

I am MH-Susceptible Now What?

A Roadmap to Answers

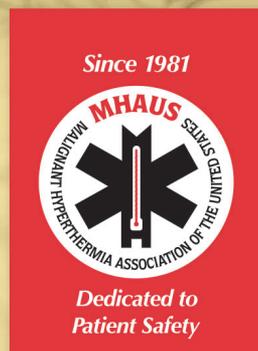


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MHAUS is a nonprofit organization devoted to the study of MH, to patient and practitioner education, and to increasing the safety of MH-susceptible individuals. Your contribution to MHAUS will support these goals.

Go to mhaus.site-ym.com/donations to make a donation of any amount toward MHAUS’ efforts to produce MH resources, such as this Roadmap to Answers.

I Just Found Out I am MH-Susceptible

Now what? Tell me...

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 volatile anesthetics and/or succinylcholine, either of which could trigger the disorder, are used.

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

MHS patients may be at increased risk for exercise/exertion-related heatstroke and other heat-related disorders. People with MH have reported a variety of physical issues, e.g., cramping muscle pain and heat sensitivity. More about this can be found at www.mhaus.org



Connecting with Other MHS Individuals

Is there someplace that has helped other MH-Susceptible 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** consisting of 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 <https://mhaus.site-ym.com/?page=PLC> to connect with them and learn more about the group.

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 special MH muscle biopsy (CHCT) can be the most definitive test, however, it is invasive and can only be done at a few select centers in the United States and Canada. Genetic testing typically involves a simple blood “draw” or mouth swab (some laboratories use saliva specimens), it is less expensive than the muscle biopsy and can be ordered through your doctor’s office or by seeing a genetic counselor. Unfortunately, genetic testing isn’t able to detect all cases of MH. Genetic testing is informative in approximately 60-80% of families where the causative genetic variant can be identified. It is especially useful if a genetic change has previously been identified in another family member. Talking with a genetic counselor can help you determine if genetic testing is right for you and your family.

MUSCLE BIOPSY 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 requires either regional or general anesthesia to surgically remove a small section of muscle from the thigh, which is then taken immediately to that facility’s lab for specifically-designed tests that subject the muscle section to MH triggers. Measurements are 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 tested immediately following removal from patient – at the same location!

CHCT (muscle biopsy) testing is available in California, Maryland, Minnesota, North Carolina and Toronto. See <https://www.mhaus.org/testing/muscle-biopsy-chct/chct-muscle-biopsy-testing-centers/> for current list and contact information. There are also several centers in Europe, who test for MH Susceptibility. See EMHG.org for locations.

MH Genetic Testing

Scheduling an MH muscle biopsy test may take from 2–8 weeks in order to obtain insurance coverage and 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. Forms you will need (CHCT Test Form Information and Request for Medical Records) can be found at the MHAUS website at <http://www.mhaus.org/patients-and-families/sample-letters>.

MH GENETIC TEST

For this option, a genetic counselor is a good first contact or the Director at one of the testing centers. 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. The director of the MH testing center will, however, need to provide the final approval and obtain consent prior to testing.

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](#), www.nsgc.org, www.aetna.com and [EMHG: United Healthcare](#) can help you locate a genetic counselor near you. Some of the testing laboratories listed on the MHAUS website have genetic counselors to assist 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 when considering the genetic testing option, that you first check with your personal insurance provider 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, Aetna (www.aetna.com) considers genetic testing for MH is indicated in the following groups:

1. patients with a positive or equivocal contracture test to determine the presence of a specific mutation,
2. individuals with a positive genetic test for MH in a family member, and
3. patients with a clinical history suspicious for MH (acute MH episode, masseter muscle rigidity, post-operative myoglobinuria, heat- or exercise-induced rhabdomyolysis) who are unable or unwilling to undergo contracture testing.

Tip: Be sure to bring your insurance card to this appointment to expedite the process.

MHAUS is presently aware of 3 genetic counselors who are experienced in MH genetics and have personally helped a number of MHS maneuver through the maze of genetic options and questions.

Jennifer L. Geurts, MS, CGC, Medical College of WI, Human & Molecular Genetics Center, Milwaukee, WI (414) 955-5889, jgeurts@mcw.edu

Deanna Steele, CGC, Children's Hospital of Pittsburgh of UPMC, Pittsburgh, PA (412) 692-7273, deanna.steele@chp.edu

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

MH Genetic Testing

After speaking with a genetic counselor, and/or testing center director, and 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 Patients page at 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**) or obtain a specimen container for a sputum sample if the laboratory supports such specimen testing
2. send it to a suitable genetic testing laboratory, such as those listed on the MHAUS website
3. request results to 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

I don't want to be tested and choose to just consider myself MH-Susceptible

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. Be aware that this choice means that all of your closest relatives will also be considered MH-susceptible unless they undergo MH muscle biopsy testing to rule out MH-susceptibility.

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 MH-susceptible individuals inform all their caregivers 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:

<http://www.medicalert.org/groups/malignant%20hyperthermia>.

Be prepared!



MH Genetic Testing

For Healthcare Professionals

All healthcare professionals caring for an MH-susceptible patient are encouraged to openly communicate and share the above 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 help keep your patient safe when MH unexpectedly strikes.

If appropriate, you may direct your patient to call the 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 it is determined 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 to keep you 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 their experiences with others who may be wrestling with the options available to them when 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.

For anyone who must deal with MH susceptibility in their family, the MHAUS website offers additional references and resources as to which facilities have taken the initiative to be prepared for MH by either studying about MH and taking an online test to confirm 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 be confident they are ready to handle an MH emergency.

When patients are deciding where to have a surgical procedure done, this list might be helpful to them in choosing a "prepared" location.

There are many resources online to help you through this often confusing topic. Another resource we have found helpful is Genetic Alliance at www.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



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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:

- **List of facilities** (city and state) whose staff have displayed enough interest in MH to purchase In-service Training and successfully complete the online test of their MH knowledge and/or have completed an MH Prep Check (brings an MH expert to the site to assess their MH plan of action) in order to act as a resource for MH-susceptibles and their families when they are considering facility options for their personal surgical needs.
- **Membership Options:**
 - Elite* (\$100/year) **with automatic updated MH educational literature.**
 - Lifetime* (\$500) **with automatic literature updates and no need to renew.**
 - MH-susceptibles and/or family members* (\$35/year).
 - Members receive 30% off most items ordered and free access to monthly webinars on topics of interest.*
 - U.S. Military membership 50% off.*
- **Blog, Facebook and Twitter Updates** to share topics of interest to MH-susceptibles and their family members, feedback and experiences from MH-susceptibles, and updates from within the MHAUS administrative office as well as from MH Experts to help clarify and enrich understanding of MH and related disorders.
- **Translations of multiple MH reference materials**
- **Historical video clips** regarding dantrolene, MH-susceptible pigs clinical data, memories from thought leaders
- **Video clip of an actual Muscle Biopsy for MH** (Caffeine Halothane Contracture Test) written in English and other languages

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 and seeks to improve the 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 website at www.nmsis.org 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 well being.

Visit our website at www.mhaus.org for any updates or ask us to add your e-mail 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, rapid heart rate, high body temperature, muscle breakdown and increased acid content. 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 questions below to assure my safety for my upcoming procedure:

1. **Has the anesthesia machine(s) been flushed according to MHAUS recommendations?** _____

2. **When was your last MH Mock Drill?** _____
(MH Mock Drills should be conducted annually)

3. **How long will I be monitored after surgery?** _____

4. **Is the MH Hotline Number available?** _____

5. **Is there dantrolene available?** _____

6. **Is the MH Cart checked and updated regularly?** _____

7. **Do you have the latest Emergency Therapy for MH Poster?** _____

8. **What anesthetics will you use during my procedure?** _____
(See list on next page for list of triggering anesthetics)

MH Triggering Anesthetics – Do Not Use

Inhaled General Anesthetics:

Desflurane

Enflurane

Halothane

Isoflurane

Methoxyflurane

Sevoflurane

Depolarizing Neuromuscular Blocker:

Succinylcholine



The Malignant Hyperthermia Association of the United States (MHAUS)

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

607-674-7901 or **800-986-4287**

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Sherburne, NY 13460

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