

Important information about your *RYR1* gene variant and anaesthetics

This leaflet provides information about your *RYR1* gene variant and what it means when having an anaesthetic. If you have any further questions or concerns, please speak to the staff member in charge of your care.

Why have I been given this leaflet?

You have a 'variant' or change in a gene called *RYR1*. Some *RYR1* gene variants are linked (or might be linked) with an inherited susceptibility to a condition called malignant hyperthermia (MH).

This leaflet explains more about MH and the precautions you should take. We recommend that you follow the advice in this leaflet even if your *RYR1* variant does not have a clear link with MH.

What is malignant hyperthermia (MH)?

- MH is a severe reaction to some anaesthetic drugs and gases. If untreated, MH can lead to death.
- MH is rare. It occurs in 1 in 30,000 to 1 in 200,000 general anaesthetics.
- Early signs of MH include a rapid heart rate and increased carbon dioxide levels. As the reaction progresses, body temperature rises, muscles become rigid, and heart rhythms can become irregular.
- Early treatment is usually successful. Treatment involves discontinuing the anaesthetic, cooling the patient and giving dantrolene. Dantrolene is an antidote available in all hospitals where anaesthetics are used.
- Very rarely MH can be triggered by stresses on the body, such as vigorous exercise or heat.

Who gets MH and why?

Susceptibility to MH most often occurs in people who have spelling differences (variants) in the *RYR1* gene, but variants in other genes have also been linked with MH susceptibility.

It is usual to find lots of gene variants in the *RYR1* gene. We are not always sure which ones cause MH and which ones do not. We might look at the genes and medical history of several members of a family to work out which gene variants are linked with susceptibility to MH. We can also see if other genetics services have found the same gene variant in another person and linked it to MH susceptibility.

Susceptibility to MH is hereditary, which means that *RYR1* variants linked with it can be passed down through the family. People with a *RYR1* gene variant have a 1 in 2 chance of passing it on to any children.

Your *RYR1* gene variant and precautions

You are susceptible to MH if you have (1) experienced an episode of MH or (2) have an *RYR1* or other gene variant linked with MH susceptibility. If we are not sure whether your *RYR1* variant is linked with susceptibility to MH, it is safest to take the same precautions:

- **Inform your anaesthetist:** Tell your anaesthetist that you are at risk of MH if you need to have a general anaesthetic. Keep an MH warning card in your wallet and wear a warning disc, such as MedicAlert, in case you are brought into hospital unconscious.
- **Communicate with your healthcare providers:** Ensure all your healthcare providers are aware you are at risk of MH. Let your genetics team know if anyone in the family experiences MH symptoms.
- **Family awareness:** Discuss this information with family members. They may also carry the gene variant and could benefit from genetic counselling and testing. If they have not been tested, your relatives should be treated as if they have MH and avoid certain anaesthetic drugs and gases.

Diagnostic testing

You may be offered a muscle biopsy test known as the in vitro contracture test (IVCT). This can help find out whether your *RYR1* gene variant is linked to MH susceptibility.

The test involves an overnight stay at a hospital in Leeds. A small sample of muscle tissue is removed and then exposed to anaesthetic drugs to observe its reaction.

The IVCT is invasive. It carries risks such as wound infection, bleeding and sensory loss. The test is not 100% accurate. However, it can provide information to guide your medical care and inform family members about their risks.

Things to avoid in day-to-day life

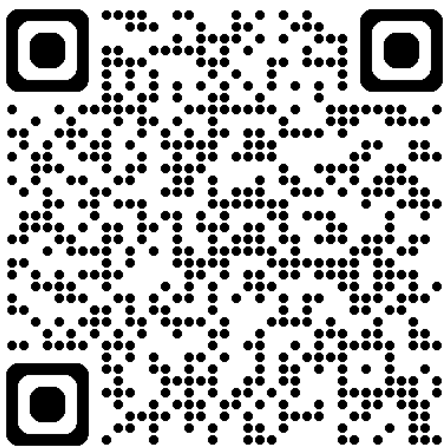
Some people who are susceptible to MH find that heat or exercise can trigger symptoms. They and their family members should avoid the activity that they find triggers symptoms. People who have not experienced adverse effects from heat and exercise do not need to avoid these activities.

How can your relatives access genetic testing?

You may be referred to your regional genetics service to discuss the option of genetic testing because a relative has an *RYR1* gene variant or you have a family history of MH. If you or your relative experience an episode of MH, please ask your GP or hospital specialist to refer you to genetic services.

Additional Resources

Royal College of Anaesthetists factsheet on Malignant Hyperthermia. Available at:



Contact us

For patients in the South West Thames region:

Department of Clinical Genetics
Southwest Thames Centre for Genomics
St. George's University Hospitals NHS Foundation Trust
Blackshaw Road
London
SW17 0QT
Telephone number: 0208 725 2805
E-mail: genetic.secretaries@stgeorges.nhs.uk
Website: www.stgeorges.nhs.uk/service/clinical-genetics/clinical-genetics/

Additional services

Patient Advice and Liaison Service (PALS)

PALS can offer you advice and information when you have comments or concerns about our services or care. You can contact the PALS team on the advisory telephone line Monday, Tuesday, Thursday and Friday from 2pm to 5pm.

A Walk-in service is available:

Monday, Tuesday and Thursday between 10am and 4pm

Friday between 10am and 2pm.

Please contact PALS in advance to check if there are any changes to opening times.

The Walk-in and Advisory telephone services are closed on Wednesdays.

PALS is based within the hospital in the ground floor main corridor between Grosvenor and Lanesborough Wing.

Tel: 020 8725 2453 **Email:** pals@stgeorges.nhs.uk

NHS UK

The NHS provides online information and guidance on all aspects of health and healthcare, to help you make decisions about your health.

Web: www.nhs.uk

NHS 111

You can call 111 when you need medical help fast but it's not a 999 emergency. NHS 111 is available 24 hours a day, 365 days a year. Calls are free from landlines and mobile phones.

Tel: 111

AccessAble

You can download accessibility guides for all our services by searching 'St George's Hospital' on the AccessAble website (www.accessable.co.uk). The guides are designed to ensure everyone – including those with accessibility needs – can access our hospital and community sites with confidence.