Two years ago, Billy Ellsworth proudly walked across the stage to receive his high school diploma, and today, as he approaches his 21st birthday, he still participates in martial arts classes. These fairly ordinary experiences are, in fact, extraordinary because Billy has Duchenne muscular dystrophy, a rare genetic disease that affects skeletal, heart, and lung muscles. Duchenne is caused by a mutation in the gene that encodes for a protein called dystrophin, which helps muscles function and repair properly. The disease is progressive, and most children with Duchenne use a wheelchair by their early teens. But not Billy.

Diagnosed with Duchenne at age four, when Billy was ten, he had the opportunity to enroll in a clinical trial for a drug called eteplirsen. For Billy and his family, this was the chance of a lifetime. His whole family is grateful that Billy was able to enroll in a clinical trial.

Five years after Billy entered the trial, eteplirsen was approved by the FDA through the accelerated approval pathway. Following approval, Billy was one of the first patients to request coverage of eteplirsen through commercial insurance. He was initially denied — an all-too-common challenge for families seeking access to FDA-approved treatments for Duchenne and other rare diseases. The drug that has helped Billy continue to walk, dress and feed himself, is continually scrutinized by payers, despite approval via a pathway that has been used for three decades. This scrutiny results in disruptions in care or, even worse, leaves behind patients that the pathway was intended to help.

Since the 2016 approval of eteplirsen, four other Duchenne treatments have been approved by the FDA — three approved through the accelerated approval pathway. The accelerated approval pathway is helping to drastically change the course of disease for patients with Duchenne.

Billy’s mom, Terri, feels a deep sense of obligation to all the boys worldwide who are living with Duchenne and their families who face increasing health, financial, and emotional challenges with every passing day. She is a dedicated activist for parents of children with a rare disease and strives to ensure that no one is denied access to the treatments prescribed by their physicians to address their health condition.

While every Duchenne patient is living their own story, the chance at a fuller life that eteplirsen has given Billy and his family is something that the Ellsworths will never take for granted. Though Billy still faces heart complications from Duchenne, he continues to beat his annual walking benchmarks, attend college classes and work on advocacy efforts. Including his trial participation, Billy has now taken eteplirsen for more than ten years. Because of the gift of time eteplirsen has given, Billy is hopeful that continued advances are on the horizon to help knock down those remaining obstacles for people with Duchenne.

“When it really comes down to it the important question that too many keep overlooking is, wouldn’t you want a life preserver of a treatment if this was your child?” – Terri Ellsworth, Billy’s Mom