HEREDITARY INCLUSION BODY MYOPATHY

PATIENT INFORMATION

WHAT IS HIBM?

HIBM is a rare genetic disease that causes muscles to slowly waste away. HIBM is not life-threatening but it may lead to physical debilitation within two decades of diagnosis. Symptoms usually begin to develop in early adulthood, between late teens to early 30's. HIBM is most common in the Iranian Jewish and Japanese communities, but affects individuals of all ethnicities.

WHAT CAUSES HIBM?

HIBM is caused by a mutation in a single gene called GNE. GNE encodes the enzyme responsible for producing sialic acid, an important component of muscle function. The body's failure to produce enough sialic acid causes muscles to slowly weaken.

OTHER NAMES FOR HIBM

Officially called GNE Myopathy, commonly known as HIBM.

ALSO KNOWN AS:
Nonaka Myopathy
DMRV – Distal Myopathy with Rimmed Vacuoles
QSM – Quadriceps Sparing Myopathy
HIBM2 – Hereditary Inclusion Body Myopathy Type 2
IBM2 – Inclusion Body Myopathy Type 2

understanding THE GENETICS

HIBM is an autosomal recessive genetic disease, meaning that both parents must be carriers of the disease in order to pass it on to their children. Carriers are unaffected; they do not have symptoms.

When two carriers have a child there is a

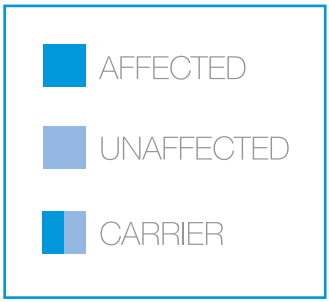
25% chance that each child will be affected by HIBM

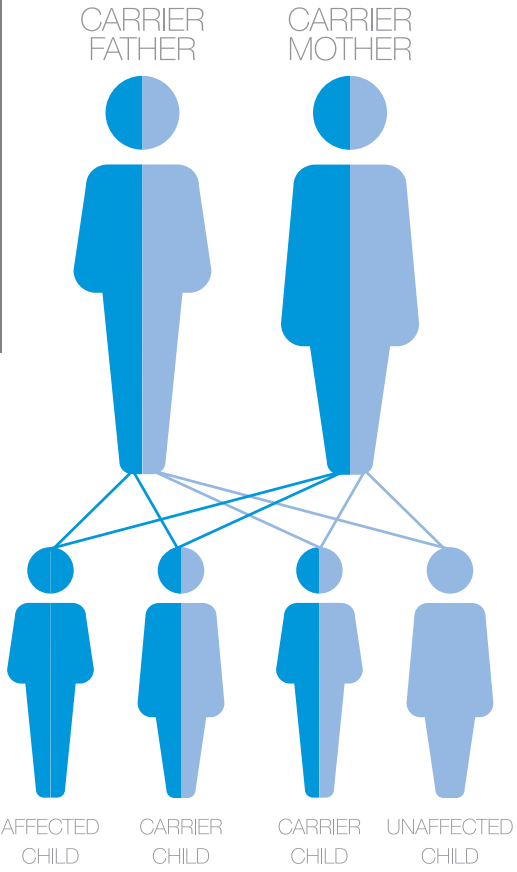
50% chance that each child will be a carrier like his or her parents

25% chance that each child will not carry any mutation for the disease, nor be affected

are you newly DIAGNOSED?

Resources exist that can offer you support (see back page for more information). It's not uncommon to be overwhelmed with a new diagnosis. Fortunately, you are not alone. Resources exist that can offer you support.





answering your **QUESTIONS**

HOW COMMON IS HIBM?

This rare genetic disorder is found among all ethnicities. However, this disease disproportionately affects the Iranian Jewish community.

About **1 in 15** Iranian Jews is a carrier of this mutation

About **1 in 1000** Iranian Jews have the disease

WHAT ARE THE SYMPTOMS OF HIBM?

Early signs and symptoms include foot drop, difficulty running or walking, frequent loss of balance, tripping, and weakness in the index finger. As time progresses, weakness may involve the hand, shoulder and neck muscles.

In most cases, muscles used in the face, the eyes, for breathing, for digestion, and for the heart are completely unaffected. Organs remain intact and unaffected.

HOW IS HIBM TREATED?

Unfortunately, there is currently no approved treatment or cure available to treat or slow the progression of HIBM. Today's treatment options are geared toward managing symptoms. These options may include consultations with physical and occupational therapists, as well as annual visits to a neuromuscular specialist. In addition, mechanical aids may be used to improve physical ability.

Clinical trials, patient registries and natural history studies are currently underway. To learn more about these opportunities, please visit www.clinicaltrials.gov and www.gnem-dmp.com.

HOW CAN HIBM BE PREVENTED?

Genetic screening is the only way to prevent future generations from being affected by this disease. It is imperative to seek genetic counseling if you are a young adult who is affected, a carrier, or at risk of being a carrier. Taking this precaution is especially important prior to starting a family.

Due to advancements in technology, two carriers can now conceive a healthy child. Please discuss your reproductive options with your physician, who can then refer you to a genetic counselor.

TREATMENT Patients should discuss treatment strategies with their physicians. Please contact Dr. Perry Shieh or Dr. Tahseen Mozaffar to further discuss your diagnosis, receive medical advice, and obtain referrals to other specialists in your area.

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GENETIC COUNSELORS These professionals help individuals and families make informed reproductive choices by educating them on the nature, inheritance and implications of genetic disorders.

GENETIC COUNSELING AND SCREENING LOCATIONS:

Advancement of Research for Myopathies, Reseda, California (818) 789-1033*
Cedars-Sinai Medical Genetics Institute, Los Angeles, California (310) 423-9914
UCLA Prenatal Diagnosis Center, Los Angeles, California (310) 825-0300

All genetic counseling sessions are confidential.
For other screening locations, please visit http://www.ndf-hibm.org.

SUPPORT RESOURCES There are a number of support options available to you. Whether you are affected by HIBM or know someone who has been diagnosed, the ability to share experiences may help fight feelings of isolation.

"HIBM/GNE Myopathy Support Group" on Facebook
Living with HIBM: www.livingwithhibm.com
Princess HIBM: http://princesshibm.blogspot.com
Tara Talks HIBM: http://taratalkshibm.blogspot.com
More support options at www.ndf-hibm.org

COUNSELING SERVICES Patients and their families might find it helpful to seek counseling services to help manage their feelings, which may range from shock, denial, anger, depression and isolation. Counseling can also help patients and family members address their individual concerns and empower them to adjust and cope with their diagnosis.

For a list of therapists who are educated about HIBM, the cultural concerns and possible stigma associated with an HIBM diagnosis, please reach out to Carol Hakimi Gelbard, LCSW clhawk13@aol.com. All communications are private and will be held in the strictest confidence.

OTHER ORGANIZATIONS THAT MAY BE HELPFUL:

Advancement of Research for Myopathies (www.hibm.org)
Genetic Alliance (www.geneticalliance.org)
Global Genes Project (http://globalgenes.org)
Muscular Dystrophy Association (www.mdausa.org)
National Organization of Rare Disorders (www.rarediseases.org)
Sephardic Health Organization for Referral and Education (www.shoreforlife.org)
Treat NMD Neuromuscular Network (www.treat-nmd.eu)

*Screening only.