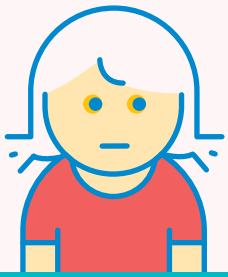
SingHealth DukeNUS
ACADEMIC MEDICAL CENTRE

Genomic
Medicine Centre

# Citrin Deficiency

SingHealth

## What is citrin deficiency?

Citrin deficiency (CD), also known as citrullinaemia type 2, is an inherited metabolic disorder in which the body is unable to make citrin, a protein that helps move substances within the cells.

Citrin helps to break down sugar (carbohydrates), make proteins and nucleotides, and get rid of toxins (ammonia) in the body. It is also important for the liver to work properly.

**Individuals with CD often prefer high protein and high fat foods, rather than high carbohydrate foods.**

## How common is CD?

It is estimated that **one in 10,000 to 38,000 people** are born with CD. It can occur in both genders, and across all races.

## What are the signs and symptoms of CD in infancy?

In babies, the signs and symptoms of CD usually begin between one and five months of age.

These include:

**Yellowish skin and eyes (jaundice)**

**Liver problems such as cholestasis (poor bile flow), enlarged liver, fatty liver**

**Low birth weight**

**Delayed growth**

**Hypoglycaemia (low blood sugar)**

**Hyperlipidaemia (high lipid levels)**

These may be seen especially when babies with CD eat foods that their bodies cannot break down, and symptoms can be triggered by long periods of fasting, illness and infection.

## What are the symptoms of CD?

Signs of CD can appear any time from birth to adulthood.

### 1 NICCD (Neonatal intrahepatic cholestasis caused by CD)

**Age of presentation**

Neonates/infants (0-1 year old)

**Signs & symptoms**

- Jaundice
- Enlarged liver
- Growth failure
- Liver dysfunction
- Fatty liver

## 2 FTTDCD (Failure to thrive and dyslipidaemia caused by CD)

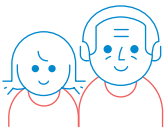
**Age of presentation**
Childhood (> 1 year old)

**Signs & symptoms**
Often a silent period with no symptoms.
If symptoms present, they may include:

- Poor growth
- Hypoglycaemia
- Hyperlipidaemia
- Fatty liver, pancreatitis, hepatoma

## 3 CTLN2 (Citrullinaemia type 2)

**Age of presentation**
Adolescents/adults
(11-79 years old)



**Signs & symptoms**

- Raised ammonia level
- Abnormal behaviour
- Brain oedema (swelling of brain)
- Fatty liver, pancreatitis, hepatoma
- Liver failure

It is important to note that not every patient with CD will develop FTTDCD or CTLN2.

## How is CD managed?

- Lactose-free formula / diet for the first year of life
- Low carbohydrate, high protein and high fat diet
- Medium-chain triglycerides (MCT) oil supplementation

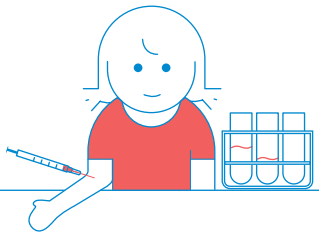
**Growth, development, intelligence quotient and general health are expected to be normal when treatment is started during infancy.**

Ongoing follow-up throughout life is required to ensure continued good health through early detection of complications.

## Is there any monitoring required?

Your child will undergo regular clinical assessments for growth, and periodic investigations by your doctor with the following assessments.

- Liver function test
- Ammonia, plasma
- Amino acid, plasma
- Lipid panel
- Galactose (< 1 year old)
- Ultrasound hepatobiliary system / abdomen



## What causes CD?

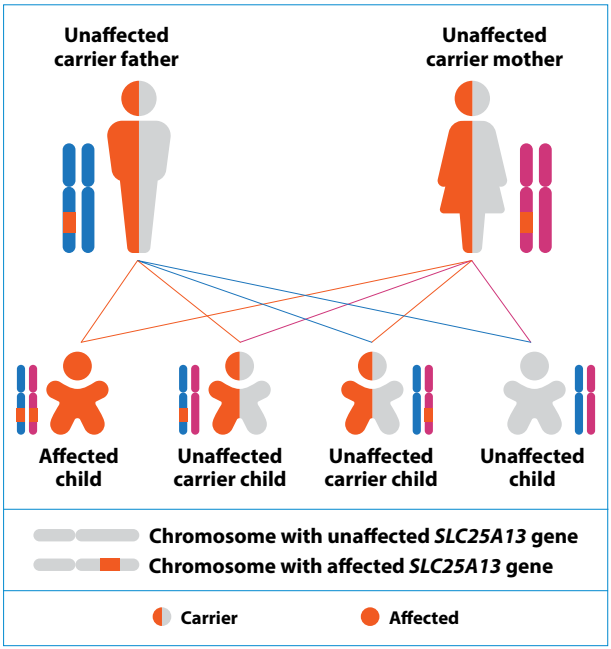
CD is caused by a change in the *SLC25A13* gene. This gene gives the body instructions for making the protein citrin. Without a working *SLC25A13* gene, the body cannot make citrin correctly, thus leading to the signs and symptoms mentioned on pages 3-4.

## How is CD diagnosed?

CD may be suspected based on its clinical presentation. The diagnosis is confirmed by further blood tests including plasma amino acids.

In addition, genetic testing can be done via blood for molecular testing of the *SLC25A13* gene.

## How is CD inherited?



Everyone has two copies of each gene in their body's cells, one copy from each parent. CD follows a **recessive inheritance pattern**. This means that a patient with CD must have two faulty copies of *SLC25A13* to cause features of CD.

An individual with one faulty copy of *SLC25A13* is known as a carrier. Typically, carriers of an autosomal recessive condition do not have any signs or symptoms.

## How likely will I have another child with CD?

Parents who are both carriers of a faulty *SLC25A13* gene have a 25% chance of having an offspring with CD.

*CD is a lifelong condition. Should you require financial assistance or emotional support, please approach your doctor for referral to a medical social worker.*

### Support Group

**Citrin Foundation**
Citrin Foundation aims to raise awareness of CD and provide support to both the medical community in tackling CD, as well as to patients and their families with CD.
**Email:** info@citrinfoundation.org
**www.citrinfoundation.org**

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