Steffens Scleroderma Foundation
Breaking the Chains

BREAKING THE CHAINS NEWSLETTER

March 2022

Degos Disease Patient Experience

Degos Disease is an ultra-rare disease and is a joint focus of the Steffens Scleroderma Foundation's endeavors. Steffens co-sponsored the Degos Disease international symposium at the National Institutes of Health (NIH) in 2018. Additionally, Dr. Lee Shapiro, our Chief Medical Officer, hosts a monthly Degos research workshop in which NIH researchers participate.

In this newsletter, our board member, Theresa Slayton, provides a first-hand account of her recent visit to NIH. We thank her for disclosing her personal experience and for her direct participation in this groundbreaking and vital Degos research at NIH.

NIH Research Study

It is important that all Degos patients, their caregivers, treating physicians, and other healthcare providers, participate in the current studies being conducted at the National Institutes of Health (NIH), which includes, National Heart, Lung, and Blood Institute (NHLBI), Vascular Institute, Malignant Atrophic Papulosis, Genetic and Rare Diseases Information Center (GARD), and NCATS Program (nih.gov). I encourage all patients who are diagnosed with or suspected of having a Degos diagnosis, to contact the National Institutes of Health (NIH) to become involved. This research is vital in the pursuit of a best practice plan of care for this ultra-rare disease. I share with you my most current experiences participating in their studies.

Following multiple communications and updates from Katherine Carney, Research Nurse with the Vascular Branch of NHLBI at NIH, I felt I was prepared to participate in a week of research studies. This was my fifth visit. Each visit has been a full week of studies so that the medical community can better understand **Malignant Atrophic Papulosis (MAP)** otherwise known as Degos disease. I feel blessed that the NHLBI at the NIH chose the Degos community to study. Marta Cardenas was a wonderful Patient Care Coordinator (PCC) and kept my husband and me on track as we moved through the daily schedule of tests and consultations.

I was informed that the heart and vascular divisions have separated to better study specific areas of the vascular system, and new government policies made it possible to share research results anonymously thus improving key public health capabilities. The logistics of becoming involved with these NIH studies are well mapped out, even providing for accommodations and reimbursement of travel costs for the participants.

Day #1 began with consent signing and blood work. Next there was an EKG, a history and physical with Becky Hufstetter NP, an extensive dermatology consult with Kelli Karacki, P.A. C, Dr. Leslie Castelo-Soccio, and Dr. Keisuke Nagao. Dr. Nagao reported that they learned valuable information from the special deep tissue skin biopsy that was done on my prior visit. They were able to identify specific immunoreactions that show that Degos disease is likely an autoimmune disorder. Next there was a surgical consult with Dr. Andrew Blakely regarding possible surgeries on my next visit. This

Bruce Cowan joins the Board of the Steffens Scleroderma Foundation.

Bruce joined our board in January 2022. He has been passionately involved with the scleroderma community for over 25 years. His wife was diagnosed with scleroderma in 1993 and tragically lost her battle to this difficult disease in 2000. He was a volunteer, a board member and for the past 5 years the Executive Director of the Tri-State Chapter of the Scleroderma Foundation, retiring in July 2021. He has worked with many of the fine people connected to the Steffens Foundation since its inception over a decade ago. Attending, assisting, and collaborating on a number of Steffen's activities and initiatives. It is his desire to bring his understanding of scleroderma as a caregiver, his business management experience, and board knowledge to the foundation, with the mutual goal to grow our outreach and expand our mission going forward in this next decade.

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follow up will be sooner than my typical one year follow up. My brain MRI had to be rescheduled for later in the week.

Day #2 included a Gastrointestinal (GI) consult with Dr Walter Lai to discuss future procedures that will assess the progression of Systemic Degos disease involving my GI system. They recommended this take place sometime over the next several months. I had an EndoPAT procedure done in vascular lab, which checks for healthy veinal and arterial health. I also received my first IV for PET CT and finally an Echocardiogram study. Day 2 was a busy day.

Day #3 included a Near Infrared Spectroscopy Study (NRIS), a Vascular consult with Dr. Kanthi and Janet Valdez, a second IV for an eye angiogram with Dr. Edmond Fitzgibbon. I also had a Pulmonary Function Test (PFTs), and a 6-minute walk test followed by a Pulmonology Consult with Karry Ryan PA-C, and Kenneth Oliver MD. This was undertaken to assess my progressive exertional dyspnea (shortness of breath during exertion). They were able to schedule the brain MRI from day 1 to the end of day 3.

Day#4 began with a third IV for a chest and whole-body Magnetic Resonance Angiography (MRA), Digit Ankle-Brachial Index (ABI) which checked blood pressures peripherally, a Coronary CT angiography, and a Rheumatology Consult with Dr. Cudricci. My day wrapped up with a carotid ultrasound.

Day#5 started with a Cardiac MRI and a targeted MRA of my left subclavian artery, both of which took about 2 ½ hours. The Rheumatologist requested additional bloodwork. We were able to start driving home around noon of day 5.

Given the knowledge that the NIH research community has already acquired from participating Degos patients, they requested we remain involved with them, so researchers can continue with further testing to better understand this disease. Their current focus is on the progression of Degos disease to the Central Nervous System (CNS), which is the brain and spinal cord. My personal experience when communicating with Degos patients from around the globe, is that I have heard of this CNS involvement presenting in young adults and children. I plan to offer all that I can by participating with NIH so that the Global Medical Community will someday have an immediate plan of care for their patients presenting with signs of Degos disease (MAP).

Student Research Projects

Giovanna Fichera is completing her entry-level doctorate degree in occupational therapy (OT) at Russell Sage College. She became interested in Scleroderma after attending the **Steffen's Scleroderma IPE** event in 2019. Ms Fichera continued to pursue this interest with three classmates, who focused their master's research around this under-investigated, autoimmune disease. The graduate students completed a study titled "3D Hand Model for Splinting Scleroderma Patients." This investigation resulted in preliminary data analysis to facilitate application of this novel splinting technique to actual patients which is Ms. Fichera's doctoral research project taking place over a 14 week period beginning in May of 2022.

Get a jump start on Spring!

Surprise Mom for Mother's Day!

Steffens Scleroderma Foundation is hosting an online fundraiser with Flower Power Fundraising.

50% of the proceeds go to the foundation

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Store closes May 15th

The purpose of this study is to determine the feasibility of a non-contact custom splint fabrication method for patients with chronic diseases suffering from hypersensitive or compromised skin integrity. Custom splinting by occupational therapists involves molding low-temperature thermoplastic material directly on patients' hands. Skin fragility and complex contracture patterns make this a daunting if not impossible task with certain populations. The study aims to recruit 20 male and female patients with either a diagnosis of scleroderma or arthritis. Participants will enroll in the study which is being conducted at Albany Memorial Hand Center.

Using photographs from a scanning application, and a 3D printer to create a precise replica of a patient's hand prior to splinting, Ms. Fichera will fabricate a custom wrist-hand-finger orthosis on the 3D model, thereby avoiding direct contact of the material on the compromised tissue of the patient's hand.

The overarching goal of this project is to create effective interventions for these patient populations by expanding splinting options and addressing the challenges associated with certain diseases. The anticipated outcome is to contribute to evidence-based practice in OT, evaluate the feasibility of the technology, improve patient outcomes, and promote best practice. The research partnership between Russell Sage College, Albany Memorial Hand Center, Steffens Scleroderma Foundation, and Precision Valve and Automation, promotes interprofessional collaboration and scholarship by laying the groundwork for future studies with custom splinting using 3D printing.

An Analysis of Referral Timelines and Intervention Patterns for Hand Therapy in Systemic Sclerosis

Hannah Bowen, Steffens research assistant, is in the process of launching three surveys with several healthcare professionals. The surveys are intended for physicians, Scleroderma patients, and hand therapists, to study referral timelines and intervention patterns for hand therapy in Scleroderma. In Scleroderma the hands are often impacted early in the onset of the illness, and may be the first to suffer from permanent damage (Fennell et al., 2020). Treatment for hands impacted by Scleroderma may be delayed or non-existent, leading to limited hand function or lifelong pain. Certified Hand Therapists (CHTs) may have patients referred to them late in the disease, or not at all, leading to poor outcomes. Raising awareness to initiate therapy in the early stages of the illness, may provide better outcomes for Scleroderma patients. This survey research aims to better understand the current availability and access to specialized upper extremity treatment for individuals with Scleroderma. It is important to examine referral by physicians, treatment from hand therapy specialists, and ease of access for the person with Scleroderma, to begin to effect the important changes needed in all three domains.

RAREDISEASEDAY.ORG

Rare Disease Day is February 28, 2022

Rare Disease Day is recognized globally to raise awareness among policymakers and the public about rare diseases and their impact on patients' lives. There are over 7,000 rare diseases that impact over 400 million people around the world.

The Steffens Scleroderma Foundation supports and promotes awareness for research toward treatments and cures for Scleroderma and Degos disease, which are rare diseases.