I am MH-Susceptible—Now What?

A Roadmap to Answers



Provided by

The Malignant Hyperthermia Association of the United States (MHAUS)

Table of Contents

I am MH-susceptible (MHS) — NOW WHAT? **2**

Connecting with Other MHS Individuals **3**

Testing Options **4**
- Muscle Biopsy Testing Information
- MH Genetic Testing Information
- Choosing Not to be Tested

Genetic Counselor & Insurance Information **7**

Healthcare Professionals Information **8**
- MH Hotline
- MHAUS Administration Contact Info
- North American MH Registry of MHAUS Contact Info
- MHAUS Website Availability

Additional Resources **9**
- MHAUS Membership Options
- The North American MH Registry of MHAUS (NAMHR)
- Neuroleptic Malignant Syndrome Information Service (NMSIS)

Patient Safety Checklist **10**

MH Triggering Anesthetics **12**

I am MH-Susceptible — NOW WHAT?

What does this mean to me and my family?

Where can we find helpful resources and information?

What is the difference between an MH muscle biopsy and MH genetic test?

Which test is the best choice for me?



First, **you have options** as to how to proceed and your personal preferences will govern which option you choose to pursue. Reading this resource is a good start.

Some things to think about as you consider your next steps:
MH-susceptible (MHS) individuals do not outwardly "look" any different from anyone else. Malignant hyperthermia (MH) usually happens in a surgical environment where gaseous anesthetics and/or succinylcholine, either of which could trigger the disorder, are used.

It may happen rapidly, gradually, or not at all in a particular surgical procedure, then may develop during a later surgery, for some reason.

MHS patients may be at increased risk for exercise/exertion-related, heatstroke and other heat-related disorders – MHAUS is following this issue more closely and will update our educational materials as it becomes available. People with MH have reported a variety of physical issues from cramping to muscle pain to heat sensitivity. More about this can be found at www.mhaus.org.

Connecting with Other MHS Individuals

Is there some place that has helped other MH-Susceptible (MHS) families?

We encourage all MHS individuals to contact the **North American MH Registry of MHAUS** via the link on the MHAUS website or by calling **888-274-7899.** Talk with staff there to determine if you should place your information in the database for future generations' use as a resource and for possible future MH research.

How can I connect with others who have this disorder to "compare notes?"

MHAUS has an active **Patient Liaison Committee** involving other MHS individuals and their family members who have experienced the concerns and questions that follow an MH diagnosis. They could provide valuable insight and direction. Visit the MHAUS website at **www.mhaus.org** to connect with them and learn more about the group. There are also various **Chapter Groups** you can join on the MHAUS website – go to the "Get Involved" button at the top of the page.

Should I be tested?

If you know you have MH, you may not 'need' to be tested. However, you may want to consider testing for the sake of family members. A muscle biopsy can be the most definitive test, but is much more invasive than the genetic test. However, you may be one of those individuals with a rare genetic variance for MH which will not be detected. The majority with MH will find a variance for MH in genetic testing.

Testing Options

Which testing options are available to me?

Muscle Biopsy (CHCT) Test

(Caffeine Halothane Contracture Test, or CHCT) is the gold standard MH test and provides the most relevant clinical data for healthcare professionals and your family, with 78% overall accuracy. This test is invasive and a small section of muscle is surgically removed from the thigh, taken immediately to that facility's lab for specifically-designed tests that subject the muscle section to MH triggers. Measurements are clinically taken to track the response levels of the muscle to the triggers in order to assess MH susceptibility level. A resource can be found at: https://www.mhaus.org/testing. Here you will be able to view an actual muscle biopsy procedure video and other related information. MH experts generally feel a child less than 10 years old is not a good candidate for CHCT testing.

NOTE: MH Muscle biopsies **cannot be done outside the CHCT testing centers listed on the MHAUS website.** Muscle MUST be processed immediately following removal from patient – at the same location!

CHCT (muscle biopsy) testing is available in Minnesota, Maryland *(military personnel and their families only)* **and Toronto.**
See https://www.mhaus.org/testing/muscle-biopsy-chct/ for the current list and contact information.

Scheduling an MH muscle biopsy test may take from 2 – 8 weeks or more in order to coordinate the surgeon, anesthesia provider, and lab personnel for the procedure date. The testing center Director and staff will be very helpful with management of your appointment. Feel free to call any of the Muscle Biopsy Testing Centers listed on our website for guidance.

MH Genetic Test

For this option, a genetic counselor is a good first contact!
Genetic testing may be the right choice for preventive and life planning measures, and to maximize your knowledge of the benefits and limitations of genetic testing. After consulting a genetic counselor, you may even determine genetic testing is not the appropriate first step for you and your family members.

Genetic counselors have the best sense of what items and data will be needed if you decide genetic testing is your chosen route for answers. If you know of a genetic counselor nearby, that may be your preferred place to start. If not, the *National Society of Genetic Counselors'* website at www.nsgc.org can help you locate a genetic counselor near you. **Important: Insurance coverage for services rendered by a genetic counselor and for genetic testing differs from company to company and state to state. It is highly recommended you check with your personal insurance provider, when considering the genetic testing option, to determine what charges they will cover and what costs you personally might incur!** Your genetic counselor may be able to help you in this process.

For instance, in the past *Aetna* (www.aetna.com) considered genetic testing for MHS individuals medically necessary for either: 1) screening clinically confirmed MHS patients for variants in the RYR1 gene considered as causative for MH by the European Malignant Hyperthermia Group (EMHG) or 2) screening at-risk relatives of patients with clinically confirmed MHS for a known familial variant in the RYR1 gene considered causative by EMHG; United Healthcare did reimburse for molecular diagnostic and genetic testing but again, check your specific plan for what portion of the testing cost will be covered; Blue Cross/Blue Shield covered genetic testing if pre-authorized by the healthcare provider. Again, check your personal plan's specific parameters as they may have changed.

Tip: Be sure to **bring your insurance card with you** for this appointment to expedite the process.

Choosing not to be Tested for MH

Should you choose not to be tested and prefer to consider yourself MH-Susceptible, this is another option available to you. MHAUS recommends verifying this through testing, but the choice is yours.

If you decide to take this path, **please be sure to make it a point to alert your family members and healthcare professionals involved in your care of this personal health concern.** It is recommended that all MH-susceptible individuals inform their surgeon, anesthesiologist and caregivers of their MHS status prior to undergoing any in-hospital or outpatient procedure. MHAUS has letters available on our website pre-formatted for your use to send to those who should know of your MH susceptibility and we encourage you to feel free to use them. Additionally, an MH ID tag of some sort is highly recommended to speak for you if you should ever become unable to speak for yourself. A link to MedicAlert's ID tag program can be found on the MHAUS website under the Patients Tab at the top of the page.

BE PREPARED!



Genetic Counselors & Insurance Info

Genetic Counselors

MHAUS is presently aware of 2 genetic counselors that are experienced in MH genetics and have personally helped a number of MHS maneuver through the maze of genetic options and questions.

Deanna Steele, CGC
Affiliated with University of Pittsburgh Medical Center, Pittsburgh, PA
PH: 412-692-7273
Email: deanna.steele@chp.edu

Christina Zaleski, MS, CGC
PreventionGenetics, Marshfield, WI
PH: 715-387-0484, x158
Email: christina.zaleski@preventiongenetics.com

Insurance Provider Information

After speaking with a genetic counselor, and/or a testing center director, if they agree that genetic testing is the right path for your personal situation, you may request a Letter of Medical Necessity to share with your contact at your insurance company; it outlines the specific codes your insurance provider will need to:

1. Consider your eligibility and
2. Determine how much they will cover based on your personal situation and plan for their pre-authorization.

A template of this letter is available on the MHAUS website on the Testing page at http://www.mhaus.org/patients-and-families/sample-letters. Fill in the blanks with information provided by the genetic counselor and print or download, as needed.

If, after considering what is covered in your particular case, you decide you should move forward with an MH genetic test, your genetic counselor will help you by arranging the following steps:

1. Set up a blood draw **(a doctor's prescription is required)**
2. Send it to a suitable genetic testing laboratory
3. Request results be sent to your genetic counselor & physician(s)
4. Interpret the results
5. Explain the results to you over the phone or at a follow up appointment and then help you assess future health plans

For Healthcare Professionals

All healthcare professionals dealing with an MH-susceptible patient are encouraged to openly communicate and share the previous resources with their patient to assist in determining which testing options are indicated in their particular situation, as well as genetic counselor resources to help in their search for answers, insight, and further discussion.

MHAUS provides a free 24/7/365 **MH Hotline at 800-644-9737** within North America and 209-417-3722 outside North America with MH experts available to assist you in your efforts to keep your patient safe when MH unexpectedly strikes.

If appropriate, you may direct your patient to call MHAUS administrative offices at **607-674-7901 or 800-986-4287** for information/assistance, **and/or** the *North American MH Registry of MHAUS* at **888-274-7899** if they want to determine if their family information should be placed in the Registry's confidential database.

Additional resources on MH testing options and other helpful information, **as well as MHAUS membership options for you to be informed and ready for MH,** can be found on the MHAUS website at www.mhaus.org.

We would like to hear from those who have gone through either type of testing and are willing to share personal experiences with others who may be wrestling with decisions about next steps that are available to them when they are told they should consider themselves MH-susceptible. Call **800-986-4287** to share your feedback and stories with others who may be searching for answers as well. Another way to share is to go to the "Get Involved" tab on the home page of MHAUS and click "Faces of MH".

For anyone who must deal with MH susceptibility in their family, the MHAUS website offers additional references and resources as to what facilities have taken the initiative to assure they are prepared for MH by either studying about MH and taking an online test to assure the material is understood **or** by holding an **MH Prep Check,** which involves bringing an MH expert to their facility to watch them perform an MH Mock Drill and review their plan of action in order to assure they are prepared to treat an MH emergency.

There are many resources online to help you through this often confusing topic. Another resource we have found helpful is Genetic Alliance at https://geneticalliance.org. This is just one of the plethora of internet options for further reference and resources available to all.

Let us know if we can be of any further help by contacting us. We are here to help!

Open Communication is a Key to Patient Safety

Additional Resources

MHAUS maintains and regularly updates a website at www.mhaus.org.

Whether a Healthcare Professional or Patient, we offer a wealth of MH information and education on our website. Our visitor's experience ranges from casual browsing for basic MH info and resources, to logging in for additional access, or to **become an MHAUS member to take advantage of membership benefits.** The website shares:

- ***Membership options***
  - Elite ($100/year) with **automatic updated MH educational material**
  - Lifetime ($500) with **automatic updates and no need to renew**, MH-susceptibles and/or family members ($35/year)
  - Members receive 30% off most items ordered
  - U.S. Military membership 50% off

- ***Blog, Facebook and Twitter Updates to share:***
  - Topics of interest to MH-Susceptibles and their family members
  - Share feedback and experiences from MH-Susceptibles
  - Updates from within the MHAUS' administrative office as well as from MH Experts to help clarify and enrich understanding of MH and related disorders

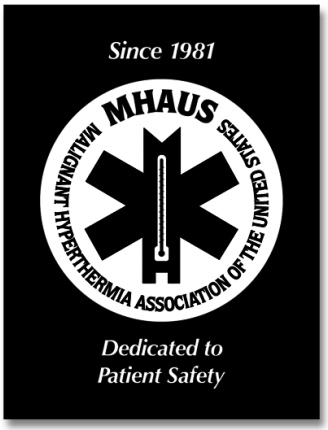
- ***Video clips and Webinars***
  - Regarding dantrolene
  - Guide to Running an MH Mock Drill
  - Managing an MH Crisis
  - Testing Options
  - How to Clean an Anesthesia Machine
  - *Actual Muscle Biopsy for MH* (Caffeine Halothane Contracture Test) written in English and other languages
  - And much more

Additional Resources continued

The North American MH Registry of MHAUS (NAMHR) is the research arm of MHAUS now at the University of Florida – Gainesville, FL which, maintains stores and analyzes MH-susceptible (MHS) and similar syndrome data in a central database, seeks to improve diagnosis, treatment, and prevention of MH. **Contact them** to include your personal de-identified MH-susceptible medical data to assist your family members and future research at **800-274-7899.**

Neuroleptic Malignant Syndrome Information Service (NMSIS) is a subgroup of the MHAUS organization and serves as an international resource center for knowledge on neuroleptic malignant syndrome in addition to other heat-related disorders and drug side effects. NMSIS has a main presence on the MHAUS website and offers NMS educational materials to psychiatrists, ED personnel and social workers who may deal with patients experiencing this debilitating disorder, as well as their family members who are concerned for their wellbeing.

Visit our website at www.mhaus.org to be aware of any updates or ask us to add your email to our *MH Hot Topics* e-Newsletter list so you can receive the recent updates from within MHAUS and the MH World every few months.



Patient Safety Checklist

Malignant Hyperthermia (MH) is a potentially fatal, inherited disorder usually associated with the administration of certain general anesthetics and/or the drug succinylcholine. The disorder is due to an acceleration of metabolism in skeletal muscle.

The signs of MH include:

- Muscle rigidity
- High body temperature
- Increased acid content
- Rapid heart rate
- Muscle breakdown

Without proper and prompt treatment with dantrolene, mortality is extremely high. The best way to protect yourself, your patients and your facility, is to be prepared before it's too late.

Malignant Hyperthermia Association of the United States (MHAUS) is a 501 (c) (3) nonprofit organization. The mission of MHAUS is to promote optimum care and scientific understanding of MH and related disorders.

For more information visit our website – www.mhaus.org

For my peace of mind, please clarify the following questions to assure my safety for my upcoming procedure.

1. Will I be first in the OR? _____
2. Has the anesthesia machines been flushed according to MHAUS recommendations? _____
3. When was your last MH Mock Drill?
   (MH Mock Drills should be conducted annually) _____
4. How long will I be monitored after surgery? _____
5. Is the MH hotline number available? _____
6. Is there dantrolene available? _____
7. Is the MH Cart checked and updated regularly? _____
8. Do you have the latest Emergency Therapy for MH Poster? _____
9. What anesthetics will you use during my procedure?
   See list next page. _____

MH Triggering Anesthetics

DO NOT USE:

Inhaled General Anesthetics

Desflurane

Enflurane

Ether

Halothane

Isoflurane

Methoxyflurane

Sevoflurane

Depolarizing Neuromuscular Blocker:

Succinylcholine



MHAUS is a nonprofit organization devoted to the study of MH, to patient and practitioner education, and to increasing the safety of MH-susceptible individuals. Your contribution to MHAUS will support these goals and possibly save a life!

For more information or questions, feel free to contact MHAUS:

Malignant Hyperthermia Association of the United States
1 North Main Street | PO Box 1069 | Sherburne, NY 13460
607.674.7901 | www.mhaus.org | info@mhaus.org