

I AM MH-SUSCEPTIBLE - NOW WHAT?

A ROADMAP TO ANSWERS



Provided by

The Malignant Hyperthermia Association of the United States (MHAUS)

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

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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 gaseous 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*, then *may develop* during a later surgery, for some reason.

Recent research is beginning to show that MHS patients may be at increased risk for exercise/exertion-related, heatstroke and other heat-related disorders – MHAUS is following this closely and will report more as it becomes available.

IS THERE SOME PLACE THAT HAS HELPED WITH 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** involving other MHS individuals and their family members who have experienced the concerns and questions that follows an MH diagnosis and 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. 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.

WHAT ARE MY OPTIONS?

You have choices as to what testing, if any, you may want to pursue in order to assure you have the most complete picture of what a malignant hyperthermia diagnosis means to you, your family and healthcare providers.

WHICH TESTING OPTIONS ARE AVAILABLE TO ME?

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 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: <http://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 California, Maryland, Minnesota, North Carolina and Toronto. See <http://www.mhaus.org/testing/centers> for current list and contact information.

Scheduling an MH muscle biopsy test may take from 2 – 8 weeks 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. Forms (*CHCT Test Form Information and Request for Medical Records*) you will need 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! 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, *Aetna* (www.aetna.com) considers 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; *UnitedHealthcare* does 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* covers genetic testing if pre-authorized by the healthcare provider. Again, check your personal plan's specific parameters.

Tip: Be sure to **bring your insurance card with you** for 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.

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Deanna Steele, CGC, affiliated with University of Pittsburgh Medical Center, Pittsburg, PA ☎ 412-692-7273 deanna.steele@chp.edu

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

After speaking with a genetic counselor, and if they agree that genetic testing is the right path for your personal situation, you will be given an important **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 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.

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 and anesthesiologist 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 [[http://www.medicalert.org/groups/malignant%20 hyperthermia](http://www.medicalert.org/groups/malignant%20hyperthermia)]. **Be prepared!**

FOR HEALTHCARE PROFESSIONALS

All healthcare professionals dealing with 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 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 for you 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. Another way to share is to go to the "Get Involved" tab on the home page of MHAUS and then 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 ready for MH.

The facilities who have taken the extra step to be prepared for MH are listed on the MHAUS website. When deciding where to have a necessary procedure done, this list might be helpful to make a choice of facility for your surgery.

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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