

South East Of Scotland Genetic Service
Western General Hospital, Crewe Road, Edinburgh, EH4 2XU
Tel: 0131 537 1116

Genetic Testing – Brugada syndrome

Information for patients

Patient Label

Brugada Syndrome

Brugada syndrome results in a disturbance to the electrical system in the heart which can lead to an abnormal heart rhythm (arrhythmia). If left untreated, these arrhythmias can cause the heart muscle to contract in an ineffective way, reducing the flow of blood from the heart around the body leading to loss of consciousness or in severe cases, cardiac arrest. Symptoms can be variable but can include fainting episodes (syncope) and or heart palpitations (flutters).

Brugada syndrome can sometimes run in families and affect one or more members as a result of a genetic change, or variant. It is important to identify such families as individuals with the condition could be at risk of sudden death. Families at risk should therefore be seen so accurate assessments, diagnosis, treatment and support from specialists can be arranged.

Healthy Living Advice

We know that for people who have or who are at risk of Brugada it is important for them to avoid certain circumstances. These include:

- **A high temperature (fever)** – if you develop a fever of 38C (100.4F) or above, take painkillers such as paracetamol to bring it down; get medical advice as soon as possible if this doesn't help
- **Drinking too much alcohol** – avoid drinking lots of alcohol in a short space of time
- **Dehydration** – get medical advice if you have diarrhoea or vomiting that doesn't go away, as you may lose a lot of fluid and might need to take special rehydration drinks
- **Certain medicines** – make sure any healthcare professional you see knows you have Brugada syndrome, and avoid medicines that can trigger the condition.

There are a number of genes that when altered can be associated with Brugada syndrome. We are able to offer you a genetic test to determine if you have alterations in these genes.

Possible outcomes of gene panel testing:

- **If a genetic change is identified in your sample**, it means we can offer a genetic test to your blood relatives. In most cases, your children, siblings and parents would each have a 50:50 chance of having this gene change. The outcome of their genetic test would ensure their cardiac screening recommendations are appropriate.
- **If a genetic change is not identified in your sample**, we will not have a blood test to offer your relatives. Individual advice would be provided about whether family members should have cardiac screening as not finding a genetic change does not necessarily mean you do not have the condition.
- **Occasionally we find an unclassified variant**. This is a change in a gene where it is not known if it was significant in causing you to have Brugada Syndrome or if it is just a natural variation that does not cause the syndrome. We will not have a blood test to offer your relatives, however personal and familial screening advice would be based on your clinical presentation of the syndrome and family history.

Please tick as appropriate	Plan of action going forward	Enter details
	You have decided to proceed with testing and we have made the following arrangements for you to receive the results:	Letter / phone call /appointment
	You have decided to get back in touch when you feel ready to proceed with testing.	

Following testing, **results are usually available within 3 months**. Results are confirmed in writing to you and copied to your referring clinician/GP.

You were seen by:	Genetic Counsellor (0131 537 1116)	Date:
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For more information and links to support organisations you can look at the website: Network for Inherited Cardiac Conditions Scotland (NICCS) at www.niccs.scot.nhs.uk/



You may also find the following website useful for information regarding up to date advice on medications to avoid with Brugada syndrome: www.brugadadrugs.org



A limitation of the genetic test is that just like much of medical science, we cannot yet know with certainty all the factors that influence health. There is very small chance that current classification of a gene variant may change as more information becomes available. This may change the clinical diagnosis, prognosis or treatment you receive.