CONGENITAL HEART DISEASE CAUSES

When parents first discover that their baby or child has a heart abnormality, it is common to ask,
- "Why has this happened?"
- "Will it happen again in our family?"

These same questions come up when a person with congenital heart disease considers having children.

The causes of heart defects.

A congenital heart defect is a heart problem which is present at birth. It is caused by abnormal formation of the heart during fetal development. Congenital heart defects are the most common types of birth defects, affecting 1 in 100 babies. In most cases, when a baby is born with congenital heart disease, there is no known reason for it.

Some of the known causes of congenital heart defects include genes, environmental factors and other factors relating to maternal health. In around 8 out of 10 cases, the reason for the congenital heart defect is unknown.

Most mothers of babies born with congenital heart disease will look critically at their own behaviours during pregnancy to try to find a cause for their child's illness.
It is important to remember that most cases of congenital heart disease have no known cause. However, some types of congenital heart defects are known to occur more often when the mother comes into contact with certain substances during the first few weeks of pregnancy, while the baby's heart is developing. Some maternal illnesses and medications taken for these illnesses have been shown to affect the heart's development. Other illnesses or medications seem to have no impact on the baby's heart. **Always consult your doctor for more information.**

Most cases of congenital heart disease have no known cause. Some of the known causes of congenital heart disease include:

- Genes – 20 per cent of cases have a genetic cause
- Other birth defects – a baby affected by certain birth defects, such as Down syndrome, is more likely to have a heart defect
- Maternal illness – illness of the mother during pregnancy (for example, rubella – now rare) may increase the risk of congenital heart disease
- Medication and drugs – medication (over-the-counter or prescription) or illicit drugs taken by the mother during pregnancy may increase the risk of congenital heart disease
- Alcohol – large amounts of alcohol during pregnancy may increase the risk of congenital heart disease
- Maternal health – factors such as unmanaged diabetes and poor nutrition during pregnancy may increase the risk
- Maternal age – babies of older women are more likely to have a birth defect than babies of younger women

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The genetics of heart defects

The risk of congenital heart disease increases when either parent has congenital heart disease, or when another sibling was born with congenital heart disease.

If there are no other family members with congenital heart disease, the chance of a congenital heart abnormality in a future pregnancy (brother or sister) is quite small, about 3-4%, as compared to the usual community risk of about 1%.

If we know the chromosomal or gene change that caused the heart defect, it can be easier to predict how likely it is that an abnormality might occur in a brother, sister, offspring or parent of the affected child.

In most cases of congenital heart disease, the cause is unknown and there are limited options for genetic testing.

As children with congenital heart disease enter reproductive age, it is important that they engage in healthy, positive and informed discussions with their parents about the heritable aspects of congenital heart disease, as this could have significant implications for future family planning.

More information

Genetic services provide counselling and information for individuals and families with, or at risk of, particular birth defects and inherited disorders. Genetic services offer diagnosis, screening and testing.

Genetic counselling services can provide detailed information about the causes of congenital heart disease and individualised recurrence risks. This is particularly important for families in which two or more family members are affected. Consult your doctor for more information and a referral.

Where can I go for further help?

People with congenital/childhood heart disease face unique challenges, treatment and often repeated surgeries for their entire lives, and support is critical at every stage of the journey – from when congenital/childhood heart disease is diagnosed through childhood, the teenage years and into adulthood.

HeartKids is the only national charity dedicated to supporting Australians of all ages impacted by congenital/childhood heart disease, the leading cause of infant death in Australia and a complex chronic disease requiring lifelong treatment.

For over forty years, HeartKids has provided timely support to infants, young people and adults living with congenital/childhood heart disease all across Australia. We also fund life-saving research, provide reliable and evidence-based information and advocate for the needs of impacted families.

Our support is a commitment for life.

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References for further reading

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For information
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