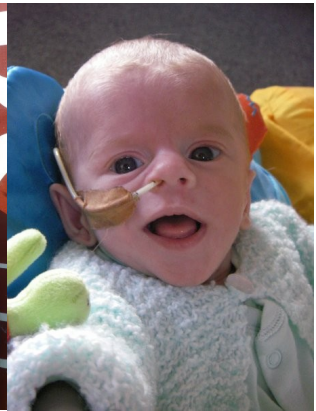


Understanding  
chromosome  
disorders

Unique



# GATA6 Syndrome



[rarechromo.org](http://rarechromo.org)

## What is GATA6 syndrome?

GATA6 syndrome occurs when one of a person's two copies of the GATA6 gene doesn't work as it should.

Our DNA is formed by 3 billion letters that contain the instructions for the function and control of all the processes within our body. Specific sets of instructions are called genes. GATA6 is one of the genes encoded in the DNA. GATA6 is important for development of different organs, including the heart and the pancreas, of a baby in the womb. If one of the copies of the GATA6 gene contains a spelling mistake (a genetic change), it doesn't work as it should and some organs in the baby (usually the pancreas and the heart) will not develop properly.

## How many people have this condition?

GATA6 syndrome is a very rare condition. To date (2016) there are just over 50 cases that have been described worldwide.

## Why did this happen?

When children are conceived, the parents' DNA is copied in the cells that make a new child. The biological copying method is not perfect and occasionally random rare changes occur in the DNA for the first time. These changes are present in the children but are not seen in the DNA of their parents. In most cases, the DNA change in GATA6 occurs 'out of the blue' in this way (this is what you may hear referred to as a 'de novo' or spontaneous change). This change happens naturally in all plants and animals and is not due to the parents' lifestyle or anything they did. In some rare cases, one parent may have the same genetic change as their child.

## Can it happen again?

If the GATA6 mutation is not found in either parent's DNA, the chance of having another child with the same genetic change would be considered extremely low. Usually, this risk would be estimated as less than 1%. If the genetic analysis of the parents of a child with GATA6 syndrome shows they carry the same genetic change, the chance of it happening again is much higher (up to 1 in 2). For specific advice about the chance of this happening again, it would be sensible to speak to a clinical geneticist or genetic counsellor.

## Can this be cured?

The whole syndrome can't be cured, but the associated health problems can be treated (see Medical concerns section on page 3).

## Most children with GATA6 syndrome have:

- Intrauterine growth retardation  
(born with low birth weight for gestational age)
- Heart defects at birth
- Complete or partial lack of a pancreas. This leads to diabetes diagnosed soon after birth and sometimes pancreas exocrine insufficiency (reduced/lack of production of digestive juice from the pancreas)

## Some children with GATA6 syndrome have:

- Underactive thyroid (hypothyroidism) from birth
- Absence of gall bladder and/or defective bile duct development
- Hernias at birth

## Medical concerns

### ■ Heart defects at birth

Most children with GATA6 syndrome are born with one or more defects in the structure of their heart. This can range from a 'hole' being present in the wall that separates the chambers of the heart to more complex problems including some of the blood vessels being in the wrong position. Some (but not all) children need to have surgery to fix these problems.

### ■ Neonatal diabetes

The majority of babies born with GATA6 syndrome have an incompletely formed pancreas and this results in them having diabetes and needing to take insulin from soon after birth for the rest of their life. Insulin will be given either by injections or via an insulin pump.

### ■ Exocrine pancreatic insufficiency

In some babies, the problems in pancreas formation results in an inability to digest food, predominantly fatty food. This is due to lack of digestive juice that the pancreas makes during eating. These babies often have abdominal pain, diarrhoea and failure to thrive. It is important to recognise these symptoms because these children need to take pancreas digestive enzymes with food (commonly called Creon, a pharmaceutical brand name) that will help them absorb their food. If they don't get treatment, they won't get enough nutrients and their growth will be impaired.

### ■ Premature birth and additional complications

Some babies are born preterm and many have growth retardation (usually with a birth weight under 2 kg). In these cases it is possible that the baby has additional medical problems such as a diaphragmatic hernia (a hole in the diaphragm) that needs to be corrected with surgery.

## ■ Absence of a gall bladder

In most people, the lack of a gall bladder does not cause symptoms and does not require treatment, but in a small number of individuals it may cause upper abdominal pain, fatty food intolerance or indigestion.

## ■ Defective bile duct development

Babies with defective bile ducts will develop jaundice (yellow discolouration of the skin and whites of the eyes) soon after birth and will require further investigation followed by surgery, called the Kasai procedure. During this procedure, the child's damaged bile ducts are removed and a loop of intestine is brought up to replace them, bile can then flow straight to the small intestine. Some children may need a liver transplant.

## Development

### ■ Physical development

As long as a child receives the appropriate amount of insulin by injection or pump and (if needed) the oral pancreatic enzymes for digestion, their physical development will not be affected.

### ■ Learning and Behaviour

Learning and behaviour can be very variable in children with GATA6 syndrome, just as it can with all children. Some children have learning difficulties and behaviour problems whilst others don't have these problems. It is not known if this is directly caused by the GATA6 syndrome or if there are other causes in some children.

### ■ Speech

Speech and language is not affected in most children with GATA6 syndrome.

## Management recommendations:

- Insulin treatment for diabetes
- Pancreatic imaging to assess if the pancreas is present
- Measurement of faecal elastase is recommended to see if there is a lack of pancreatic digestive juice
- If indicated, treatment with pancreatic enzymes is needed to improve food digestion and growth
- Regular assessment by a dietician for the adequacy of the pancreatic enzyme replacement
- Treatment with fat soluble vitamins (vitamin A, D E, K) for children who are on pancreatic enzyme replacement

## Families say ...

*“ Despite a very tough start to life, Finlay has surpassed our expectations and is leading a healthy, active, happy life. While he may be smaller than his peers he makes up for it with an infectious enthusiasm, fitting in well at school. His ability to deal with the daily burden of his diabetes needs without complaint is inspiring.”*

## Inform Network Support



### Rare Chromosome Disorder Support Group

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