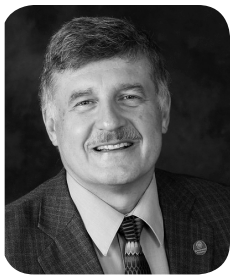
Clinical Trials — Our New Reality

By Mathew J. Toth, Science Director, Barth Syndrome Foundation

"We need our Barth brothers to step up and volunteer as much as they can. These volunteers not only have the potential to help themselves, but they also will help their fellow Barth brothers who cannot participate, and they may help other people afflicted with similar serious diseases." ~ *Matthew J. Toth, PhD, Science Director, Barth Syndrome Foundation*



Dr. Matt Toth

It has been almost 12 years since I first came to the Barth Syndrome Foundation (BSF) and attended its biennial conference. At the 2006 BSF conference, I was introduced to many of you, and I spoke briefly about what we all wanted to hear — how will we find a treatment for Barth syndrome? Though it has taken a long time, the hope I spoke about in 2006 is now coming into focus.

At the 2016 BSF conference (and before), we began to hear about the term clinical trials, and several other BSF members and I spoke to many of you about what clinical trials were and how important they are in the process of finding a specific treatment. I gave a presentation about eight distinct therapeutic ideas that were being pursued. In April of 2017, the company Stealth Biotherapeutics initiated a clinical trial with Barth syndrome individuals testing their lead compound Elamipretide — the TAZPOWER study. That study is soon to be followed by another pharmaceutical clinical trial using the pharmaceutical Bezafibrate which will take place in the United Kingdom — the CARDIOMAN study. We anticipate a third trial starting soon that involves gene therapy, which is very exciting due to its revolutionary approach for treating human disease. Clinical trials of other therapies are also being planned that will keep up the therapeutic assault on this cruel disease. BSF is very fortunate and blessed to have researchers, clinicians, and members who have worked hard and struggled to be where we are now — enrolling volunteers to test specific treatments for Barth syndrome.

Just as BSF asks for donations, BSF now asks for volunteers for these clinical trials. What I said to you in 2006 has now reached a point where therapies have to be tested. Our small community needs to realize that without the timely volunteering for clinical trial testing, nothing will be advanced. If we do not complete these initial clinical trials, there will not be any more of them. Researchers and drug developers will lose interest and enthusiasm for finding a treatment for Barth syndrome. Guys with Barth syndrome will never get better.

Clinical trials are experiments on humans that test the usefulness of a particular therapy in a scientific way. All clinical trials benefit from what has come before, and volunteering for any clinical trial has great value. No one can predict whether one therapy or another (or a combination of therapies) will be useful if at all, but the process of performing any clinical trial gives the healthcare and research community a great deal of information about the disease it is tested on. We need our Barth brothers to step up and volunteer as much as they can. These volunteers not only have the potential to help themselves, but they also will help their fellow Barth brothers who cannot participate, and they may help other people afflicted with similar serious diseases.

I recently attended several meetings at the major funding and drug approval arms of the US government — the National Institutes of Health (NIH) and the Food and Drug Administration (FDA), respectively. There is new thinking taking hold at both of these organizations that heralds a renaissance in how advances in healthcare are made. A remarkable consortium of academic, industrial, and patient-advocate organizations is focusing their efforts on the patient and on rare disease sufferers in particular. The NCATS division (National Center for Advancing Translational Science) of the NIH has put together a "toolkit" to do the very things BSF has been doing for years, and it describes what we should be and what we are doing in this era of clinical trials for Barth syndrome. No rare disease will be left behind by NCATS. Patient advocate organizations like BSF are an integral and essential part of this consortium. BSF represents the only people who can volunteer to test these new drugs/treatments for Barth syndrome in the clinical trials.

The vision I described at the 2006 BSF conference is here now. This reality of the need for volunteers from our community to test therapies is exciting but sobering. The research that the BSF has supported over the years through its grant program, through its conference clinics that members have participated in, and through the scientific-medical discussions/debates that take place at the conferences and beyond, have all contributed to where getting us we are now. BSF is in a positon to finally reap the rewards of what it has carefully sown and cultivated over the years. The major medical research and pharmaceutical approval agencies of the US government are now explicitly encouraging and endorsing what BSF has been doing since its inception — fostering a vibrant and caring scientific-medical-patient community to help our boys, and others like them. We are united in a struggle to lessen the suffering of not only our own members, but that of others with similar diseases. Please make sure to help us make the future better and consider volunteering for clinical trials.

(Photo courtesy of Amanda Clark [2016])