

WINTER 2021

THE CMTA Report

www.cmtausa.org



MEET CRYSTAL EMERY— WRITER/PRODUCER/FILMMAKER

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Inhibitors Funded

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Like Son

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OUR MISSION: To support the development of new drugs to treat CMT, to improve the quality of life for people with CMT and, ultimately, to find a cure.
OUR VISION: A World Without CMT.

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THE CMTA Report

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Email the CMTA at info@cmtausa.org



DEAR FRIENDS,

I'm sure I'm not the only one who was happy to see 2020 end. It's more than a little ironic that a number depicting clarity of vision came to represent a year filled with confusion bordering on chaos. But with your help, the CMTA not only persevered, it thrived.

The CMTA community thrived because our members share our passion for our mission—finding a cure for CMT. The pandemic did nothing to diminish that passion—though it did force us to pivot to new ways of holding branch meetings, walking 4 CMT and shifting to research that could be done in a bubble. But, as David Tannenbaum noted in our special issue on COVID-19, people with CMT have had to deal with strangeness and adversity all of their lives. According to David, “The uncertainty and fear of the COVID-19 crisis is not all that different from the uncertainty and fear we face in living with CMT.”

This issue is dedicated to everyone—men, women and children, teachers, doctors and writers—who is helping the CMTA by spreading the word about CMT. It's a constant, ongoing battle to educate the many, many people who still have not heard of the disease. But it's a vital step toward finding a cure—identify the problem, make people aware of the problem and ask people to help fund a cure. Dave Loy got a massive shark tattoo on his arm so that he can talk to people about CMT when they ask about it. Rick Clemente makes and gives away gorgeous wooden pens. When people ask about the pens, he says the price is a donation to the CMTA. Sarah Kesty entered her lesson plan about CMT into a contest, and Jeff Seitzer is publishing his memoir about CMT. Filmmaker Crystal Emery tells us how she prevails over her CMT despite its impact on her arms, legs and respiratory system.

We'll be bringing you more in-depth information on STAR's progress in an upcoming special issue, but for now I'd like to highlight a few examples of the tremendous research progress made in 2020, including:

- \$2.5 million invested in research
- 19 joint preclinical treatment studies this year with leading pharma/biotechs developing treatments for CMT
- 32 total alliance partners from top biotech, pharma and gene therapy labs around the world
- 50 active research projects and studies and eight more projects approved in recent months
- More than 30 of the leading CMT scientists and gene therapy experts from around the globe working with our STAR Advisory Board
- Research tools for biotech companies to use in testing potential therapies for types CMT1, CMT2, CMT4 and CMTX
- Investments in the discovery of new genes that cause CMT

We look forward, as always, to a brighter future for everyone with CMT. And we wish each of you a very Happy New Year.

With warm regards,

A handwritten signature in black ink that reads "Amy Gray". The signature is fluid and cursive.

AMY GRAY, Chief Executive Officer

COVID-19 Vaccine and CMT—Q & A



What are the side effects of the COVID vaccine? Will they affect my CMT?

The known side effects of current and likely-to-be authorized COVID-19 vaccines are similar to those of the annual flu shot and include muscle soreness at the injection site, fever, tiredness, body aches and headache. It's important to note that at this time we do not know whether or not the COVID-19 vaccine will have a different or more serious set of side effects for CMT patients. You should talk to your doctor about that possibility.

What impact will the vaccine have on possible future gene therapy treatments or medications?

The Food and Drug Administration's authorization of the

Pfizer/BioNTech vaccine did not mention any contraindications for genetic therapies or other medications particularly important to the CMT community, but you should consult your clinician about whether the vaccine will have an adverse impact on any future course of treatment. Should there be any contraindications in the future, we will update our information accordingly.

What safety measures are in place for the COVID vaccine?

The U.S. vaccine safety system ensures that all vaccines are as safe as possible. The Centers for Disease Control has developed a new tool, v-safe, as an additional layer of safety monitoring to increase our ability to rapidly detect any safety issues with COVID-19 vaccines. V-safe is a

new smartphone-based, after-vaccination health checker for people who receive COVID-19 vaccines.

How long will it take to work? Will I get proof of vaccination?

You won't get the full protection from the Pfizer-BioNTech vaccine until about a week after the second dose, based on clinical trial data. The researchers found that the vaccine's protection started to emerge about 10 days after the first dose, but it only reached 52 percent efficacy, according to a report in the *New England Journal of Medicine*. A week after the second dose, the efficacy rose to 95 percent. You should receive a vaccination card or printout that tells you what COVID-19 vaccine you received, the date you received it and

where you received it. You should receive a paper or electronic version of a fact sheet that tells you more about the specific COVID-19 vaccine you are being offered. Each authorized COVID-19 vaccine has its own fact sheet that contains information to help you understand the risks and benefits of receiving that specific vaccine.

What if I forget to get the second dose on time?

Both the Pfizer-BioNTech and Moderna vaccines have two doses, with the booster shot coming a few weeks after the first. Pfizer-BioNTech's second dose comes three weeks after the first, and Moderna's comes four weeks later. The second dose provides a potent boost that gives people strong, long-lasting immunity. If for some reason you fail to get the second shot precisely three weeks after the first, you don't have to start all over again with another two-dose regimen.

What will happen if serious side effects crop up after the vaccine is rolled out?

Once a vaccine starts to reach large numbers of people, it's possible (and not uncommon) for a small number of severe "adverse events" to occur. Many existing vaccines, including the flu shot, also can cause rare complications, including Guillain-Barré syndrome, seizures and sudden unexplained death. While this sounds frightening, the risk is minuscule when considered over the millions of people who are safely vaccinated each year, and some of these complications can be triggered

by the virus itself. Health officials will investigate each event to see if it's simply coincidence—or if it could have been caused by the vaccine. While everyone should be prepared to hear about these reports, they should not be a cause for worry or prompt you to delay getting the vaccine. Your risk of severe complications from COVID-19 is far higher than your risk of complications from the vaccine.

I've had COVID-19 already. Do I need the vaccine?

It's safe, and probably even beneficial, for anyone who has had COVID to get the vaccine at some point, experts said. Although people who have contracted the virus do have immunity, it is too soon to know how long it lasts. So for now, it makes sense for them to get the shot. The question is when. Some members of the CDC advisory committee have suggested people who have had COVID in the past 90 days should be toward the back of the line.

Will the vaccine work on older people?

All the evidence so far suggests that the answer is yes. The clinical trials for the two leading vaccines have shown that they work about the same in older people as younger people. As the vaccines get distributed, the vaccine makers and the CDC will continue to monitor the effectiveness of the vaccine in people 65 and older who, because of age-related changes in their immune systems, often don't respond as well to vaccination as younger people do. But just as certain flu vaccines have been developed to evoke a stronger

FOR MORE INFORMATION about the COVID-19 vaccine and CMT, go to www.cmtausa.org/covid.

immune response in older people, it's possible that one of the new vaccines could emerge as a better option for this age group. It's just far too soon to know.

Will these vaccines put a dent in the epidemic?

The coronavirus vaccines will be much less effective at preventing death and illness in 2021 if they are introduced into a population where the virus is raging—as is now the case in the U.S. A vaccine that's 95 percent effective, as Moderna's and Pfizer's versions appear to be, is a powerful fire hose. But the size of a fire is still a bigger determinant of how much destruction occurs. According to the authors of a paper in the journal *Health Affairs*, at the current level of infection in the U.S. (about 200,000 confirmed new infections per day), a vaccine that is 95 percent effective—distributed at the expected pace—would still not be enough to end the terrible toll of the virus in the six months after it was introduced. Almost 10 million or so Americans would contract the virus, and more than 160,000 would die. Measures that reduce the virus's spread—like mask-wearing, social distancing and rapid-result testing—can still have profound effects. Public health officials hope that people will continue to take these precautions at least until the country reaches a vaccination rate of 70 to 75 percent. ☆

DISCLAIMER: Nothing shared on these pages should be construed or is intended to be used for medical diagnosis or treatment. It should not be used in place of the advice of your physician or other qualified health care provider. Should you have any emergency questions or concerns, please contact your physician or health care provider immediately. Always consult with your physician or other health care provider to gain clarification regarding any health care related questions. This content was sourced from the CDC, the CMTA Scientific Advisory Board, the Muscular Dystrophy Association and the New York Times.

Filmmaker Crystal Emery: Energy in Motion and Spirit

BY MARCIA SEMMES



CMT took the use of her arms and legs from writer/producer/filmmaker Crystal Emery, but it can't touch her indomitable spirit. "I refuse to be defined by the body I inhabit," she says, adding, "As a deeply passionate and creative individual, I am so much more than a Black woman living with a life-altering physical disability. I am energy in motion and spirit first."

Crystal's latest documentary, "The Deadliest Disease," is about the scourge of racism in health care, with COVID-19 as Exhibit A. She held a virtual screening narrated by Soledad O'Brien in May 2020 and the film will be broadcast on American Public Television in 2021.

Crystal grew up in New Haven, Connecticut, with a passion for acting. She wrote and directed her first play in the fifth grade. In the sixth grade, she began falling without tripping. While she intuited that something was seriously wrong, everyone else wrote it off as clumsiness. After one particu-

larly bad fall, her left hip slipped out of place, requiring major surgery and landing her in a body cast for two months, an experience she calls "nothing short of traumatizing."

She walked more slowly after being liberated from the cast but continued to take dance classes and play softball. Her orthopedist said she needed more physical therapy, and her physical therapist said she was "lazy." Faced with the doctors' criticisms and lack of tangible guidance, she eventually stopped going to therapy altogether. Crystal began to experience a slight drop foot in tenth grade and by the end of high school, she couldn't walk the 12 blocks from high school to her job downtown.

In 1981, after years of slow decline, Crystal was diagnosed with a form of CMT that affects both her limbs and her respiratory system. Once again, it didn't stop her—after earning a BA from the University of Connecticut, she began her professional career in theater and film with an apprenticeship with

"Obstacles are those frightful things you see when you take your eyes off your goals."

renowned theater director Lloyd Richards ("The Piano Lesson"). She polished her craft under director Bill Duke ("A Rage in Harlem"), then went on to earn a master's in media studies from The New School for Public Engagement.

Since then, Crystal has produced socially conscious storytelling that celebrates the triumph of the human spirit on a variety of platforms. In addition to "The Deadliest Disease," her filmography includes "Black Women in Medicine," both of which she wrote, directed and produced. The latter has been seen by 12 million people globally. Her writing has appeared in numerous publications, including *Time* magazine and *The Huffington Post*. Her published works include the first two volumes of the Little Man children's

book series; *Against All Odds: Black Women in Medicine*; and *Master Builders of the Modern World: Reimagining the Face of STEM* (science, technology, engineering and math).

Crystal is also the founder and CEO of URU The Right to Be, Inc., a non-profit content production company that fosters communication and understanding among diverse people by discovering and applying tangible multimedia solutions at the intersection of the arts, humanities, science and technology. Crystal's goal is to move all stakeholders toward a more equitable and humane world. She also conceived, designed and launched the innovative Changing the Face of STEM national educational initiative in 2015. The culmination of this initiative was a 2017 congressional forum in Washington D.C., in conjunction with the National Academy of Science, attended by more than 500 STEM professionals, policymakers, educators, students and members of the media.

Today, Crystal uses a wheelchair and a ventilator, relying on friends and health aides to help her eat, bathe and get in and out of bed. It takes her 2.5 hours to get from bed to wheelchair in the morning. That's a challenge, she says, but the bigger challenges come in the way CMT affects her ability to hang out with friends, go to her family's homes and work as an artist.

Nonetheless, she says, "CMT doesn't dominate me. It's just like any other obstacle. You just have to figure out how to get around it." Quoting Henry Ford, she added, "Obstacles are those frightful things you see when you take your eyes off your goals." ☆

CMTA Board Approves Grants to Jackson Lab to Study Two Inhibitors

The CMTA Board of Directors approved two research grants to Robert Burgess, PhD, at the Jackson Laboratory in 2020.

The first grant of \$45,000 will be used to explore HDAC6 (histone deacetylase 6) inhibitors, a potential therapeutic approach for multiple forms of CMT. The many genes associated with CMT make it unlikely that a single treatment will work for all forms of the disease, but preclinical studies with HDAC6 inhibitors, which have been shown to reduce motor and sensory deficits, have demonstrated promising results in several mouse models of CMT. Based on these promising results, scientists believe that HDAC6 inhibitors might be beneficial in treating a wide array of neurodegenerative conditions including demyelinating (Type 1 and 4) and axonal (Type 2) CMTs.

Dr. Burgess, a member of the CMTA's Scientific Advisory Board, will use mouse models of several forms of CMT to determine which types may be candidates for treatment with HDAC6 inhibitors and whether HDAC6 inhibitors may be of therapeutic benefit across a variety of CMT types. The latter will help inform whether patients with genetically undiagnosed cases of CMT are likely to benefit from this therapeutic strategy, or whether only select forms of CMT may respond to this treatment.



Dr. Robert Burgess, PhD

With the second grant of \$110,000, Dr. Burgess will study whether inhibiting SARM1 is of therapeutic benefit in multiple forms of CMT. SARM1 (sterile alpha and toll interleukin receptor motif containing protein 1) was identified in fruit flies as being actively involved in axon degeneration and similar effects were shown in SARM1 knockout mice.

The hypothesis is that inhibition of SARM1 may be beneficial in some, but not all, forms of CMT. The results obtained in these five CMT

Preclinical models of HDAC6... have demonstrated promising results in several mouse models of CMT.

models (1A, 1X, 2E, 2D, 2S) will be definitive and publishable and are not part of a larger effort on SARM1 in the Burgess lab. However, these results are important for the field and relevant to drug development efforts that are already underway. Furthermore, though this proposal is a pilot of modest scale, the results will inform future research on just how generally applicable SARM1 inhibitors may be in CMT. ☆

Two New Members Join Advisory Board

The CMTA welcomed two new members to its Advisory Board in 2020, part of its mission to provide enhanced expertise in a wide variety of fields to the community.



Christine Murray, MD

CHRISTINE MURRAY, MD, is a board-certified reproductive endocrinologist and infertility (REI) specialist. She specializes in in vitro fertilization and polycystic ovary syndrome with a special interest in preimplantation genetic testing of embryos, a technology that helps families deal with genetically inherited disorders and offers an approach to building families with an increased knowledge of, and options to address, genetically inherited syndromes.

She has spent 20 years in Vermont as an academic physician involved with the training of medical students, residents and fellows. After medical school at the Mayo Graduate School of Medicine, she underwent OB/GYN residency training at McGill University in Montreal, Quebec, followed by a fellowship in reproductive endocrinology and infertility. From 1999-2014, Dr. Murray worked at the University of Vermont Medical Center and was an associate professor in the Division of Reproductive Endocrinology. In addition to a busy clinical practice, Dr. Murray became residency program director in 2004 and held that position for eight years. In 2015, Dr. Murray opened Northeastern Reproductive Medicine to bring affordable, comfortable fertility care to patients wishing to build their families.

Dr. Murray became interested in the CMT community through her close friendship with the Ouellette family, who started the Vermont Cycle (and Walk) for CMT. She is interested in helping families understand their reproductive options and hopes that her extensive connections within the reproductive medicine community can provide answers and assistance.



Teresa Carroll, MS, PhD

TERESA CARROLL, MS, PHD, is an organismal biologist with more than 25 years of experience in higher education, primarily as an associate professor of biology in Missouri. In 2016, she moved to South Carolina, where she continued teaching at a small regional university and is currently writing topic-specific manuals for use in undergraduate biological labs.

Diagnosed with CMT1X in 1994, Teresa is an advocate and long-time supporter of the CMTA and has spent many hours since her diagnosis studying the scientific literature on CMT1X in an effort to understand her condition. Given that her career included taking complex biological information and making it comprehensible for undergraduate biology majors, Teresa is excited to have the opportunity to use those skills to build patient-friendly communications that help her fellow CMT patients and their families better understand the disease. ☆

Walks 4 CMT Raised Nearly \$180,000

CMTA volunteers hosted 26 virtual walks in 2020, raising nearly \$180,000 despite the disruptions and restrictions caused by the pandemic.

Because the 2020 Walks 4 CMT were virtual, participants were not limited by time or location. Teams held smaller walks with their families and walk leaders held virtual events using Zoom and social media. Many kicked things off with a fun activity like a Bingo or trivia game night, then walked for an entire month, some in smaller groups, all with proper social distancing.

The Syracuse, NY Branch kicked off its walk with a face mask fashion show on Zoom and went on to beat both last year's record and their goal for this year. According to Branch Leader Mike Casey, "After a couple of weeks, I decided that if we were going to make a Virtual Walk successful, we had to keep a positive outlook. So, I used my email and was on Facebook on a very regular basis asking for donations. I got more exposure



for our walk in the local newspaper, and I was featured on a local program on our ABC affiliate in Syracuse."

Mike went on, "We should have a good time and gather, even during these rough times with the pandemic. I feel that a positive outlook helps with meetings and other activities. We were very fortunate this year to have the Bucks County, PA and Albany, NY Branches join us. I can't wait until next year when we can have a walk in person."

The CMTA is one of the few national non-profit organizations with a walk campaign organized

and led solely by volunteers. Any city or town can host a Walk 4 CMT—all it takes is one leader to step forward. The CMTA makes it simple to host a walk with a Walk4CMT.org website, turnkey templates for sponsorship and marketing and support from our dedicated staff. ☆

If you want to be part of this amazing success, look for a 2021 walk at www.Walk4CMT.org. Or if you're ready to host a Walk 4 CMT in your community, contact CMTA Events Manager Julie Tarle at julie@cmtausa.org.

THE CMTA THANKS all the leaders and participants for their flexibility and creativity in raising research funds during this difficult time. The leaders are:

CALIFORNIA

Los Angeles: Alani Price and John Ramos
Palo Alto: Ori Bash and Tau O'Sullivan
San Diego: Kendall Trout

COLORADO

Denver: Carol Ris

DISTRICT OF COLUMBIA

Washington: Steve Weiss

FLORIDA

Miami: U of Miami CMT Clinic
Parkland: Lara Rustici
Tampa: Sarah Gentry

GEORGIA

Atlanta: Jeannie Zibrida

ILLINOIS

Chicago: Doreen Pomykala

MAINE

Peaks Island: Mary Louie

MARYLAND

Baltimore: Clark Semmes

MASSACHUSETTS

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Nashville: Teresa Shoaf

TEXAS

Dallas/Ft. Worth: Thomas Roderiguz

WASHINGTON

Seattle: Emily Osborne

ONTARIO, CANADA

Fergus: Kelly Hall

2020 WALK 4 CMT HIGHLIGHTS

26 Walk 4 CMT Events

459 Participants

11 Sites Raised Over \$5k

5 Sites Raised Over \$10k

DC Walk Raised a Record-Breaking \$42,000

Top Team "The Mighty Quinn" in Washington, DC Raised \$22,810

\$180,000 Raised for CMT Research



Parkland (FL) Walk 4 CMT

Advances in Genetics: New Tools for Prospective Families

BY CHRISTINE MURRAY, MD

Advances in genetic technology have allowed for the development of powerful tools to assist people as they begin to plan their families. Multiple techniques are available to provide prospective parents with valuable information to plan and prepare. This overview, while not comprehensive, offers a look into what modern genetic analysis can do.

A review of basic genetics provides a good starting point. Human beings have cells containing 46 chromosomes: Two will code for gender at birth (X and Y). The remaining 22 autosome pairs consist of maternal and paternal DNA. These paired chromosomes contain hundreds of thousands of DNA strands—genes that code for all the traits that make us who we are. Defects in any of these genes can lead to specific disorders. Such single-gene mutations can be inherited in an autosomal recessive manner, which means that an affected child received one defective gene from each parent.

The parents, each having only one affected gene, are considered silent carriers and are not affected by the disorder. Their offspring has a 25 percent chance of having the disorder and a 50 percent chance of being a silent carrier.

Examples of autosomal recessive disorders are cystic fibrosis, sickle cell anemia and spinal muscular atrophy. Some disorders are autosomal dominant—inheriting only one abnormal gene will lead to the

disorder. Huntington's disease and Marfan syndrome are examples of autosomal dominant inheritance. X and Y chromosomes can also carry genetic mutations: Hemophilia and fragile X syndrome have an X-linked inheritance pattern.

The Human Genome Project, a multinational scientific collaboration completed in 2003, is widely considered one of the most ambitious scientific undertakings of all time. The finished project sequenced 3 billion DNA letters covering 99 percent of the human gene-containing regions.

It is hard to overstate the tremendous impact of this undertaking. The results have been used to identify a genetic source for a multitude of medical issues, including cancers, as arising from specific mutations. Understanding the genetic basis of disease opens the door for specific diagnostic tools and treatments.

GENETIC CARRIER SCREENING

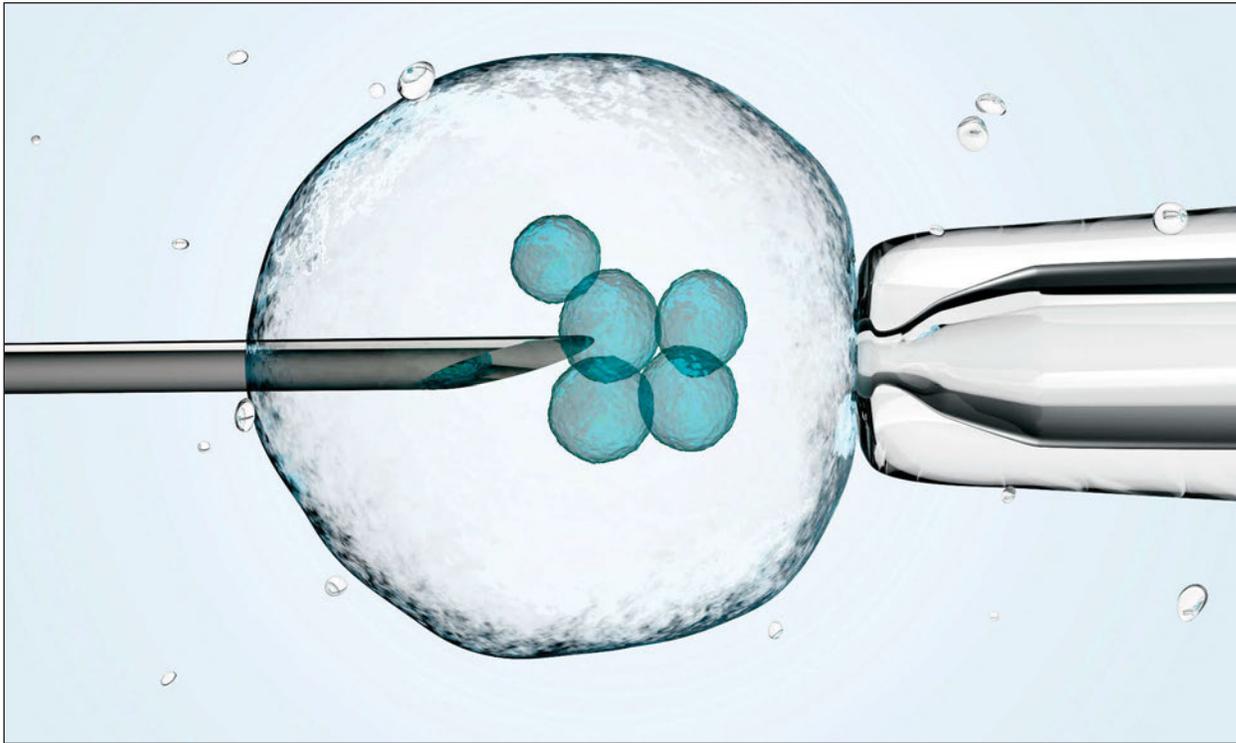
Carrier screening refers to a blood test that can identify silent gene mutations. Prospective parents are now routinely offered this screening to identify some of the most common mutations like the genes for cystic fibrosis (CF), spinal muscular atrophy and fragile X syndrome. This basic carrier panel is offered to one potential parent. If a silent mutation is identified, the other parent is tested. There are expanded panels recommended for parents based on their ethnic backgrounds. Tay Sachs, sickle cell anemia and the thalassemias

are more common among certain populations. Comprehensive carrier screening panels now test for nearly 300 single-gene defects. The American College of Obstetrics and Gynecology recommends that women have genetic carrier screening as part of their routine prenatal evaluation. Reproductive options for at-risk couples include IVF with preimplantation genetic testing (PGT), conception with donor eggs or sperm or adoption.

NON-INVASIVE PRENATAL TESTING/SCREENING

NiPT is a genetic test that is performed once pregnancy has been established. It requires a simple blood draw and can be done as early as nine to 10 weeks of pregnancy. The test analyzes tiny fragments of cell-free DNA from the developing placenta to assess the genetic health of the developing baby.

Unlike carrier screening, which provides information about specific genes, NiPT is used to detect chromosomal disorders that are caused by the presence of an extra or missing copy (aneuploidy) of a chromosome. NiPT primarily looks for trisomy 21 (Down syndrome), caused by an extra chromosome at position 21, trisomy 13 and 18 and extra or missing copies of the X and Y chromosome. NiPT can also identify gender. NiPT is a screening test, which means that it cannot give a definitive answer about whether the pregnancy is affected. The test can only estimate whether the risk of having certain conditions is



increased. An abnormal result is usually followed by more definitive testing (amniocentesis or chorionic villous sampling).

PREIMPLANTATION GENETIC TESTING

Since 1974, IVF has been used to help millions of couples who were unable to conceive for a variety of reasons: blocked Fallopian tubes, issues with ovulation or sperm production and endometriosis, to name a few. It is estimated that over 8 million babies have been conceived via IVF since Louise Brown—the first IVF success—was born in 1974.

The advent of preimplantation genetic testing (PGT) has added another group of prospective parents: those who have family or personal histories of genetically inherited conditions or who are at a higher risk of chromosomal anomalies due to advanced maternal age or recurrent unexplained pregnancy losses.

PGT refers to the process of creating embryos via IVF and

then performing a biopsy of the embryo to remove a small number of cells that can then be tested. Typically, an embryo is grown (incubated) in a laboratory for five days after fertilization. A 5-day-old embryo (blastocyst) has an inner cell mass—which is destined to become the baby—and an outer cell mass (trophoblast) that will ultimately develop into the placenta and amnion. Using an incredibly small and precise laser, a small opening is made in the wall of the embryo and cells are removed from the trophoblast. The embryo is then frozen while the cells are tested.

PGT-A tests the cells for an abnormal number or pairing of chromosomes (aneuploidy). Selecting embryos with a normal number and arrangement of chromosomes (euploid) to transfer back to the patient significantly increases the chances of a healthy pregnancy.

PGT-M refers to testing the embryos for single-gene mutations like the one that causes CF. A probe is created that can

detect the presence of the specific mutation gene: Two copies indicate an affected embryo; one copy would identify embryos destined to become genetic carriers of CF and those without probe activity would be considered unaffected.

To illustrate, a prospective mother is tested for cystic fibrosis and found to be a silent carrier of a CF mutation. Her partner is then tested and is also a carrier of a CF mutation. There is a 25 percent chance of having a baby with cystic fibrosis, which affects multiple organs—particularly the lungs. Incredible advances in treatments have been made and, armed with this knowledge, the couple may elect to proceed with conceiving on their own. They could also decide to conceive using an egg or sperm from a donor who does not carry the CF mutation. They might choose to adopt. They can also elect to have IVF and test their embryos for cystic fibrosis.

PGT-M is now widely utilized to identify embryos

(continued on page 13)



Youth Council Member Emily Stuffed (front row, center) held a fundraiser and awareness event for CMT at her high school in Florida in 2020.

Youth Council Gearing Up for Big Fundraising Year

Members of the CMTA’s Youth Council raised more than \$7,000 for the CMTA in 2020 and look forward to keeping the momentum going in 2021.

“This is the year we get the whole community of youth involved because together there is no stopping us,” the Youth Council’s Fundraising Committee—Elijah Tolz, Paola Martinez and Evan Zeltsar—said in a statement.

The three are spread out on opposite coasts but share a passion for fundraising to cure CMT and are “determined and ready to encourage other youth in the community to make a difference.”

Plans for 2021 include a virtual bingo game, publication of

“Walk a Mile in Our Braces,” a book charting the experiences of 75 youth with CMT and the first-ever Global Online Dance-a-thon 4 CMT.

The council has also developed a “Fundraising E-Kit,” a presentation filled with information on the different ways to fundraise and get involved. The E-kits are downloadable and tailored specifically for situations like school fundraisers, Walks 4 CMT and birthday celebrations. They are available on the CMTA website.

Two young fundraisers shared their motivation for helping the CMTA. According to

Maya Grochowska, “Attending Camp Footprint and meeting so many other kids with CMT inspired me to be more open about my CMT. And it made me want to raise money for the CMTA. Meeting other kids with CMT drove home that I’m not alone and all of us with CMT deserve hope for a cure.”

Hannah Roberts said she is “inspired to raise money for the CMTA because of its commitment to research and passion for community. I’m inspired because of my tribe and the impact for future generations living with Charcot-Marie-Tooth disease.” ☆

FOR MORE INFORMATION on getting involved with the CMTA’s Youth Program, go to www.cmtausa.org/youthprogram.

GENETIC TESTING

(continued from page 11)

affected by a great number of genetically inherited disorders that demonstrate a specific pattern of inheritance. Some disorders, however, are considered multifactorial. There may be a genetic component, but other factors are required to develop a specific disease. For example, some women may have a BRCA mutation. While this confers a significantly increased risk of breast and ovarian cancer, not everyone who carries the mutation will develop the disease. Some genetic disorders carry a highly variable inheritance pattern.

CMT is in this category. CMT is caused by mutations in genes that support or produce proteins involved in the structure and function of either the

peripheral nerve axon or the myelin sheath. More than 100 genes have been identified in CMT, with each gene linked to one or more types of the disease. In addition, multiple genes can be linked to one type of CMT. Moreover, the inheritance pattern can be autosomal dominant, X-linked or autosomal recessive. It can also appear as a de novo mutation.

While these variations present a challenge to probe development for PGT-M, it is still an option for many. Perhaps as many as 50-60 percent of cases involve the duplication of the *PMP22* gene on chromosome 17. Traditional PGT-M can be considered in such cases. Some cases can involve the deletion of this gene or some other variant; these cases typically require phasing-clinical testing of the patient's parents. Presently, de

novo mutations would not be identified by traditional PGT-M techniques.

Advances in genetic and reproductive technologies can expand reproductive options for many who, until recently, had very few. They can provide knowledge to aid in planning and preparedness for pregnancies affected by heritable medical disorders or chromosomal abnormalities and ultimately expand our approach to the diagnosis and treatment of complex diseases.

For more information, a helpful start can be found at ReproductiveFacts.org. ★

Christine Murray, the newest member of the CMTA Advisory Board, is a board-certified reproductive endocrinologist and infertility (REI) specialist. She specializes in in vitro fertilization and polycystic ovary syndrome with a special interest in preimplantation genetic testing of embryos.

WHETHER IN-PERSON OR VIRTUAL, LET'S HAVE SOME FUN IN 2021!

2021 CAMP FOOTPRINT REGISTRATION

2021 Camp Footprint: August 9-14

While we are not yet sure what camp will look like in 2021, we do know we will have a great time together, whether that's virtual or in-person!

We're asking all interested families to register for camp as usual and get all of your online forms completed. Monthly updates will be provided to all registered families.

Register at cmtacamp.org



cmtacamp.org



INNERVATORS

HONOR ROLL

ACCORDING TO THE DICTIONARY, an innervator is a nerve stimulator. According to the CMTA, an **INNERVATOR** is an action-oriented donor who gives monthly to the organization, sustaining STAR (Strategy to Accelerate Research), our premier research initiative, as well as important community initiatives like Camp Footprint, Patient/Family Conferences and educational programs. All of these programs require steady, reliable support from committed donors. Please join the Innervators today! www.cmtausa.org/cmta-innervators

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"I am an Innervator because I know the budgetary challenges nonprofits face in terms of planning. So often the bulk of giving occurs late in the fourth quarter. If enough people provide monthly giving over the course of the year, it will provide the CMTA with the stable funding it needs to fulfill its mission of ridding the world of this affliction." —David Coldiron

WHY I GIVE:

For anyone thinking of donating to the CMTA or its STAR research program, Howard Landis has four simple words of advice: "Now is the time."

Howard has a late-onset form of CMT 2 for which the culprit gene has not yet been identified, but the fact that the exact cause hasn't yet been pinned down doesn't stop him from getting involved and giving. As he noted, "The research community is doing its part and is making tremendous advances. Now is the time to join the CMTA in supporting them with your contributions, and now is the time to have your genome sequenced if you have an unidentified form of CMT. Your funds and your genomes will undoubtedly make a difference."

Howard is particularly interested in the work that Dr. Stephan Züchner is doing on unidentified Type 2s at the Genesis Project at the University of Miami. Genesis, a cloud-based database designed for storing and analyzing genomic data, recently identified a new form of CMT affecting an estimated 3,000 people in the United States. Howard is on the board of Genesis, which is available to the worldwide research community, making the challenge of identifying culprit genes easier. With more genomes and exomes from CMT patients available to researchers, the research community could identify many more forms of CMT.

Individuals with an unidentified form of CMT who have

their genome sequenced may have hundreds of disease-causing mutations, Howard explained. Most of these variants are harmless—everyone has thousands of mutations—and determining which one is not is the challenge. The challenge is much easier if there is a second and a third patient, who might be halfway across the world, with the same mutation. "It is a gross oversimplification to say that identifying culprits is a numbers game, but there is a sense in which this is true," Howard explained. He noted that the Genesis Project faces a number of barriers—i.e., many people with unidentified forms of CMT do not know that they have the disease and many who have been diagnosed have not had their genome or exome sequenced. In addition, some researchers and companies don't wish to share their data.

Breaking down these barriers is more important than ever given the progress being made with gene therapy and other treatments, Howard said, noting that the Genesis researchers who recently located a new gene also identified an existing medication that will treat and possibly cure the disease. "That sort of thing just wasn't possible even five years ago," he added.

Howard's own CMT journey was different than most. He had

HOWARD LANDIS

no significant symptoms when he was diagnosed 12 years ago at the age of 54. He grew up on a small farm in Pennsylvania and wasn't aware of any CMT symptoms in his earlier years although he has a high instep and was prone to ankle sprains. He went on to work in Manhattan as an accountant and consultant for a couple of years and then spent most of his career with a small private equity firm in Connecticut. He was referred to a neurologist by an orthopedist treating him for a shoulder injury. The orthopedist noticed some wasting in the area between his thumb and index finger and thought it might be CMT. A nerve conduction test confirmed his hunch.

Howard thinks his father, who passed away several years ago, probably had CMT. He was never seen by a neurologist, but

his genome has been sequenced and if Howard's culprit gene is identified researchers will be able to determine if he had the disease too. Howard has one sibling and one child and so far neither shows signs of CMT.

Howard initially viewed the CMTA as a resource for learning more about the disease, then later began attending

Patient/Family Conferences and online webinars. He says that his experiences have been uniformly positive and that he "continues to be amazed at the caliber and dedication of the

people that are working to support CMT patients and the research community that is working hard to find treatments and cures for CMT."

Howard says the CMTA fits into his philosophy of giving, which is whether it makes a difference. Ideally, that means giving to an

Now is the time to join the CMTA in supporting the research community...



Howard Landis

organization that is well run, he added, noting that the CMTA has a 4-star rating from Charity Navigator—and a clearly defined mission that has a reasonable chance of succeeding.

Currently retired and living in South Florida, Howard closely follows CMT research and lives out his own philosophy of giving—making a difference in the lives of everyone with CMT. ★



BECOME AN INNERVATOR!

Support the CMTA's Life-changing Initiatives with a Monthly Gift

Complete and return to: CMTA • PO Box 105 • Glenolden, PA 19036

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Charge my Visa MasterCard American Express Donate online at www.cmtausa.org/donate

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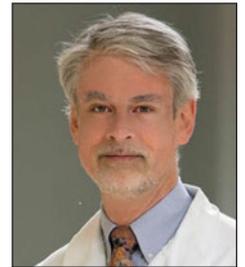
To give a gift of stock or learn about leaving a legacy gift to the CMTA, please call or email Jeana Sweeney, 800-606-2682 x106 / jeana@cmtausa.org.

Three New CMTA Centers of Excellence

The CMTA designated three new Centers of Excellence in 2020, bringing the international total to 40. Adding to the CMTA's wide network of clinical support ensures that as many people as possible have access to expert advice and care. Like all of the centers, they are led by professionals with deep experience in treating CMT.



NEW JERSEY DR. FLORIAN THOMAS, at the Hackensack University Medical Center in New Jersey, has cared for people with CMT and engaged in CMT research for over 30 years. He has long been active with CMT advocacy groups, and in 1998 co-founded the CMTA peer support group in St. Louis, Missouri. He is fellowship-trained in neuromuscular research and board-certified in neurology and neural repair and rehabilitation. He is the chair of the Neuroscience Institute and the Department of Neurology at Hackensack Meridian School of Medicine. He has published on several CMT subtypes, identified, with an international team of collaborators, a novel CMT disease gene and spearheaded two of the first clinical drug trials for CMT in the United States. Additional treatment studies are being initiated.



For appointments, call 551-996-1324 or email Annerys.Santos@HMHN.org.

COLORADO DR. MICHELE YANG is a pediatric neurologist at Children's Hospital Colorado with certification in neuromuscular disorders and electrophysiology. She trained at the Children's Hospital of Pittsburgh in pediatrics and child neurology, at Beth Israel Deaconess Medical Center in electromyography and at Children's Hospital of Philadelphia in pediatric neuromuscular medicine.



For appointments, call 720-777-3907 or email Allison.ballard@childrenscolorado.org.

CALIFORNIA DR. ALEXANDER FAY, at the University of California San Francisco (UCSF), is a pediatric neurologist with a focus on neuromuscular disorders. He obtained his PhD in biophysics and his MD from UCSF and completed his child neurology residency and neuromuscular fellowship at Washington University in St. Louis. Since joining the faculty at UCSF, he has devoted his time to identifying a novel form of CMT in a large family from South America, developing CRISPR-based therapeutics for CMT2, serving as an investigator on several clinical trials for childhood neuromuscular diseases and expanding his neuromuscular practice to include UCSF Benioff Children's Hospitals in both San Francisco and Oakland. He is a native of the Bay Area and is proud to be serving this community.



For appointments, call 415-353-7596 or email Audrey.glancy@ucsf.edu.

Named In 2020

CMTA CENTER OF EXCELLENCE

Cedars-Sinai Medical Center (Los Angeles).....	Drs. Matthew Burford and Richard Lewis
Children's Hospital Colorado (Aurora)*	Dr. Michele Yang
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Children's Hospital of Pittsburgh (Pittsburgh)*	Dr. Hodas Abdel-Hamid
Children's National Hospital (Washington, DC)*	Dr. Diana Bharucha-Goebel
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Kane Hall Barry Neurology (Dallas/Fort Worth)*	Dr. Sharique Ansari
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Ohio State University (Columbus).....	Dr. Samantha LoRusso
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Rush University (Chicago)*	Dr. Ryan D. Jacobson
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Wayne State University (Detroit).....	Dr. Jun Li

*These Centers of Excellence are not part of the INC.

INTERNATIONAL

The Children's Hospital (Westmead, Australia).....	Dr. Manoj Menezes
The National Hospital for Neurology & Neurosurgery (London, England)	Dr. Mary Reilly
C. Besta Neurological Institute (Milan, Italy)	Dr. Davide Pareyson
University of Antwerp (Edegem, Belgium)	Dr. Jonathan Baets

CMTA CENTERS OF EXCELLENCE are patient-centric, multidisciplinary CMT clinics where children, adults and families affected by CMT can be assured of receiving comprehensive care by a team of CMT experts. The Centers roughly correspond to the 21 international sites that make up the NIH Inherited Neuropathies Consortium (INC)—a group of academic medical centers, patient support organizations and clinical research resources sponsored in part by the CMTA. The centers will become even more important as the CMTA begins clinical trials, which will depend on how much we know about the “natural history” of CMT—how different types of CMT progress over time and whether novel medications are slowing the course of the disease. Much of that information will be supplied by the Centers of Excellence.

CMTA CENTERS OF EXCELLENCE
www.cmtausa.org/coe

Editor's Note: This piece is excerpted from Jeff's memoir, *The Fun Master*, which portrays his struggles with CMT and his journey as caregiver to a son in fragile health. The book will be published by SparkPress in 2021. Jeff wrote about his training routine in the Spring 2020 issue of *The CMTA Report*.

The Feet of Dorian Gray (An Excerpt)

BY JEFFREY SEITZER

I approached the waiting room of the CMT clinic at Wayne State University full of anticipation. In 30 plus years struggling with CMT, I had only met one person outside my family who had it. I imagined being greeted there by a throng of fellow CMT sufferers eager to share stories and exchange shoe tips. Alas, the waiting room was empty.

After a few minutes, I was led into an examination room, where I was joined by Dr. Michael Shy, who told me, "You have the strangest case of CMTX we have ever seen. In terms of your neural response, muscle mass, muscle tone, you appear to have a very mild case of Type 2. In a male, this is rather surprising. By age 60, men with CMTX typically require a cane or even AFOs to walk."

I explained about breaking out of the plastic leg braces in

my twenties because my calf muscles had gotten so big from vigorous exercise.

"I can imagine," he said. "Your calves look great."

I raised my eyebrows, surprised anyone would say that about my calves. I asked him if he agreed with the neurologist who told me that in some cases people plateau at a level with limited decline, then experience a precipitous decline much later. "Am I merely waiting for the other foot to drop, so to speak?"

I asked, secretly pleased with my play on words.

"I don't think so," he answered. "CMT is slowly progressive, so your condition will worsen. But you've established a certain trajectory. Your degeneration should continue along the line already established."

"Your case is interesting in another respect," he continued. "To understand how unusual it is, let me go over some of the most important aspects of your type of CMT."

I was looking at him while he spoke, but my mind was elsewhere. It was like a waking dream moving back in time. First, I was speaking with the neurologist at the University of Chicago, who said I might not even have CMT; then I was speaking with the mysterious stranger who told me about reconstructive foot surgery; followed by the 20-something

shoe salesman who suggested I get plastic rather than metal braces. All of them gave a new direction to my life. Sensing that this moment was important as well, I worked hard to concentrate on what he was saying.

"Your case is better than most CMTX cases," I heard him say. "There is less atrophy. Why is unclear."

"That's a lot to take in," I said, glancing at my notes, which looked like an experimental poem. Content to let my wife

Janet, family science officer, sort out the technical details, I focused on my new principal worry, the difference between my right and left sides. "If my left leg, the good leg, not the

right, is the lead indicator of the degeneration produced by CMT, then I feel I'm in good shape. Is that your sense as well?"

"Perhaps," he said. "We will have to study your case more. It may not be possible to determine that conclusively. The fact that you've had encephalitis and CMTX, though, is highly interesting. Your form of CMT involves a protein, Connexin 32. It is a little different from other proteins affected by CMT in that it is located in both the central and peripheral nervous systems. It is in a little different location in each. But there is a possible connection with encephalitis. In a very small number of cases, some activity like high altitude

In 30 plus years struggling with CMT, I had only met one person outside my family who had it.

Jeffrey Seitzer and son Ethan



INPUT ON CMT AWARENESS IN SCHOOLS SOUGHT

CMTA Advisory Board member and educator Sarah Kesty is seeking input from the community on how to encourage CMT awareness in schools.

Sarah entered a contest sponsored by Sarepta Therapeutics that asked educators to create lessons that build understanding of, and empathy for, rare diseases and those affected by them. The winning lessons will be posted on sharemylesson.com.

“Rare Lessons” is a program intended to promote the development and implementation of rare disease lessons in the K-12 classroom. According to the company, “We believe that rare disease education is an important component of disease awareness and diversity and inclusion within the classroom setting and that the study of rare diseases sets the foundation for enhanced education throughout students’ academic lives.”

Kesty said she was honored to participate in Rare Lessons, which fosters the empathy- and understanding-building she would like to see in all classrooms. Noting that her CMT was a particular challenge when she was younger, she said, “Lessons like these would have truly helped my classmates, teachers and me better understand the impact and nature of rare diseases.”



Sarah talks with Emaya G. about the characters in Sarah's book, *Everyone Has Something*.

Sarah is seeking answers to the following questions from the community:

- What would you like other students to understand about CMT?
- What do school leaders need to know to improve your child's experience at school?
- What challenges do you face in sharing about CMT with others?

Drop Sarah an email at info@cmtausa.org and she'll address your concerns in an upcoming article. ★

climbing or deep-sea diving can produce a metabolic shock, which, in turn, might cause a temporary central nervous system problem, in your case perhaps encephalitis, with some of the symptoms you've had.”

I was stunned. “Whoa,” I said. “What are the chances of that?”

“Not high. There might be 10 or 12 known cases of this happening.”

I just shook my head in disbelief. After a pause, he asked, “Well, do you have any other questions?”

After a deep breath, I said, “A neurologist told me that alcohol damages the sheathing around nerve fibers, so I should give it up immediately. I drink a

couple glasses of red wine every day,” I added, knowing it was often more, rarely less. “One is for my heart health, and the other because I like it so much.” The proportion was actually weighted decidedly toward the latter, but I figured he didn't need to know that. “Is it something I should cut out altogether?” I asked, bracing myself to receive the bad news.

“Whatever you are doing, keep it up. It's really working. In fact, you might want to share some of your experiences with the support group, particularly your approach to conditioning. They may benefit from it.”

“I'll do that,” I said, adding, “It might be a multivolume work.”

I was positively giddy while I waited for the elevator. You'd think I'd just been told that with a simple change of diet or some easy to take medication, all my troubles would be over. I still had to contend with the residual effects of encephalitis and the slow, progressive decline of CMT. But I would not end up like my mother or my grandfather, who were both severely hobbled at a much younger age than I was then. I could manage my condition well without fear of complete incapacity. For the first time since my diagnosis at age 12, I felt truly lucky. ★

Jeff teaches at Roosevelt University and has published recently in Adoptive Families Magazine, The Omaha World-Herald, Brevity Nonfiction Blog, Hippocampus, and elsewhere.

The Surgical Correction of the CMT Foot: IS SURGERY RIGHT FOR YOU?

PART 4 OF 4:

It Starts (and Ends) with the Toes

BY GLENN B. PFEFFER, MD



Dr. Pfeffer is an orthopedic foot and ankle surgeon and co-director of the CMTA's Center of Excellence at Cedars-Sinai Medical Center, Los Angeles. He is also a member of the CMTA's Advisory Board.

This is the last in our series on CMT foot surgery. I've saved the toes for last, but perhaps they should have been first, as they are the first affected by CMT neuropathy. Someone once wrote, "Toes are at the end of your feet, but the start of your journey," and that is certainly true when it comes to CMT.

CMT is a length-dependent neuropathy, which means that the muscles farthest away from the spine are affected first. The small muscles in the foot are therefore the first to weaken. These small intrinsic muscles stabilize the toes, keep them straight and help them move in a coordinated fashion. As these small muscles atrophy, the toes start to curl into what is commonly referred to as a "clawed" position (shown below).

At first, the toes remain flexible, but over time they become fixed in their clawed position. It is often the baby toe (fifth toe) that starts to deform first. Parents who are concerned about their children's feet might watch out for this clawing of the fifth



Clawed toes



toe as an early sign of CMT foot involvement, although there is no telling whether it will worsen.

It is important to recognize early on what is going on with the toes, as stretching exercises may help prevent a fixed contracture of the toe joints so that surgery can be avoided.

The stretching program is quite simple: Stretch each of the toe joints out into a straight position. If the middle joint (proximal interphalangeal joint) is flexing down, stretch it straight. If the metatarsal-phalangeal (MTP) joint at the base of the toe is extending up, stretch it down. Do the same thing with all of the other joints. Hold the joint straight for 10 seconds once or twice a day. A physical therapist can help you make sure you are stretching the right way. It shouldn't take more than one or two visits to help set you up with a home program. My dentist used to tell me to

"brush, brush, brush." I tell my CMT patients with toe problems to "stretch, stretch, stretch." Just like brushing, keep up the stretching for the rest of your life. Even if you already have some fixed contractures of the joints (where they won't straighten completely), a stretching program may help prevent further progression.

Sometimes even the best stretching program doesn't stop the toes from getting worse. It's important to understand what is going on in this situation. The foot intrinsic muscles, as we discussed, are usually affected first with CMT, but the tibialis anterior (TA) is often not far behind. The TA is the large muscle in the anterior leg that lifts the ankle. The toe extensor muscles also contribute to lifting the ankle (dorsiflexion).

You can see that when a normal ankle goes up, the toes go up as well. Once the TA starts

to weaken, the toe extensor muscles have to work harder and harder to help lift the ankle. And with the toe extensors working harder than normal, the toes begin to deform into a clawed position. It happens so slowly that few patients realize what is going on. Every day in the office I see a CMT patient who can move their ankle up but only because of the strong toe extensors. Good for the ankle, but ultimately very bad for the toes, which get deformed by the tendon over-pulling with each step.

As the toes deform into a clawed position a lot of bad things can happen to the foot. The cushioning fat pad underneath the toes starts to be pulled out of position. The bones (metatarsal heads) become more prominent on the bottom of the foot, with nothing but skin between them and the floor. Walking barefoot on a hard surface becomes intolerable. Because the toes are no longer in touch with the ground, balance is affected and push-off strength is diminished, which further compromises gait. Custom cushioned shoe inserts (orthotics) can help, along with a shoe that has a roomy toe-box (extra-depth shoes).

To understand what has to be corrected with surgery, it's useful to take a closer look at the toe deformity. There are three

joints in the small toes and two in the big toe, and they commonly all deform in different directions. That multiplane deformity is what makes toe surgery tricky. The loss of intrinsic muscle function causes the joint at the base of the toe, the metatarsal-phalangeal (MTP) joint, to extend upward. The middle joint of the small toe, the proximal interphalangeal (PIP) joint, flexes downward and the joint closest to the nail in the distal interphalangeal (DIP) joint can go in either direction. The goal of surgery is to straighten all the joints and end up with toes that fit comfortably into normal shoes.

There is no consensus on the best approach to toe surgery. While all surgeons agree that the release of soft tissue contractures is essential, they disagree on the role of PIP fusion, tendon lengthening and tendon release. Over the years, my approach to CMT toe deformity has changed. In non-CMT patients, division of the long flexor tendon (FDL) of the toe is commonly performed. The FDL muscle arises in the leg, extends down to the foot where it becomes tendinous and inserts onto the tip of the toe. The FDL tendon/muscle is often a deforming force, and its division can help correct the toe deformity. The division can be done through a tiny incision underneath the toe. Two other

muscles flex the toes down in a non-CMT patient, which means that division of the FDL will go largely unnoticed.

In CMT patients, however, these other muscles are often very weak or completely paralyzed. Preservation of the FDL is therefore important, as it is the only working muscle that provides flexion power to the toes.

An even more important reason not to divide a strong FDL is that it can be used to reconstruct ankle motion in the future if the CMT disease progresses. Please look at my Instagram (@Charcot-marietoothsurgery), and you will see many videos of patients who have avoided braces by transfer of the FDL to restore ankle motion. A surgical fusion of the PIP joint is often performed when the FDL is preserved to prevent a recurrence of the deformity. Fusions of the PIP joints have little impact on function. The small joint in the big toe, the interphalangeal joint, is another joint that often requires fusion during toe surgery, with hardly any compromise in function. A final key element of the surgery is to take the toe extensors, if they are strong, and transfer them to the top of the foot where they will help lift the ankle.

Understandably, toe surgery is usually the last priority for CMT patients. A crooked foot and loss of ankle function need to be corrected first to keep a person walking and out of a brace. Sometimes everything can be reconstructed during the same surgery, but not always. There is a limit to what the foot can tolerate during an operation, and a second surgery for the toes may be needed. It's a lot to go through, but absolutely worth it. The process is a marathon, not a sprint, but so gratifying when you win. ☆

This is the fourth in Dr. Glenn Pfeffer's four-part series on The Surgical Correction of the CMT Foot. The other three parts are:

Part 1: IS SURGERY RIGHT FOR YOU,
Winter 2020 CMTA Report, page 14

Part 2: TENDON TRANSFERS,
Spring 2020 CMTA Report, page 4

Part 3: THE ROLE OF JOINT FUSION,
Summer 2020 CMTA Report, page 21

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CMTA Zooms First Patient/Family Conference

BY LAUREL RICHARDSON

More than 200 people gathered for the CMTA's first Zoom Patient/Family Conference on Saturday, November 7, 2020, in a rare opportunity for attendees to hear from CMT experts worldwide.

Those experts covered a wide variety of topics. Dr. Steven Scherer and Tanya Bardakjian, MS, LCGC, both of the University of Pennsylvania (UPENN) presented on adult neurology/genetics, and Dr. Alexander Fay and Matt Hall, MS, of the University of California San Francisco (UCSF) gave a presentation on pediatric neurology/genetics.

Midday breakout sessions were led by CMTA Advisory Board members and/or therapeutic experts from CMTA Centers of Excellence. They included tangible, valuable information on physical therapy, bracing, emotional wellness and staying active with CMT.

The afternoon session began with a robust STAR research update led by CMTA Board Chairman Gilles Bouchard. He was joined by four researchers who sit on the CMTA Scientific Advisory Board: Drs. John

Svaren (University of Wisconsin), Steven Scherer (UPENN), Kleopas Kleopa (Cypress Institute of Neurology and Genetics), and Stephan Züchner (University of Miami).

Attendees then moved into breakout sessions by CMT type: demyelinating (Types 1, 4 and X) or axonal and unknown variants (Type 2). The type-specific sessions provided in-depth updates on the many research projects underway for each subtype.

Drs. John Svaren and Kleopas Kleopa led the demyelinating session. Dr. Kleopa updated attendees on his CMTA-funded studies that proved gene therapy is feasible in rodent models of CMT1X and CMT4C. The CMTA is actively supporting the efforts of several gene therapy companies to develop new CMT gene therapies. Dr. Svaren told listeners about the CMTA's partnership with InFlectis BioScience to develop agents to restore myelin protein balance for CMT1A and CMT1B. Phase 1 clinical trials have concluded and InFlectis is gearing up for Phase 2 trials.

Drs. Steven Scherer and Stephan Züchner led the other afternoon STAR breakout session in an in-depth look at the progress being made with CMT

Type 2s (axonal forms) and unknown variants. They talked about Dr. James Wilson's work with Passage Bio at the University of Pennsylvania using gene therapy to treat CMT2A. The CMTA is also funding the work of Drs. Bruce Conklin and Luke Judge of the Gladstone Institutes and the UCSF Departments of Medicine and Pediatrics, who are exploring the therapeutic application of genome editing technology (CRISPR) to CMT2A, CMT2E and CMT2F.

Dr. Züchner shared information about the newly discovered SORD gene. According to Züchner, "[T]here are over 3,000 patients in the US alone [with the SORD mutation] and the best part is we think it's treatable." When the breakout sessions finished, all attendees and speakers met back in the main Zoom "room" for a Q & A session with the panelists.

The silver lining of holding the conference online was that CMT patients from all over the world were able to participate. The CMTA will continue planning virtual education events and conferences in 2021 with the hopes of having as many people take part as possible. ☆

Laurel is the CMTA director of community outreach.

To view the 2020 Patient/Family Conference video recording, visit www.cmtausa.org/pfcs/.

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Widower Continues to Raise Funds in Wife's Name

BY MIKE DRIEDGER

“Look for the helpers,” Mr. Rogers said. Rick Clemente is one of them. While he doesn't have CMT himself, he raises money and awareness of the disease to honor his late wife Celia.

Rick has a rather unique approach to both. The retired physical therapist and human anatomy teacher is a skilled amateur woodworker who got his start making pens, letter openers and utensils to make Celia's life with CMT a little easier. She suggested that woodworking might be a nice retirement hobby and asked him to make pens and letter openers for every member of their family. After a couple of classes, he put a lathe in their garage and soon had it outfitted as a full-fledged workshop.

Celia died of cancer more than three years ago, but he still makes the pens and whenever anybody asks about them, he uses the opening to talk about CMT. He also offers to sell the pens. The cost? A donation to the CMTA.

Rick is committed to the organization because of the support and friendship that members of the Pittsburgh Branch showed Celia. Her diagnosis was a long time coming. She had no family history of CMT and was erroneously diagnosed with a number of other illnesses, including ALS. Visits to the Cleveland Clinic and the Mayo Clinic were fruitless. Not until a

After Rick's wife attended her first branch meeting, "She became herself again."

local neurologist suggested genetic testing was she finally diagnosed.

Rick says the most important thing in this story is the help the branch gave his wife, who taught the deaf. By the time Celia attended her first branch meeting, she was at wit's end, he recounts. "The people there could not have been more welcoming," he says, adding, "She found friends and companions who let her know she wasn't crazy." Celia was

struck by the fact that branch members looked just like her with their braces, wheelchairs and difficulty holding pens. The meeting had a profound impact on Celia, he says, and afterward, "She became herself again."

Rick's woodworking has slowed a bit due to arthritis in his hands. But he continues to attend branch walks with various family members and proudly wears his walk T-shirts. Like his pens, they provide an opening for him to talk about CMT. And because he is a helper, that's just what he does. ☆



Rick Clement with some of his pens

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WHAT'S ON YOUR MIND? *Ask David.*

Dear David:

I wanted to relate an incident that rattled me at first but ultimately had a happy ending. I am a middle school teacher in a small Midwestern public school. I love being a teacher despite some classroom difficulty with balance because of my CMT. I also have some weakness in my hands so I am always dropping materials, much to my kids' delight.

The other day I was in front of the classroom trying to teach a history lesson when one of my more rambunctious students blurted out, "What happened to your leg?" I always figured I would get this question someday, but it caught me off guard. I turned beet red and sheepishly tried to explain CMT to my class, but it's hard enough trying to explain my CMT to friends, let alone seventh graders. I have some feelings of shame and embarrassment but I hide them pretty well and try very hard not to let them prevent me from participating in and enjoying my life.

Fortunately, I had a break after the class and a colleague suggested that I take the opportunity to go back and lead a discussion on people that we know who have physical challenges. I was extremely nervous, but I did just that and the students were more than willing to give examples of people they knew who had physical problems. Many examples were from their own families: One boy spoke of his grandfather who

is in a wheelchair from a Vietnam War injury and spent lots of time teaching him about cars. Another spoke about her brother who had cerebral palsy. After a while, I honestly didn't know if our discussion was for them or me.

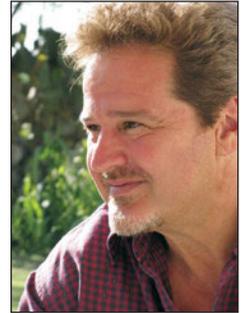
Even though I had a way to go in accepting my own CMT, I saw that I could transform my self-consciousness into helping others learn to accept their differences. The best part of the day came when the boy who had asked me about my CMT saw me struggling to carry materials out to my car and asked me if I wanted some help. Driving home I choked up with tears, thinking that maybe I taught them a lesson about empathy, the most important lesson of all. Just wondering how you think I handled the situation. —Ms. B.

David replies:

Dear Ms. B., Thanks so much for sharing a story that I know many in our community can appreciate. When someone asks "the question," most of us are often caught off guard. I think that we are all so private with our feelings about our CMT that we are shocked when that boundary is crossed. We are unaccustomed to sharing our thoughts and feelings in public. This is another good reason to attend support groups and CMT conferences whenever possible to receive this support from others who understand what we experience daily.

Your student probably triggered the feelings of inadequacy and vulnerability that we often go

David Tannenbaum answers questions from readers in his column "What's On Your Mind? Ask David" regularly in The CMTA Report. David has an LCSW degree and has been a psychotherapist in New York City for the past 30 years, specializing in helping others with the task of growing emotionally and spiritually through physical challenges. "My CMT has been my greatest challenge and my best teacher in life," says David.



to great lengths to hide. Our vulnerability is human and can be transformed into power if we use it to connect to others. Everyone feels vulnerable in some area of their lives, and many of us struggle with our imperfections. Be courageous in telling your story and you will find that many will respect your fearlessness. Remember that anxiety is who you think you should be and peace is who you are.

Telling your story gives others permission to tell their stories and believe me, after 40 years of practice as a therapist, I can tell you that everyone has one! When we freeze up in our self-consciousness, it is simply a reminder that we have a bit more work to do in releasing some old pain around our physical challenges. Identifying these thoughts, which often have to do with the past or the future, and then coming back to the present moment will help whenever you can remember to do so. Take this opportunity to identify and then gradually be free from your fear thoughts. Take pride in your being and know that your true identity has nothing to do with your physical body. Self-acceptance makes your light shine brighter and provides a powerful example for all to follow. You are a true teacher. ☆

Former Pastor Spreads the Word about CMT

BY MARCIA SEMMES

When David Loy and his wife Cathy give to the CMTA, they are giving not for themselves, but for future generations. “At 64 years old, I may not see the cure in time to help me, but I do not want another child to go through the ridicule, snubbing and multiple misdiagnoses that I did when I was young,” he says.

David’s road to diagnosis was full of twists and turns. Born in Iowa in 1956, he says it was apparent early on that something was not right: He couldn’t run, his feet were misshapen and he was always in pain. But doctors had no answers for his issues and often told his parents that it was all in his head.

In elementary and middle school, PE teachers told him he just wasn’t trying. He was picked last, or not at all, for teams, and the other kids made fun of him. That finally changed in high school. One teacher saw something others did not and brought him into the theater department, where he found a place he could both excel and hide.

After graduating from college with a business degree, David began working in the telecommunications industry, then moved to the educational testing field. After that, he “took a left turn” into non-profit work as the full-time pastor of Community of Christ in Cedar Rapids, Iowa. When the recession hit in 2008, he became the executive director of a non-profit that worked with families whose children had been taken away because of addiction and neglect. David’s last career move was to United Way of East Central Iowa, where he was a financial manager. He retired on disability in 2015.

David’s wife Cathy has accompanied him on his CMT journey since they married in 1979.

Luckily for him, she was a nurse and encouraged him to look for a diagnosis, but answers were still few and far between. Not until 1993 did he finally find out what was wrong, and then only by chance. He went to the Mayo Clinic for an eye problem and while he was there, a doctor took a look at his feet and said that he needed to have a neurologist do an EMG. The EMG confirmed CMT.

After David was diagnosed, he learned that his cousin in California also had CMT. In 2011, she directed him to the CMTA website for more information, which eventually led him, one “wonderful day,” to the newly opened clinic of Dr. Michael Shy. There he received wonderful care, benefited from Dr. Shy’s vast knowledge of CMT, had a DNA study to confirm that he had CMT1A and took part in a clinical trial.

Seeing Dr. Shy made him realize, he says, that the CMTA “was not just a source of reliable information, but also the leading organization in the fight to find the cure.” Since that time, he and Cathy have tried to give what they can for this fight and to encourage others to do the same.

David is aware that the search for a cure for any disease is a slow process with many medical and legal hurdles, but he thinks the CMTA’s search “is moving at a steady and often breakneck pace in comparison to other diseases.” That’s why he gives and that’s why he spreads the word about the disease,

The CMTA is “not just a source for reliable information, but also the leading organization in the fight to find the cure.”

sporting a shark and nerve tattoo that provides an easy opening to tell anyone who asks about CMT.

The former pastor is still preaching these days, but now his sermon is about

CMT. There are many places to put your money, he says, but this crippling disease will be cured within a very reasonable time. “Great advancements have been made,” he says, adding “Wouldn’t you like to say you helped eliminate it from this world?” ☆



David Loy



CMTA BRANCH NEWS

SAN DIEGO, CA

Eight members of the San Diego Branch welcomed orthotist Ara Mirzaian from the Hanger Clinic to their December 5, 2020, meeting. He shared great information and answered members' questions on bracing and orthotic options for people with CMT. A member who has benefited from Ara's years of prosthetic and orthotics experience recommended that anyone in need contact him at his Encinitas office with questions regarding bracing and orthotics for CMT.

★

DESTIN, FL

It didn't take long for the nine people attending the September 26, 2020, inaugural meeting of the Destin, FL Branch to begin sharing their CMT stories and building camaraderie. Members bonded over common experiences and shared information about pain management, bracing, medications, physical therapy, occupational therapy and resources available through the CMTA. Laurel Richardson, the CMTA director of community outreach, talked about the organization and its purposes. The group also discussed pre-

senters and topics of interest for future quarterly meetings, which in the short run will likely be virtual.

★

NEWBURY, NH

Sara O'Dougherty from Granite State Independent Living joined five members of the Newbury, NH Branch on Zoom October 1 to share valuable information about helping people live independently. Sara explained that the agency's goal is not just to provide transportation for disabled people, but to also teach them about home care, adding accommodations into their homes, building ramps and much more. The next meeting will be in February.

★

TULSA, OK

The Tulsa, OK Branch welcomed guest speaker Laurel Richardson to its fun and festive branch meeting on December 5. Everyone wore holiday attire or "ugly Christmas sweaters" and played fun virtual games. They also talked about how everyone is doing this year and how everyone is dealing with CMT.

★

BUCKS COUNTY, PA

The Bucks County Branch welcomed guest speaker Dragan Vujovic to its November 21 meeting. Dragan is working on the ACT-CMT study with Dr. Steven Scherer at the CMT Center of Excellence at UPenn in Philadelphia. He talked about what kinds of data are being gathered through the study, how it will contribute to CMT treatments and what is on the horizon for other types of CMT research. The group also spent time talking about how everyone is holding up during such a challenging year.

CHESTER COUNTY, PA

Nine members of the Chester County Branch welcomed Jonah Berger, the CMTA national youth programs manager, to their December 12 meeting. He told the group about the huge success of taking Camp Footprint online in 2020 and upcoming projects. He also talked about his personal experience with CMT and how his positive attitude helps him to stay motivated to enjoy life. The branch also discussed the results of their fundraising efforts in 2020, which totaled \$7,200 between virtual walk and bingo events. They reviewed the CMTA Patient/Family Conference topics available online and a member shared a favorite session from attending the live conference.

★

PITTSBURGH, PA

Eleven members of the Pittsburgh Branch welcomed three guest speakers to their November 14 meeting: Elizabeth Ouellette, Laurel Richardson and Julie Tarle. Elizabeth, who is a longtime patient advocate and CMTA board member, shared an incredibly informative presentation on "CMT & Mythbusting: Separating Fact from Fiction." The presentation helped to eliminate some of the confusion surrounding CMT as it relates to other diseases. Laurel shared information about resources and education for managing and living with CMT. She also talked about the many upcoming virtual meetings and events available to the community, which can be found at www.cmtausa.org. Finally, Julie, the CMTA's special events manager, shared a WALK 4 CMT update and talked about the organization's amazing strides in event fundraising.

CMTA WELCOMES NEW FLORIDA BRANCH

The CMTA welcomed a new branch to the family in 2020, this one in Destin, Florida. Branch leader Ted Spring is determined to tell the world about CMT.

Ted and his wife moved to Destin not too long ago and soon after he bravely raised his hand to start a branch in that region. Due to COVID, they have only met virtually but the group looks forward to getting together in person when it's safe to do so.

Ted said he wanted to start a branch to help others avoid some of the issues he faced on his road to diagnosis. He was misdiagnosed with herniated discs in 1968 and underwent spinal surgery. The symptoms didn't abate with surgery and he wasn't diagnosed with CMT until 2015. Orthotics changed his life he says, not just because they helped him to walk but also because of the interest the braces attracted. The many questions he got about the braces gave him an opening to talk about CMT. The experience left him determined "to do my part to inform, educate and help people benefit from the work of CMTA," he said.

Ted says he continues to be "surprised" about the lack of knowledge people have concerning the disease and the availability of support services once they have been diagnosed. "Support services, including CMTA branches, help people get connected, informed and more involved in treating their CMT," he says.

To find a branch in your area, go to www.cmtausa.org/branches/.





CMTA Branches

Most CMTA Branches can be accessed
online at www.cmtausa.org/branches

ALASKA

Anchorage Area

Lisa Hubert
907-223-4566

ARKANSAS

Little Rock

Laurel Richardson
800-606-2682, Ext. 112

ARIZONA

Phoenix Area

Pam Palmer
480-236-2445
Christina Fisher
623-742-8921

CALIFORNIA

Antelope Valley Area

Donna Murphy
661-317-6332
Danielle Metzger
661-317-6533

Los Angeles Area

Alani Price
310-710-2376
John Ramos
951-318-5669

Orange County Area

Beth Dorin
949-929-2908

Sacramento Area

Laurel Richardson
800-606-2682, Ext. 112

San Diego Area

Annette Van Veen
760-473-5014
Kendall Trout
760-632-5654

South Bay Area

Ori Bash
408-829-4562
Tau O'Sullivan
916-806-2173

COLORADO

Denver Area

Ron Plageman
303-929-9647
Dick Kutz
303-988-5581

CONNECTICUT

Hartford

Roy Behlke
239-682-6785
Diane Lindsay
860-670-4417
North Haven
Lynne Krupa
203-288-6673

DISTRICT OF COLUMBIA

Washington, DC

Steven Weiss
Kimberly Hughes
301-962-8885

FLORIDA

Central Florida

Linda Davis
Mitch Davis
863-875-4239

Destin Area

Ted Spring
850-368-1097

Jacksonville Area

Tim Nightingale
904-504-1953
Stephanie Burkhalter
904-710-3771

Melbourne Area

Laurel Richardson
800-606-2682, Ext. 112

Naples

Roy Behlke
239-682-6785
Bill Millar
202-309-5685

Sarasota Area

Rachel Rivlin
941-284-0766

Tampa Bay Area

Edward Linde
813-712-4101
Sarah Gentry
941-706-5348

West Palm Beach

Phil Lewis
561-307-0100
Eileen Martinez
561-901-5566

GEORGIA

Atlanta Area

Jeannie Zibrida
404-307-6519

HAWAII

Honolulu Area

James Cuizon
808-450-1236

IOWA

Iowa City Area

Jeffrey Megown
319-981-0171

ILLINOIS

Chicago Area

Jay Pate
630-888-4673
Doreen Pomykala
815-351-1328

INDIANA

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Aimee Trammell
574-304-0968
Indianapolis Area
Laurel Richardson
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KANSAS

Kansas City Area

Tammy Adkins
314-608-6889
Aron Taylor
913-744-5674

St. Louis Area

Payton Rule
618-401-4822
Amanda Rule
618-698-3039

Springfield Area

Jessica Brantner
417-468-8049
Jessica Hardy
417-434-1656

Wichita Area

Karen Smith
316-200-0453

LOUISIANA

Baton Rouge Area

Laurel Richardson
800-606-2682, Ext. 112

MAINE

Portland Area

Mary Louie
207-450-5679

MARYLAND

Baltimore

Clark Semmes
410-350-4812
Easton
Laurel Richardson
800-606-2682, Ext. 112

MASSACHUSETTS

Boston

Mimi Works
617-913-4600
Jill Ricci
978-887-1014

MICHIGAN

Grand Rapids Area

Laurel Richardson
800-606-2682, Ext. 112
Amy Reynolds
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Mid-Michigan Area
Jonah Berger
303-827-4218

MINNESOTA

Central Minnesota

Jo Smith
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Minneapolis Area
Laurel Richardson
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MISSOURI

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Aron Taylor
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Amanda Rule
618-698-3039

Springfield Area

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417-468-8049
Jessica Hardy
417-434-1656

NORTH CAROLINA

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Carrie Johnsen
704-904-2828
Mona Doshi
980-339-8560

Durham Area

Jeanne Boehlecke
919-942-7909
Rick Nelson
919-889-9776

Wilmington Area

Laurel Richardson
910-515-8488

NEBRASKA

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Brandon Lederer
402-680-0502

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Newbury Area

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Central New Jersey

Mark Willis
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Jacqueline Donahue
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Albuquerque Area

Gary Shepherd
505-296-1238

NEVADA

Las Vegas Area

Martha Boadt
231-852-4251

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Buffalo Area

Peter Morris
716-866-3519
Maryann Ciskal
716-435-3899

Kristen Braun

716-270-3095
Syracuse Area
Michael Casey
315-439-9325

Albany Region

Cara Leath
914-262-1108
Westchester Area
Beverly Wurzel
Frank Wurzel
201-224-5795

OHIO

Cincinnati Area

Jill Stuhlmuller
513-254-4065
Jo Koenig
513-607-2822

Cleveland Area

Shelly McMahon
440-781-8329
Columbus Area
Jessica Diamond
216-570-6432

OKLAHOMA

Tulsa Area

Lonna Henry
918-961-1418
Natasha Karki
405-413-1831

OREGON

Grants Pass

Jessica Barton
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541-846-8525

Portland Area

Laurel Richardson
800-606-2682, Ext. 112

PENNSYLVANIA

Bucks County Area

Julie FitzGerald Schell
315-573-3919
Chester County
Carol Aruffo
610-405-9291

Ashley Trout

484-364-9334
Harrisburg
Erin Weierbach
717-379-7504

Johnstown Area

J.D. Griffith
814-539-2341
Jeana Sweeney
814-269-1319

Northwestern Area

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Pittsburgh
Debra Czarnecki
412-331-6744

SOUTH CAROLINA

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Zack Boyd
803-622-6565
Kyle Bryant
803-378-6202

Greenville Area

Rebecca Lauriault
864-918-2437

TENNESSEE

Nashville Area

Teresa Shoaf
615-772-8810

TEXAS

Austin Area

Nate Halk
512-415-6097
Dallas/Fort Worth
Thomas Roderiguez
817-913-8477

El Paso Area

Jo Ann Longoria
915-526-0676
Olivia Longoria
915-491-0786

Houston Area

Miranda Burge
512-784-3086

VIRGINIA

Blacksburg/ Roanoke Area

Karen Brown
540-252-9453
Fredericksburg
Leigh Van Doren
540-370-1968

Harrisonburg Area

Laurel Richardson
800-606-2682, Ext. 112
Suffolk Area
Holly Hall
757-477-6122

WASHINGTON

Seattle Area

Denise Snow
206-321-1261
Emily Osborne
425-220-4225

WEST VIRGINIA

Charleston Area

Karen McClure
304-548-4413

WISCONSIN

Madison Area

Debi Weber
608-712-8709
Milwaukee Area
Lois Hawkins
414-249-0390

CANADA

Southern Ontario

Kelly Hall
519-843-6119
Toronto Area
Linda Scott Barber
416-997-5084

Michael Driedger

647-680-7601

Interested in starting a branch in your area?

Contact CMTA Director of Community Outreach
Laurel Richardson at laurel@cmtausa.org.

Like Father, Like Son

BY MARCIA SEMMES

CMT tears some families apart, but in a strange twist of fate, it brought Kansas City Branch Co-Leader Aron Kyle Taylor together with the 19-year-old son he had never met.

Aron is well known in the CMT community, not just as part of a mother-and-son pair of co-leaders, but also as a musician, a rapper and the composer of “Giving Toesday,” the CMTA’s 2020 post-Thanksgiving fundraiser.

Aron is from the small town of Parsons, Kansas, but has lived in the Kansas City area for the past 15 years. He was diagnosed with CMT in the second grade, though his specific type (2A) wasn’t confirmed until 2019.

Even with the challenges of CMT, Aron was able to keep playing varsity baseball as a pitcher and infielder through his senior year of high school when he was sidelined by a foot injury. Today, he continues to struggle with stairs and occasional falls but feels fortunate that his CMT is still fairly mild.

Now 43, Aron is an IT professional at a law firm in Kansas City, with experience in tech training, public speaking and resume building. In that capacity, he gives motivational talks to various groups. So when his friend Jess asked him to speak at a job fair for a group of young adults, he said yes without hesitation. After Aron’s talk about resumes and interviews, his

friend Jess was talking to a participant named Ethan Kyle Sargent (who goes by Kyle), a computer programmer.

As the three spoke, Jess observed that Kyle and Aron had a number of similarities in addition to working in the same field, including their distinctive CMT legs. Kyle had never met anyone with CMT before so Aron invited him to a branch meeting. When Aron gave Kyle a business card

bearing the name of his music production company, Ridiculous Beats, Kyle remarked that he was a musician too. Aron noted that “Kyle” was also his middle name.

The lightbulb moment came when

Aron asked Kyle his last name. On being told that it was Sargent, the name of a long-ago girlfriend, Aron threw his hands in the air and exclaimed to the entire room, “This is my son.” Simultaneously, Kyle said, “This is my dad.”

Since making their “Hallmark movie connection” on October 18, 2019, Aron and Kyle have talked multiple times daily. They live just 20 minutes apart and see each other several times a week. Noting that Kyle programmed video games at the age of 14 and is a talented guitarist, the proud father praises his son as smart and funny.

Aron looks forward to being able to help his son navigate some of the difficulties of CMT, like braces, and says he feels a responsibility to be a role model for his newly discovered son.

Together the two visited a counselor, who noted the simi-

Since making their “Hallmark movie connection” Aron and Kyle have talked multiple times daily.



Aron Taylor and son Kyle

larities in their posture and body language and quickly recognized the deep love Aron has for his long-lost son, “This is meant to be,” she told them. Aron agrees, adding “It’s a happy ending.”

It’s also a new beginning: Sharing their love of music and technology, the father and son are already collaborating on music together. “It’s going to be beautiful,” Aron says. ★



CMT PATIENT MEDICATION ALERT:

Definite high risk (including asymptomatic CMT):

Taxols (paclitaxel, docetaxel,
cabazitaxel)
Vinca alkaloids (Vincristine)

Moderate to significant risk:

Amiodarone (Cordarone)
Arsenic Trioxide (Trisenox)
Bortezomib (Velcade)
Brentuximab Vedotin (Adcetris)
Cetuximab (Erbix)
Cisplatin and Oxaliplatin
Colchicine (extended use)
Dapsone
Didanosine (ddl, Videx)
Dichloroacetate
Disulfiram (Antabuse)
Eribulin (Halaven)
Fluoroquinolones
Gold salts
Ipilimumab (Yervoy)
Ixabepilone (Ixempra)
Leflunomide (Arava)
Lenalidomide (Revlimid)
Metronidazole/Misonidazole
(extended use)
Nitrofurantoin (Macrochantin,
Furadantin, Macrobid)
Nitrous oxide (inhalation abuse)
Nivolumab (Opdivo)
Pembrolizumab (Keytruda)
Perhexiline (not used in US)
Pomalidomide (Pomalyst)
Pyridoxine (mega dose of
Vitamin B6)
Stavudine (d4T, Zerit)
Suramin
Thalidomide
Zalcitabine (ddC, Hivid)

Uncertain or minor risk:

5-Fluorouracil
Adriamycin
Almitrine (not in US)
Chloroquine
Cytarabine (high dose)
Ethambutol
Etoposide (VP-16)
Gemcitabine
Griseofulvin
Hexamethylmelamine
Hydralazine
Ifosfamide
Infliximab
Isoniazid (INH)
Lansoprazole (Prevacid)
Mefloquine
Omeprazole (Prilosec)
Penicillamine
Phenytoin (Dilantin)
Podophyllin resin
Sertraline (Zoloft)
Statins
Tacrolimus (FK506, Prograf)
Zimeldine (not in US)
α-Interferon

Negligible or doubtful risk:

Allopurinol
Amitriptyline
Chloramphenicol
Chlorprothixene
Cimetidine
Clioquinol
Clofibrate
Cyclosporin A
Enalapril
Glutethimide
Lithium
Phenelzine
Propafenone
Sulfonamides
Sulfasalazine



The Charcot-Marie-Tooth Association
P.O. Box 105
Glenolden, PA 19036
1-800-606-CMTA (2682) FAX (610) 499-9267
www.cmtausa.org

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WHAT IS CMT?

- ▶ More than 3 million people worldwide have CMT, which is one of the most commonly inherited nerve disorders and affects the motor and sensory nerves.
- ▶ CMT is slowly progressive, causing the loss of muscle function and/or sensation in the lower legs and feet, as well as hands and arms.
- ▶ Men and women in all ethnic groups may be affected by CMT.
- ▶ CMT is genetic, but it can also develop as a new, spontaneous mutation.
- ▶ CMT can vary greatly in severity, even within the same family.
- ▶ CMT causes structural deformities such as high-arched or very flat feet, hammertoes, hand contractures, scoliosis (spinal curvature) and kyphosis (rounded back).
- ▶ CMT can also cause foot drop, poor balance, cold extremities, cramps, nerve, muscle and joint pain, altered reflexes, fatigue, tremor, sleep apnea, hearing loss and breathing difficulties.
- ▶ CMT rarely affects life expectancy.
- ▶ Some medications are neurotoxic and pose a high risk to people with CMT, notably Vincristine and Taxols. See full list (at left) of medications that may pose a risk.
- ▶ More than 100 different genetic causes of CMT have been identified.
- ▶ Many types of CMT can be determined by genetic testing. Please consult with a genetic counselor (www.nsgc.org) or your physician for more information.
- ▶ Although there are no drug treatments for CMT, a healthy diet, moderate exercise, physical and/or occupational therapy, leg braces or orthopedic surgery may help maintain mobility and function.
- ▶ The CMTA's STAR research program and extensive partnerships with pharmaceutical companies are driving remarkable progress toward delivering treatments for CMT, bringing us closer to a world without CMT.