

AEVR RARE EYE DISEASE BRIEFING

On May 25, AEVR held a Congressional Briefing that focused on rare eye disease entitled: Rare Eye Disease: Research Bringing Treatments to Patients. The briefing featured topics around rare retinal disease and Thyroid Eye Disease (TED). Ben Shaberman, Vice President of Science Communications for the Foundation Fighting Blindness provided insight into research advancements that have been supported by the National Eye Institute (NEI) in rare retinal disease, and AEVR Executive Director, Dan Ignaszewski, shared information about the impacts of TED. In addition, patients who are living with Usher Syndrome and TED shared their experiences living with these diseases.

Classified as diseases that affect less than 200,000 Americans annually, rare diseases represent an area where significant research advancements are being made by NEI-funded research, private foundation-funded research, and the pharmaceutical and biotechnology industries.

Mr. Shaberman began the briefing with an overview of the retina and how various retinal degenerative diseases affect the ability of the retina to detect light and transmit information to the brain for processing. He detailed the individual impacts of rare retinal diseases present differently and how they can lead to progressive vision loss and blindness. He explained how private foundations, the NEI, and the industry play important roles in advancing research to bring treatments to patients.

Mr. Shaberman shared the advancements being made in gene therapy and emphasized the potential of currently funded research at NEI that seeks to use pluripotent stem cells that are derived from a patient's own blood that could be utilized to grow RPE cells that could be transplanted subretinally to provide future treatments to patients.

Patient Advocate Peggy Borst discussed her experience living with Usher Syndrome and Retinitis Pigmentosa and how the progressive vision loss she's experienced since her twenties has impacted her and her family. She shared that she knows there is likely a day when she will progress to full vision loss beyond the tunneling and

spotty vision she currently has, but that she hopes to see advancements that can stave off more vision loss and potentially restore vision in the future.

AEVR Executive Director, Mr. Ignaszewski, shared that in addition to rare retinal disease, TED is also considered a rare eye disease. Mr. Ignaszewski explained how TED is an immune system disorder resulting in hyperthyroidism causing the tissue around the eye to become inflamed resulting in swelling of eyelids, eye bulging, double vision, and additional vision loss. Like some of the advances in the retinal space, researchers have been able to draw on NEI-funded research to develop targeted therapies that help treat TED resulting in new treatments for patients beyond the pre-existing steroids and surgical interventions most used.

Natalie Roberts and LaQuilla Harris, two individuals living with TED shared their experience and how it has significantly impacted their lives, and how important research advancements have been in bringing new treatments to market that have helped improve their outcomes.



Patient Advocates Natalie Roberts and Peggy Borst, Dr. Shaberman, and AEVR Executive Director Dan Ignaszewski

AEVR thanks its sponsoring Member organizations for their support for the briefing:

