Gene Therapy Advances for Canavan Disease

Gene therapy is still in its early stages and more research is needed before the therapy will be ready for clinical trials for humans, but as the FDA’s chief, Scott Gottlieb, recently said, “What was once just a theory may have the potential to treat and cure some of our most intractable and vexing diseases.”

This summer a new company, ASPA Therapeutics, brought together researchers and parent leaders to discuss the needs of the Canavan disease community. ASPA, named after the gene aspartoacylase that is mutated in children suffering from Canavan disease, was formed to help Dr. Guangping Gao’s gene therapy advance toward a clinical trial.

Dr. Gao’s study in mice was co-funded by the Canavan Foundation and National Tay Sachs and Allied Diseases (NTSAD). It showed the disease could be slowed as a result of injecting the gene responsible for producing the enzyme into mice bred to have a Canavan-like enzyme deficiency.

ASPA helps move therapy forward

ASPA is actively working to combine the efforts, where possible, of two prior initiatives focused on better understanding how Canavan disease progresses. This step is an important precursor to any Canavan study in humans that might be approved by the FDA; no date for the start of the clinical trial has been established.

Canavan Foundation President Orren Alperstein, who was present at the ASPA Therapeutics gathering in July, says “I am encouraged and excited by the involvement of ASPA, a company experienced in overcoming the regulatory hurdles to clinical trials.”

Update on Programs

Expanding access to information on carrier screening is a major way the Canavan Foundation carries out its mission.

When President Alperstein asked for a show of hands from supporters at last year’s benefit, it was obvious that almost everyone knew someone who would benefit from information about screening for Jewish genetic diseases. Four children were born in the U.S. in the past few years as a result of doctors conducting inadequate carrier screening tests, underlining the need for more education.

Expanded distribution

Over the past year we expanded our distribution of educational materials to Ob-Gyn practices, offering brochures to practices in nearly a dozen cities with significant Jewish populations.

We also started a pilot program to contact primary care practices in New York, prompted by Ob-Gyns who expressed frustration at not being able to screen women who come in already pregnant. Close to 200 primary care practices ordered nearly 5,000 brochures. We have already begun to expand the program across the country, discovering that primary care doctors’ interest in ordering brochures is almost equal to Ob-Gyns.

Synagogues welcome annual deliveries

As in prior years, we contacted synagogues nationwide to restock brochures and posters prior to the High Holy Days. We distributed over 25,000 brochures and nearly 300 posters to more than 600 synagogues. Many recipients said they are grateful for our materials and have come to expect our annual calls.

The Canavan Foundation website remains a vital source of information on Canavan disease and the need for genetic carrier screening, with an average of nearly 200 unique visitors every day. In 2018, we updated the website to improve functionality and ensure that every link and listing is up to date.
Expanded Carrier Screening Recommended

The American College of Obstetricians and Gynecologists’ (ACOG) committee on genetics recently strengthened its recommendation for carrier screening, emphasizing that each Ob-Gyn should establish a standard screening practice. ACOG explained that in today’s multiracial world, as the cost of genetic testing comes down it may make sense to screen everyone before pregnancy, not just targeted ethnic groups such as Ashkenazi Jews. The Canavan Foundation applauds ACOG’s recommendation to expand screening, as we know there are still holes in the system. This year, two Ashkenazi couples had children diagnosed with Canavan disease — one couple had twins — after believing they had been fully screened. Both later found out that testing was done for only a small group of diseases, not the broad panel of rare genetic diseases as they had believed.

The Canavan Foundation continues to advocate for increased awareness and implementation of pre-conception carrier screening. We recommend that couples consult with a genetics professional before being tested to ensure that screening covers the full spectrum of genetic diseases, then request to see the results report to be sure complete testing was carried out.

Canavan Foundation supporters enjoy Carousel

In 2018 the Canavan Foundation theater gala featured Rodgers and Hammerstein’s “Carousel.” Before the show we were treated to a fascinating dialogue between executive producer Joey Parnes and Justin Peck, the wunderkind choreographer. Peck talked about his development from dancer to choreographer, and the difference between choreographing for New York City Ballet and for Broadway. Orren Alperstein offered her heartfelt thanks to our indefatigable benefit chairs, Deedy Goldstick and Pat Hirschhorn, and to David Goldstick for his many years of service on behalf of the foundation.