Welcome to the GNEM-DMP!

Welcome to the fourth GNE myopathy Disease Monitoring Programme (GNEM-DMP) newsletter and thank you for your continued support and participation in the GNEM-DMP. Our newsletter is intended to provide you with regular updates on the GNEM-DMP and provide you with scientific updates related to GNE myopathy. We welcome your feedback and suggestions on this newsletter.

In this Edition

- GNE myopathy Research and Clinical Trials with ManNAc at the US National Institutes of Health
- Patient Organisations and What They Do
- The Importance of Patient Advocacy for Rare Diseases
- Aceneuramic acid Phase 3 Clinical Trial Update (Ultragenyx) - New sites are now open
- GNEM Registry Update
- Mobility and Use of Wheelchairs/Scooters in Registry Participants
- Participant Story: ‘My Journey So Far’ – Mark

To join the GNE Patient Registry please visit: www.gnem-dmp.com
For more information on the GNEM-DMP contact: HIBM@treat-nmd.eu
For more information about Ultragenyx Pharmaceutical Inc. please visit: www.ultragenyx.com
For more information about TREAT-NMD please visit: www.treat-nmd.eu
Researchers at the US National Institutes of Health (NIH) have been conducting extensive research since 2001 to learn more about the underlying cellular defects in GNE Myopathy. One of their studies (in 2007) in mutant mouse models demonstrated that oral ManNAc therapy improved pathologic features of both GNE Myopathy and glomerular diseases.

ManNAc, or N-acetyl-D-mannosamine, is a naturally occurring sugar and an intermediary in the production of sialic acid, a component of muscle and other tissues. Decreased muscle sialic acid levels are believed to contribute to the clinical aspects of GNE Myopathy. The National Center for Advancing Translational Sciences (NCATS) and National Human Genome Research Institute (NHGRI), completed two pivotal animal toxicology studies to demonstrate ManNAc drug safety, and allowed successful filing of the Investigational New Drug (IND) application to U.S. Food and Drug Administration (FDA). In addition, the FDA has provided orphan designation for ManNAc in GNE Myopathy.

A Phase 1 clinical study was completed at the NIH Clinical Center (Bethesda, Maryland, USA), assessing pharmacokinetic properties and safety of ManNAc in GNE Myopathy patients (ClinicalTrials.gov Identifier: NCT01634750). These clinical studies demonstrated that ManNAc is well-tolerated by patients and that twice-daily oral ManNAc dosing led to significant and sustained increase in the circulating levels of sialic acid. In collaboration with NCATS and NHGRI researchers, ManNAc is currently being investigated in an open-label Phase 2 study in GNE Myopathy (ClinicalTrials.gov Identifier: NCT02346461). A multi-center clinical trial is being planned. The NIH is collaborating with Escala Therapeutics, Inc., a New York based company focused on development of drugs in rare disorders.

At the NIH Clinical Center (Bethesda, USA), ~45 patients with GNE Myopathy have been followed on a prospective, single-center Natural History Study (ClinicalTrials.gov Identifier: NCT01417533). This study started in September 2011 and collects health and biological information over time in order to understand how GNE Myopathy disease affects patients’ strength, function and quality of life. The information collected gives researchers insights into which measures can be used to demonstrate response to therapy in clinical drug trials for this slowly progressive disease. As part of their participation, patients also receive information about the status of their disease and how to better handle medical aspects related to GNE Myopathy.

For more information on the NIH research and clinical studies: Nuria Carrillo (carrilln@mail.nih.gov) or Marjan Huizing (mhuizing@mail.nih.gov)
If you have any questions about patient advocacy or any of the information in this article please contact patientadvocacy@ultragenyx.com

Julie Kelly is a freelance consultant working in the rare disease arena. She is a Patient Services Consultant, supporting Patient Advocacy for Ultragenyx, and in this article shares her experiences and advice in relation to the importance of patient advocacy for rare diseases, such as GNE Myopathy.

“I have been working in the area of rare disorders for many years, and have been very fortunate and privileged to connect with many patient advocacy groups during this time. Their advice, help and guidance has always been of great benefit to me as a person, to help me have a better understanding of the needs of patients, their carers, and their families.

The aim and focus for patient advocacy groups is in principle the same as that of the medical teams who provide care for you - they are focused on you and your needs. They provide invaluable help, support, and information when and where needed, and look at many other aspects of living with a rare disease, such as social care and support, how to access resources which can help you in daily life, and much more. They have worked tirelessly with awareness raising of rare disorders, and ensure that the word “rare” does not mean “invisible”. Working to try and ensure an early diagnosis and putting patients and families in contact with those who can help is also key to their work. At all times they have worked closely with the medical teams, and with compassion and care.

Patient Advocacy Groups can also influence planning and decision making within the healthcare environment, which can in turn have an impact on the lives of those they support. Government, health care providers, and those who make decisions on resources planning can all benefit from connecting to Patient Advocacy groups, as their insight and experience can be of great help to better understand the needs of patients and families.

Patients and families themselves who are diagnosed with a rare disorder can at times feel isolated and alone with their condition. I believe there is great value for those who choose to connect with others, and who are experiencing the same challenges as you, can be of real help, and help you feel that you are not alone. Of course there are patients and families who might prefer to remain private and not connect to others, but still need and appreciate the help of a professional body who understands their needs and what they have to face on a daily basis.

Patient advocacy groups work closely with the medical teams so that there is a shared understanding of your needs. This extends the care and guidance you receive from your doctors, nurses and medical teams, and can also look at other aspects of daily life and where you may need help and guidance. Sometimes it can be the little things in life which make all the difference.

Knowing that there is someone you can call on to ask for help and advice, to connect you to others who are experiencing some of the challenges you have faced, and to ensure you do not feel you are alone is of such value and importance to us all.”

If you have any questions about patient advocacy or any of the information in this article please contact patientadvocacy@ultragenyx.com

---

Muscular Dystrophy Ireland (MDI)
Muscular Dystrophy Ireland (MDI) is a voluntary organisation, which was established in 1972 by a small group of people in the west of Ireland to support families who had a member with muscular dystrophy. Since then it has grown considerably and it now has a membership of over 700 members and a network of branches throughout Ireland.  
www.mdi.ie/

Associazione Gli Equilibristi HIBM – (Italy)
This is a non-profit organisation with the primary aim of spreading the knowledge of GNE Myopathy. Established in 2012, its mission is to raise funds in order to promote and support the medical and scientific research aimed at the study of this disease, form a network of patients with GNE Myopathy in order to ensure there are direct updates on the disease and be a means of communication  
www.gliequilibristi-hibm.org/

Distal Muscular Dystrophy Patients Association (PADM) – (Japan)
PADM is a patient organization based in Japan which is very active in pursuing a treatment for GNE Myopathy. There are over 130 members in this association which was established in April 2008.  
www.enigata.com/index_e.html

GNE Myopathy International
An international group of GNE myopathy patients, family and friends, with the mission to generate awareness among patients and communities worldwide about this rare genetic disorder, and to provide information and support to patients. Located in many countries including Asia, Europe, Middle East, and United States of America.  
www.gne-myopathy.org/

Muscular Dystrophy UK (MD UK) – (UK)
MD UK is a charity for the 70,000 people living with muscle-wasting conditions in the UK. Since 1959, it has been supporting families living with muscle-wasting conditions. They provide vital information, advice, resources and support for people with these conditions, their families and the professionals who work with them.  
www.musculardystrophyuk.org/

Neuromuscular Disease Foundation (NDF) – (USA)
NDF is a non-profit organization launched in 2006 whose mission is to raise awareness and encourage testing for GNE Myopathy and to direct funding to scientists who are working to find a treatment and cure for neuromuscular diseases, including GNE Myopathy. Our vision is a world free of GNE Myopathy today and for generations to come.  
www.ndf-hibm.org/

Advancement for Research for Myopathies (ARM)
ARM is a non-profit organization with the primary goal of speeding up bio-medical research on IBM2, the Autosomal Recessive form of Hereditary Inclusion Body Myopathies (HIBM). Founded by HIBM patients in 2000, ARM’s mission is to support and inform patients and their families, to raise funds for research, to encourage researchers to study this rare disorder and to ultimately find a cure for HIBM.  
www.hibm.org/arm/home
Phase 3 Randomized, Double-Blind, Placebo-Controlled Study to Evaluate Sialic Acid Extended Release Tablets in Patients with GNE Myopathy (GNEM) or Hereditary Inclusion Body Myopathy (HIBM) - New sites are now open

Ultradynex Pharmaceutical is seeking participants for a phase 3 study of investigational aceneuramic acid extended release (Ace-ER) tablets (also known as sialic acid extended release, or SA-ER) for the treatment of GNE myopathy, also known as hereditary inclusion-body Myopathy (HIBM) and Nonaka distal myopathy.

The study will collect information on the safety and efficacy of Ace-ER in people with GNE Myopathy. As the study is placebo-controlled, half of enrolled patients will receive the study drug and half will receive a placebo (sugar pill). After the study has been completed, all participants may have the opportunity to take part in an extension study in which they will receive treatment with Ace-ER.

The study will take place in selected centers throughout the world. The following centers are currently open for recruitment:

<table>
<thead>
<tr>
<th>Country</th>
<th>Location</th>
<th>Status</th>
<th>Contact Information</th>
<th>Principal Investigator(s)</th>
</tr>
</thead>
<tbody>
<tr>
<td>United States, California</td>
<td>Irvine, California, United States</td>
<td>Recruiting</td>
<td>Tel: 714-456-8520 <a href="mailto:bminton@uci.edu">bminton@uci.edu</a></td>
<td>Tahseen Mozafar, MD</td>
</tr>
<tr>
<td>United States, Missouri</td>
<td>Saint Louis, Missouri, United States</td>
<td>Recruiting</td>
<td>Tel: 314-362-1626 <a href="mailto:rennar@neuro.wustl.edu">rennar@neuro.wustl.edu</a></td>
<td>Alan Pestronk</td>
</tr>
<tr>
<td>United States, New York</td>
<td>New York, New York, United States, 10016</td>
<td>Recruiting</td>
<td>Tel: 212-263-6628 <a href="mailto:swapnil.parma@nymc.edu">swapnil.parma@nymc.edu</a></td>
<td>Heather Lau, MD</td>
</tr>
<tr>
<td>United States, New York</td>
<td>New York, New York, United States, 10029</td>
<td>Recruiting</td>
<td>Tel: 212-659-1477 <a href="mailto:luca.fierro@msmsm.edu">luca.fierro@msmsm.edu</a></td>
<td>George Diaz, MD</td>
</tr>
<tr>
<td>Bulgaria</td>
<td>Sofia, Bulgaria</td>
<td>Not yet Recruiting</td>
<td><a href="mailto:teodoratch@abv.bg">teodoratch@abv.bg</a></td>
<td>Ivailo Tournev, MD</td>
</tr>
<tr>
<td>Canada, Ontario</td>
<td>Hamilton, Ontario, Canada, L8N325</td>
<td>Recruiting</td>
<td>Tel: 905-521-2100 x 76929 <a href="mailto:hatchere@hhsc.ca">hatchere@hhsc.ca</a></td>
<td>Mark Tarnopolsky, MD</td>
</tr>
<tr>
<td>France</td>
<td>CHU La Réunion - site GHSR</td>
<td>Recruiting</td>
<td>Tel: 0262.35.96.73 <a href="mailto:julie.ruiz@chu-reunion.fr">julie.ruiz@chu-reunion.fr</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, New York</td>
<td>New York University School of Medicine</td>
<td>Recruiting</td>
<td>Tel: 212-521-2100 x 76929 <a href="mailto:hatchere@hhsc.ca">hatchere@hhsc.ca</a></td>
<td>Mark Tarnopolsky, MD</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, California</td>
<td>Irvine, California, United States, 92697</td>
<td>Recruiting</td>
<td>Tel: 714-456-8520 <a href="mailto:bminton@uci.edu">bminton@uci.edu</a></td>
<td>Tahseen Mozafar, MD</td>
</tr>
<tr>
<td>France</td>
<td>CHU La Réunion - site GHSR</td>
<td>Recruiting</td>
<td>Tel: 0262.35.96.73 <a href="mailto:julie.ruiz@chu-reunion.fr">julie.ruiz@chu-reunion.fr</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, Missouri</td>
<td>Washington University School of Medicine</td>
<td>Recruiting</td>
<td>Tel: 314-362-1626 <a href="mailto:rennar@neuro.wustl.edu">rennar@neuro.wustl.edu</a></td>
<td>Alan Pestronk</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, New York</td>
<td>New York University School of Medicine</td>
<td>Recruiting</td>
<td>Tel: 212-263-6628 <a href="mailto:swapnil.parma@nymc.edu">swapnil.parma@nymc.edu</a></td>
<td>Heather Lau, MD</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, New York</td>
<td>New York University School of Medicine</td>
<td>Recruiting</td>
<td>Tel: 212-521-2100 x 76929 <a href="mailto:hatchere@hhsc.ca">hatchere@hhsc.ca</a></td>
<td>Mark Tarnopolsky, MD</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, California</td>
<td>Irvine, California, United States, 92697</td>
<td>Recruiting</td>
<td>Tel: 714-456-8520 <a href="mailto:bminton@uci.edu">bminton@uci.edu</a></td>
<td>Tahseen Mozafar, MD</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, Missouri</td>
<td>Saint Louis, Missouri, United States, 63110</td>
<td>Recruiting</td>
<td>Tel: 314-362-1626 <a href="mailto:rennar@neuro.wustl.edu">rennar@neuro.wustl.edu</a></td>
<td>Alan Pestronk</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, New York</td>
<td>New York University School of Medicine</td>
<td>Recruiting</td>
<td>Tel: 212-263-6628 <a href="mailto:swapnil.parma@nymc.edu">swapnil.parma@nymc.edu</a></td>
<td>Heather Lau, MD</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, California</td>
<td>Irvine, California, United States, 92697</td>
<td>Recruiting</td>
<td>Tel: 714-456-8520 <a href="mailto:bminton@uci.edu">bminton@uci.edu</a></td>
<td>Tahseen Mozafar, MD</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, Missouri</td>
<td>Saint Louis, Missouri, United States, 63110</td>
<td>Recruiting</td>
<td>Tel: 314-362-1626 <a href="mailto:rennar@neuro.wustl.edu">rennar@neuro.wustl.edu</a></td>
<td>Alan Pestronk</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, New York</td>
<td>New York University School of Medicine</td>
<td>Recruiting</td>
<td>Tel: 212-263-6628 <a href="mailto:swapnil.parma@nymc.edu">swapnil.parma@nymc.edu</a></td>
<td>Heather Lau, MD</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, California</td>
<td>Irvine, California, United States, 92697</td>
<td>Recruiting</td>
<td>Tel: 714-456-8520 <a href="mailto:bminton@uci.edu">bminton@uci.edu</a></td>
<td>Tahseen Mozafar, MD</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, Missouri</td>
<td>Saint Louis, Missouri, United States, 63110</td>
<td>Recruiting</td>
<td>Tel: 314-362-1626 <a href="mailto:rennar@neuro.wustl.edu">rennar@neuro.wustl.edu</a></td>
<td>Alan Pestronk</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
<tr>
<td>United States, New York</td>
<td>New York University School of Medicine</td>
<td>Recruiting</td>
<td>Tel: 212-263-6628 <a href="mailto:swapnil.parma@nymc.edu">swapnil.parma@nymc.edu</a></td>
<td>Heather Lau, MD</td>
</tr>
<tr>
<td>France</td>
<td>Institut de Myologie GH Pitié-Salpêtrière</td>
<td>Recruiting</td>
<td><a href="mailto:n.reguiba@institut-myologie.org">n.reguiba@institut-myologie.org</a></td>
<td>Anthony Behin, PH</td>
</tr>
</tbody>
</table>

To learn more about this trial, including inclusion and exclusion criteria and how to participate, please visit: [www.clinicaltrials.gov/ct2/show/NCT02377921](http://www.clinicaltrials.gov/ct2/show/NCT02377921).
As of March 2016 there are now 225 participants (Fig 1) in the GNEM-DMP Registry, from 25 different countries. The map to the left shows the distribution of participants globally, highlighting the amount of participants from each country who have so far signed up. The average age of the registry participants is 39.5 years old (age range of participants extends from 20.5 to 70.1 years old).

The data that you provide in the registry gives us a great insight into the global GNE myopathy community and we appreciate you taking the time to complete your questionnaires at your first visit, 6 months later and then at yearly intervals going forward.

Update on the Registry Component of the GNEM-DMP

To answer this question we looked at your answers to the registry questionnaires. According to this analysis first signs of GNE Myopathy occur around 28-29 years of age, but occasionally as early as 15 years of age or as late as 50 years of age. Weakness in arms and hands would usually be felt five years later than weakness in the legs. Difficulty sitting up right without support becomes noticeable seven years after the first symptoms of GNE Myopathy. Wheelchairs are used on average from 39 years of age, but this is also spread out widely between people in their early twenties to others in their fifties. More than 30% of registry participants stopped working because of the muscle weakness at an average age of 33-34 years.

Our observations are based on a preliminary data analysis and may change once more information becomes available. We think the estimated time line of events is useful as it helps disease management and to be prepared. Of course it is important to remember that not all of these symptoms will occur in every person diagnosed with GNE Myopathy – some will live for many years and into post middle-age, unaffected by all of these symptoms.

GNE Myopathy is a slowly progresssing disease. FAQ: How quickly will I get worse / how slow or fast could it be?

Fig 1: Worldwide Distribution of GNEM-DMP participants

Fig 2: Breakdown of Participant Mobility – From Onset until Requiring Use of Wheelchair/Scooter (Average and Range)
Mark is from the United States of America and in 2003 was diagnosed with the muscle wasting condition Hereditary Inclusion Body Myopathy (HIBM), also known as GNE Myopathy. Mark is currently participating in the GNEM-DMP International Registry at Newcastle University, which aims to increase understanding of the condition and drive clinical trials and research forward and could lead to effective treatments. Below, Mark talks about living with the condition and shares some stories about himself.

My story begins as most do - by being born, which in my case was in 1965. My parents allowed me to try different things growing up, such as music lessons (hated them) and playing football (I liked that). I was, and still am the first person in my entire family to obtain a bachelor’s degree. After graduating I had a few jobs before settling in to an organisation where I worked as a fraud investigator, staying there for nearly twenty years. I married a beautiful woman named Phyllis and have two great daughters (college students).

At around the age of 32 I injured my knee, which resulted in me requiring therapy. During this initial evaluation it showed that my calves and hamstrings were also unusually weak, so I started to work these muscle groups into the therapy sessions as well. Over time my knee got better but I could not improve the other muscle groups. The therapist I was seeing suggested that I should visit a Neurologist. The first Neurologist that I visited said that they believed I had ALS (Amyotrophic lateral sclerosis). Needless to say panic ensued; however testing proved I did not have ALS. Each subsequent visit to the Neurologist resulted in me undertaking further tests, all of which came back negative. After eight months of this the first Neurologist gave up. A couple of months later I posted online (in the early days of the internet) about my symptoms and was referred to a doctor at the University of Medicine and Dentistry of New Jersey, who after a few tests (including another muscle biopsy) confirmed that I had HIBM. At this point the diagnosis was just a mild annoyance to me.

Flash forward a few years and I continued to follow-up with the doctor who was monitoring my increasing weakness. I tried using IVIG which seemed to help, but this had heavy side effects. At this point steps or stairs were challenging and I was beginning to trip and fall. Moving from sitting position to standing was also more difficult, but still possible. Ultimately I installed a stair lift in my house as the 15 stairs were just too much for me. As my falls became more frequent I started to use a cane to assist me. At my place of work, adjustments to my duties were made so I continued in my employment until 2006. Since leaving employment, I have kept myself busy by doing odd jobs as well as raising my kids.

In 2005 I connected with Advancement of Research Myopathies (ARM) in California (HIBM.ORG) and gave them samples to review for their research (I’m a member on the Facebook group - HIBM / GNE Myopathy Support Group). As time progressed I ended up taking one too many falls and on one occasion I fell fracturing two vertebrae. I was sent to rehab and have been in a power wheelchair ever since. Presently, I require assistance to do most things in my day to day life.

There have been a number of problems I have encountered which have impacted me financially since my diagnosis. I had to pay for the addition of a ramp so I can access my house from outside (which I discovered has its own problems during heavy snow). Also, as I live in a remote area of the USA called West Milford where transport for people in wheelchairs is not easily obtained, I have had to buy a ramp van in order to help me get around. The extra expense caused problems with my mortgage company and so we ended up having to leave our house. We moved into an apartment which is much more wheelchair friendly, and we are now attempting to purchase a wheelchair friendly place of our own.

The experiences I encountered during this difficult time, led me to finding out about a programme called ‘Workability’ that is ran here in the USA. I was able to access financial assistance via Medicaid (a social health care programme) - the only catch being, I was required to be in employment in order to receive the assistance. After dozens of dead end interviews I connected with an online company that teaches English as a second language. However, in order to qualify I was required to return to school to get a teaching certificate, which I successfully obtained. Since this I have been working as a teacher. With Medicaid I now have access to the essential services that I require in my everyday life to enable me to get by. This program has been a life saver. I highly recommend that you look into the social health care programmes available to you in your country of residence.

This disease is hard on me and my family. I must adapt to doing things very differently than “normal” people. Using grabbers to pick up things, working from the power chair for every aspect of daily life, relying on others for assistance, not being able to go places with friends and family because of steps/stairs, being unable to open a jar or lift more than a few pounds and having to be driven by others are examples of this. I have also had some trouble with depression since my diagnosis, but have worked through it. The hardest part I think is knowing there really is nothing that can stop the progress of the disease.

But now the bright side. A long time ago, back when I knew there was no treatment I began to study alternate and herbal remedies. Through much trial and error, I have come up with a regime of things I believe are helpful for my wellbeing, including vitamins minerals and protein supplements. Being in contact with others does also help, allowing me to realise I’m not alone. The most important thing that has gotten me through this ordeal is Phyllis standing by me. Knowing she loves me makes everything else seem tolerable. No matter what is thrown at me I have gotten through it, and so can you.

If you would like to contribute to a future edition of the GNEM-DMP newsletter with a story of your own, please contact us at HIBM@treat-nmd.eu

Please be aware that this article contains the views and opinions of Mark and does not necessarily imply that TREAT-NMD or the International GNE Myopathy Registry endorse them.