My name is Colin Werth and I'm a lifelong resident of Farmville, Virginia located just over an hour west of Richmond. In addition to my work in the IT and marketing field, I am an advocate for the rare disease community. I was diagnosed with Duchenne Muscular Dystrophy at age 3 in 1998.

Duchenne is the most common form of muscular dystrophy. It is a disease caused by mutations in the Dystrophin gene which is the largest gene in the human body. Dystrophin is a vital protein for anchoring muscle cells and providing them with needed stability, so they don't break down easily. Without this, my muscles get damaged and over time lose strength. Duchenne is a progressive disease which gets worse over time and is fatal. About one out of every 5,000 boys has Duchenne, and about 20,000 worldwide are born with it each year. Duchenne mostly affects males and reaches across all races and cultures. It is estimated that there are about 15,000 boys and men, as well as a few women, living with Duchenne today in the United States.

With regards to trial design, it is important to make sure to include the entire patient population living with a certain rare disease. Often clinical trials for diseases like Duchenne only include younger patients who are still ambulatory. This is because many popular tests to show drug efficacy are related to ambulation, such as climbing stairs, running 10 meters, and walking for 6 minutes. However, this leaves out the patients like me who are further along in disease progression and are unable to walk anymore. It is important to make sure that we are not forgotten. As patients living with progressive diseases we are willing to take any treatment we can get to help us. Not being able to walk is one thing, but when you lose arm function practically all your independence is taken away. So, just make sure when you are planning trials to make sure no one is left out.

There is lots of natural history data on Duchenne that could be augmented as a separate trial arm to avoid the need to use placebo control data. Delaying of patient access to gene therapy in the Duchenne population is of concern since once muscle function is lost it cannot be regained.

In addition, it is important we include patients in the clinical trial process more. Including those living with a disease such as Duchenne in the decision-making process is vital to success. People living with rare diseases are the best resource out there since they understand what it's like to live with a condition firsthand. Patients deserve a say in how these treatments should be studied in order to have the best possible trial outcomes and get novel drugs out there faster.

It is important that the FDA and pharmaceutical companies realize that traveling for medical care and to participate in trials is especially difficult for someone with complex needs and requires use of a wheelchair like me. Anyone traveling knows how complicated travel logistics can be. Add on top of that a complex medical condition and travel planning becomes much more daunting. So, if you can, eliminate as much travel as possible and consider multiple locations around the country. Or have labs drawn locally for patients and do follow up visits virtually. If that is not feasible, make sure to have a robust travel assistance program available to patients. Also, consider extended stays in apartments near the trial locations so patients don't have to travel back and forth to their home locations.

Not only is gene therapy going to help Duchenne patients, but it can benefit patients living with many other rare diseases as well such as those discussed on this panel. Gene therapy can also benefit larger populations as well, such as those affected by diseases including Alzheimer's, Parkinson's and even cancer. This is definitely a technology that will become more and more popular in the coming years.