

## What are the chances that we will have another child with CHD?

Most cases of CHD occur without a family history.

A genetic counsellor or genetic specialist can work out the specific chance for your family.

The table below is a general guide to working out that chance. These figures are worked out based on any other family member who have or had CHD.

Recurrence risks of CHD	
No family history	Approx. 1%
One child with CHD	1-6%
More than two children with CHD	3-10%
Mother has CHD	2-20%
Father has CHD	1-5%
More than two first degree relatives (parent or brother/sister)	Up to 50%
Child diagnosed with genetic condition	Up to 50%
Child with diagnosed single gene cause (non-syndromic)	Up to 50%

## More information and support:

If you have any questions or would like more information about what is in this brochure, please contact:

**Heart Centre for Children Genetics Clinic:**  
(02) 9845 1333

## Additional information and support can also be found at:

**Centre for Genetics Education:**  
(02) 9462 9599 or [www.genetics.edu.au](http://www.genetics.edu.au)

**Heart Centre for Children Psychology Service:**  
(02) 9845 0088

**HeartKids Australia:** [www.heartkids.org.au](http://www.heartkids.org.au)

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the childr<sup>en</sup>'s hospital at Westmead



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# Congenital Heart Disease

How and why did this happen?

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HCC4904/0319



The Sydney children's Hospitals Network  
care, advocacy, research, education

## What is congenital heart disease?

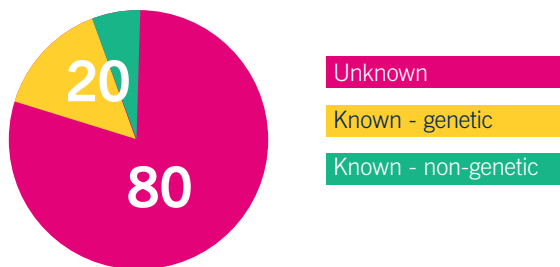
Congenital heart disease (CHD) is any problem with the child's heart that is present at birth. CHD affects about 1 in every 100 babies born. In Australia more than 2400 babies are born with CHD each year.



There are many kinds of CHD, some are minor and do not require any treatment. Others require treatment such as surgery.

## Causes of CHD

For every 100 children who are born with CHD the cause is known or can be worked out for about 20.



## Known causes of CHD

There are two types of known causes: genetic and non-genetic.

### Genetic causes of CHD

In some cases a problem with the child's genes has led to the heart condition.

We all have about 20,000 genes in the cells of our bodies. Some of these genes are important for how our heart develops and works.

The genes are organised into strings of genes called chromosomes. We all usually have 46 chromosomes that are in pairs. So we have two copies of each of the 23 chromosomes. We inherit one copy from our Mum and one from our Dad. A problem with one or more of these genes can cause a genetic condition.

### Genetic conditions

Sometimes the gene problems can cause a "genetic syndrome". In these cases more than just the heart can be affected.

These syndromes can occur due to being born with:

- Extra copies of one of the chromosomes
- A change in sections of one of the chromosomes
- A variation that has made one of the 20,000 genes that are important for the heart become faulty.

More rarely, CHD is not part of a genetic syndrome and is the only problem present. Changes affecting more than 30 different genes cause these forms of CHD.

If a genetic cause for the heart condition is suspected, parents may be referred to a Genetics Service where more information about the condition as well as possible genetic testing options can be discussed.

## Non-genetic causes of CHD

In rare cases, other factors, such as environmental factors, may have caused the CHD. We still do not understand the role of these factors in causing CHD.

Research is continuing but these basic recommendations may help to reduce the risk of a heart condition developing and should be considered before you fall pregnant:

- Talking to a doctor about any medications you plan to take during pregnancy
- Seeking advice such as taking a multivitamin containing folic acid before and during pregnancy
- Completing rubella (German measles) vaccinations
- Managing any health conditions, such as diabetes, as well as possible

## Unknown causes of CHD

For every 100 children with CHD, the exact cause is unknown in 80 of them.

Recent research suggests that in these cases, the heart condition most likely develops due to a combination of both genetic and environmental factors. So, rather than just one gene becoming faulty, variations in many genes involved in heart development may interact with each other and with environmental factors to cause CHD.

