glucose accumulation. It is a rare disease, occurring in only one in 90,000 to 500,000 live births. NDM can be mistaken for the much more common type 1 diabetes, but type 1 diabetes usually occurs later than the first 6 months of life. There are two types of NDM: permanent and transient NDM. Permanent NDM is a lifelong condition. Transient NDM is diabetes that disappears during infancy, however, it may appear again later in life.

## MODY (maturity onset diabetes of the young)

MODY is also a rare form of diabetes that occurs later in life (during adolescence or early adulthood). There are several different types of MODY, so a genetic test needs to be carried out to determine what type it is. MODY can be mistaken with the much more common type 2 diabetes, which usually occurs in overweight or obese subjects.

## What are the symptoms of monogenic diabetes?

The most common symptoms of this condition are:

- Excessive thirst (also known as polydipsia) and increased urination (also known as polyuria)
- Dehydration
- Ketoacidosis (a diabetic complication that occurs when the body produces high levels of acid in the blood (ketones)).

In neonatal diabetes mellitus, some other symptoms may also be present:

- Intrauterine Growth Restriction (A condition in which the baby is smaller than he or she should be at birth)
- Neurological defects.

## How is monogenic diabetes diagnosed?

The body has more than 30,000 individual genes. Mutations (changes) in one gene can lead to monogenic diabetes. So far, more than 20 different genes have been linked to monogenic diabetes. Individuals with monogenic diabetes can pass it on to their children or future generations. Your diabetologist might ask for genetic testing to decide about the best form of treatment.





## Can several members of the family have monogenic diabetes?

Monogenic diabetes is caused by a mutation, or change, in a gene. It means that it runs strongly in families, usually affecting someone in each generation. Usually, there is a 1 in 2 risk of an affected person passing the condition to an offspring.

## What is the treatment for monogenic diabetes?

The treatment of monogenic diabetes is mainly insulin replacement therapy, using insulin injection. Monogenic diabetes may sometimes be treated with oral sulfonylureas, depending on the genetic cause of the diabetes.



## What happens in adolescence or in adulthood?

Usually, monogenic diabetes is a lifelong condition. In the transient form of neonatal diabetes, diabetes disappears within the first years of life. Nevertheless, recurrence of diabetes is common in adolescence or adulthood.

A number of health complications can occur in adults with monogenic diabetes, depending on which gene is affected. The following complications are relatively common:

- Diabetic ketoacidosis in poorly treated patients
- Developmental delay in various degrees, such as learning disabilities or mild muscle weakness
- Macroglossia – a larger than normal tongue.



## What are other sources of useful information?

The goal of this leaflet is to provide a basic overview on the diagnosis and treatment of Monogenic Diabetes.

Educational material can also be found by contacting the following organisations:

- **European Society for Paediatric Endocrinology**  
Starling House  
1600 Bristol Parkway North  
Bristol  
BS34 8YU  
espe@eurospe.org  
Telephone +44 (0) 1454 642246  
www.eurospe.org
- **British Society of Paediatric Endocrinology and Diabetes**  
bsped@endocrinology.org  
<https://www.bsped.org.uk/>
- **Child Growth Foundation**  
info@childgrowthfoundation.org  
Telephone +44 (0) 208 995 0257  
www.childgrowthfoundation.org
- **The Endocrine Society**  
www.endo-society.org



You can also consult your Specialist Team for additional information in your local area.



# Monogenic Diabetes

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This leaflet is part of the **Hormone Disorders Series**

The following are also available:

- Growth Hormone Deficiency
- Puberty and the Growth Hormone Deficient Child
- Congenital Adrenal Hyperplasia
- Constitutional Delay of Growth and Puberty
- Multiple Pituitary Hormone Deficiency
- Craniopharyngioma
- Intrauterine Growth Retardation or Small for Gestational Age
- Hyperthyroidism
- Hypothyroidism
- Type 2 Diabetes and Obesity
- Diabetes Insipidus

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