NEUROMUSCULAR DISEASE FOUNDATION

A WORLD FREE OF GNE MYOPATHY TODAY AND FOR GENERATIONS TO COME.

ARE YOU NEWLY **DIAGNOSED?**

It's not uncommon to be overwhelmed with a new diagnosis. The Neuromuscular Disease Foundation has several free offerings for GNEM patients. Simply scan the QR code below to access patient resources.

WHAT CAUSES **GNEM?**

GNE Myopathy is believed to be 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 weaken over time.

WHAT IS **GNEM MYOPATHY?**

GNEM is a distal myopathy; a rare genetic disease usually starting at the feet, causing muscles to slowly weaken. GNE Myopathy is not life-threatening, but it may lead to physical debilitation within two decades of symptoms' onset. Symptoms usually begin to develop in early adulthood, between late teens to early 30's. GNEM exists in all races and nationalities, world wide; however, ongoing natural history studies* show elevated carrier rates in certain populations of Eastern European and Asian heritage; including Jewish, Persian, Uzbeki, Arab, East Indian, Korean and Japanese among others.

**For more details see clinicaltrials.gov 10: NCT01784679 and NCT01417533.*

UNDERSTANDING THE GENETICS

GNE Myopathy 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 2 carriers have a child there is a:

25% chance that each child will be affected by GNE Myopathy

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

WHERE TO GET SCREENED AS A CARRIER FOR GNE MYOPATHY

For more information about genetic screening or to order a genetic test kit, contact one of our partners: JScreen: jscreen.org or The Open Medicine Institute: openmedicineinstitute.org

If you think you may have weak muscle symptoms you can get an online diagnosis at: Jain-foundation.org/alda

ANSWERING YOUR **QUESTIONS**

HOW COMMON IS GNE MYOPATHY?

This rare genetic disorder is found among all ethnicities. A study done by the National Institutes of Health (NIH) predicts that between 20,000-40,000 patients exist globally. NDF's own studies* have found concentrated populations in certain cities in the Middle East and Eastern Europe to have carrier rates as high as 1 in 4.
**Dr. Nuria Carrillo, NIH and Dr. Hossein Khademian in collaboration with Dr. Siavash Kurdistani, UCLA.*

WHAT ARE THE SYMPTOMS OF GNE MYOPATHY?

Early signs and symptoms include foot drop, difficulty running or walking, frequent loss of balance, tripping, and weakness in the pincer grip. As time progresses, weakness may involve the index fingers, 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 GNE MYOPATHY TREATED?

There is currently no approved treatment or cure for GNEM. With the support of NDF, the NIH is in the process of developing MaNAc, a medication that could potentially slow down the progression of the disease. Breakthrough technology is making human gene therapy trials possible, and NDF is committed to and presently funding such research. However, today's practical treatment options are geared toward managing symptoms and early detection. 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 clinicaltrials.gov and gnem-dmp.com.

HOW CAN GNE MYOPATHY BE PREVENTED?

Genetic screening and preimplantation genetic diagnosis can 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. 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.

For information about genetic screening at a medical facility or in the privacy of your own home, please visit CureGNEM.org/programs/advocacy.

CONTACT **US**

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