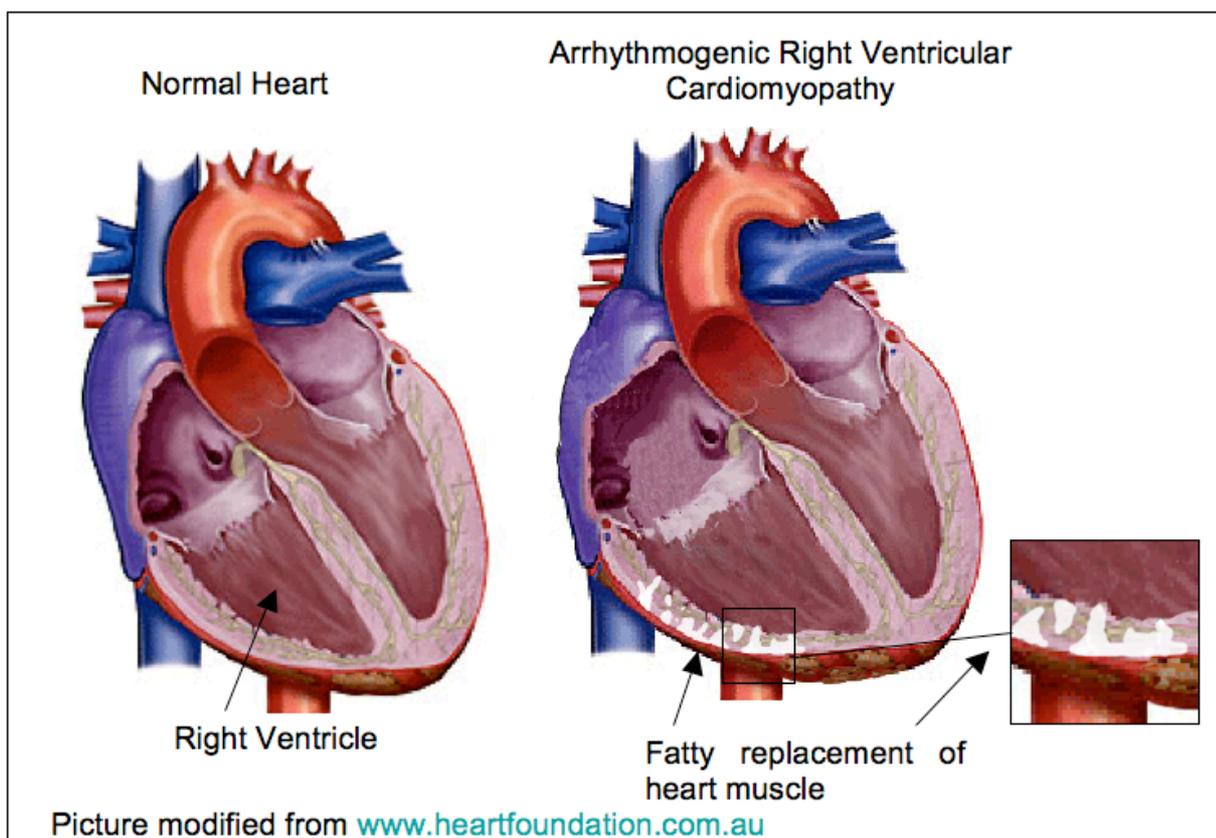


Last updated 06 December 2012

## Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)

### What is Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)?

Arrhythmogenic right ventricular cardiomyopathy or ARVC (previously known as arrhythmogenic right ventricular dysplasia or ARVD) is a condition of the heart muscle, where the normal heart muscle cells on the right-hand side of the heart are replaced by fat and scar tissue. These changes in the heart muscle can cause two main problems. First, it can cause changes in the electrical system of the heart leading to heart rhythm abnormalities. Second, the right side of the heart can become enlarged and not pump as it should, which leads to heart failure. These changes in the heart can lead to a range of symptoms that varies from patient to patient. Some people with ARVC may have no symptoms while others may develop palpitations, chest pain, dizziness and fainting episodes. In the most serious instances, it can lead to heart failure and premature death.

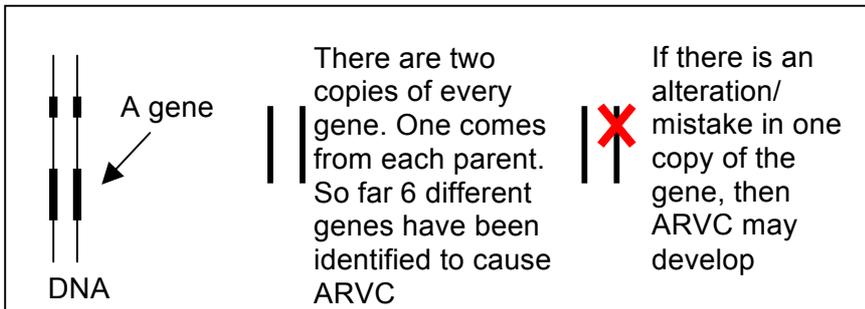


### What causes ARVC?

ARVC is a genetic condition caused by a problem in the genes involved in keeping the heart healthy. Our body is made up of millions of cells. There are many different types of cells, including brain cells, liver cells and heart cells to name a few. Each cell contains 46 chromosomes, which hold the genetic material that decides features such as the colour of our

eyes and whether we are tall or short. These 46 chromosomes are grouped into 23 pairs, one of each pair coming from mum and the other from dad.

If you imagine a chromosome as being like a ball of wool, you could stretch it out into one long strand this would be your DNA. Along the length of DNA there are regions called genes. As there are two copies of every chromosome, there are also two copies of every gene (as

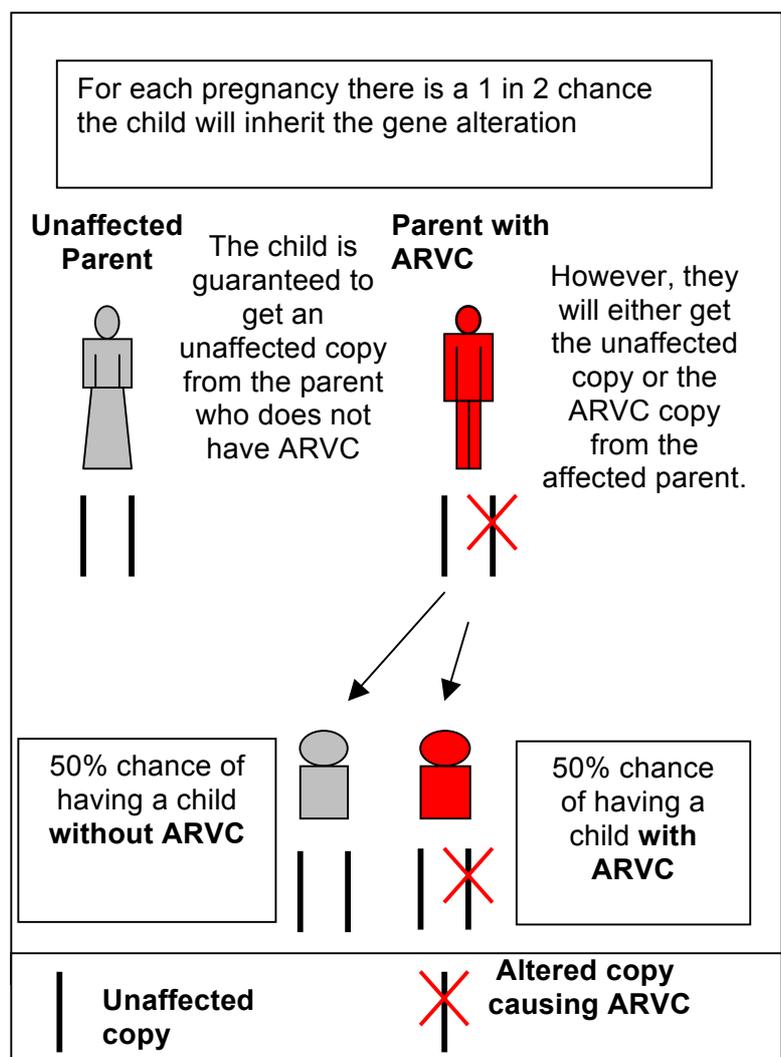


mentioned, one from each parent). Genes act as recipes to make certain things in the body, and each recipe is unique based on the order of units it is made up of. If there is a mistake in one of these genes (a mix-up in the order of these units) it may lead to the

development of ARVC. This mistake is known as a gene alteration. In the case of ARVC we know of alterations in at least six genes that can cause the condition. If there is an alteration in one of these genes ARVC may be result.

### How is ARVC inherited?

ARVC is known as an autosomal dominant condition, whereby the gene with the alteration “dominates” over the unaffected gene to cause the condition. The term “autosomal” means it affects males and females equally. An affected person has inherited one altered ARVC copy of the gene from one parent, and one unaffected copy from the other parent. Likewise, if a person who carries this ARVC copy of the gene has a child, then **the chance that the child will be affected is 50%, or 1 in 2**. This is because they are guaranteed to get an unaffected copy of the gene from the unaffected parent, and they can either get an altered (ARVC) copy or an unaffected copy from the parent with the condition (see diagram right).



Rarely, a person may have a gene alteration that has not been inherited from their parents. Instead, the alteration has arisen some time after conception. This means siblings are not likely to develop the condition, however, the gene alteration can still be passed down to any children of the affected person.

## What is genetic testing?

Genetic testing involves looking for gene alteration in the genes known to cause ARVC. At present, at least six genes have been identified to cause ARVC. Gene testing of five of the known genes is available in centres around the world. If you would like to know more about centres offering commercial genetic testing, please look at our Information Sheet on Cardiac Genetic Services, contact the Registry Coordinator on [registry@centenary.org.au](mailto:registry@centenary.org.au) or (02) 9565 6185 or speak to your regular cardiologist or doctor.

Genetic testing can affect applications for life insurance. If you would like to know more please visit the Financial Services Council website at [www.fsc.org.au](http://www.fsc.org.au) for current guidelines (go to the Search bar and type in “Genetic”) or call (02) 9299 3022.

## Will a diagnosis of ARVC change my lifestyle?

Relatively little is known about the clinical aspects ARVC. Based on the limited available information on this condition, following are the key lifestyle changes and medical treatments recommended for people with ARVC:

- **Avoid competitive sports and strenuous physical activity:** Competitive sports include those that require significant exertion, for example: Touch football, Basketball, Netball, Squash, running and even social games that you may not think are too strenuous. We recommend people avoid competitive sports because it may be associated with severe cardiac symptoms and possible sudden death.
- **Regular light exercise:** It is important to maintain a healthy lifestyle. Regular light exercise, such as walking, is not only good for your heart but also good for your general health.
- **Regular check-ups with your cardiologist:** It is important to monitor the progression of your condition, and you should also report any new symptoms.
- **Consult with your GP or cardiologist before taking any new medications:** It is important to talk to your doctor before taking any new medications, including over-the-counter medications, complementary medicines, natural products or herbal remedies.
- **Encourage relatives to be screened:** As mentioned, parents, siblings (brothers/sisters) and children of an affected person are at risk of also being affected. Screening of relatives involves them seeing a cardiologist where they will have a physical examination, an ECG, an echocardiogram (ultrasound of the heart) and possibly a magnetic resonance imaging (MRI) scan. Find a specialist cardiac genetic service like the Genetic Heart Disease Clinic at RPAH’s Sydney Heart Centre (tel: 02 9517 2011) or speak to your cardiologist about screening family members.

## Further information:

If you have any further questions, please contact:

Your local cardiologist or GP

**Australian Genetic Heart Disease Registry** [www.heartregistry.org.au](http://www.heartregistry.org.au)

Tel: 02 9565 6185 or email [registry@centenary.org.au](mailto:registry@centenary.org.au)

Visit the Registry website today to find out how to enrol. Your participation can help save and improve lives by improving our knowledge of genetic heart conditions in Australia.