From: Dirk Johnson <dirklori@gmail.com> **Sent:** Wednesday, October 18, 2023 8:58 PM **To:** Pharmacy Services <rxinfo@dhcfp.nv.gov>
Subject: Rett syndrome and Daybue

Hello! My name is Laura Johnson and I am from Las Vegas, NV. My daughter, Camilla, is 16 and has Rett Syndrome. She is the youngest of four daughters and she is loved dearly. Her first year of life was a blissful time as we were completely unaware that anything was amiss with her. She was a bright-blue eyed, towed headed blond hair beauty with chubby cheeks and she was and is still a joy to our family. At 12 months, I became concerned that she wasn't crawling. So I put her in physical therapy and she quickly began to crawl. With continued therapy, she walked on her own at 22 months. We knew that her tone was low and we had some concerns, so we brought her to a neurologist. Many tests were ordered and everything came back normal. We then saw more specialist who then diagnosed her with Autism. I knew in my gut that there was something more going on with her as I saw her lose the few words she could speak, and she started dropping things and being unable to turn the pages of her books. Her head also measured small. It took another year and a half of seeing many specialists and ordering more genetic tests, before we had our answer.

Camilla had Rett Syndrome. The neurologist was surprised as she didn't have the obvious symptoms at the time. I contacted rettsyndrome.org (Camilla is front and center of the website with her cute pigtails and beautiful smile.) I was referred to Katie's clinic in Oakland, CA and we have received excellent care there ever since. As I learned more about Rett Syndrome, I became aware that Camilla would most likely go through several stages of deterioration, but I was in complete denial as Camilla was doing pretty well at the time. Camilla has since followed each debilitating stage of Rett Syndrome as outlined and it has been very difficult to accept and see my beautiful girl slowly deteriorate. She developed seizures at the age of 5, and we have been trying every possible treatment for the past 11 years without any success. Camilla's seizures have become frequent and severe often lasting over ten minutes and requiring emergency medicine to stop them. Camilla attends a special needs school with a nurse and has seizures there often with frequent calls to 911 and urgent calls to me to get to the school quickly, which is a 40 minute drive away. This has been especially difficult as I am nervous all day that I will get an urgent call from the school and it has also greatly affected my ability to work as a RN. We often take Camilla to Phoenix and California to see specialists because specialty care here in Las Vegas is very poor. Camilla walked well and independently until around age 8 and now she requires two people assisting her to walk. Spasticity has taken over her body, which necessitates botox injections and daily stretching and bracing. Camilla is unable to speak, but she uses a Tobii eye gaze computer to communicate and she can tell you quite well what she is thinking and needs. She is a bright girl trapped in a body that keeps her from doing the simplest of tasks like brushing the hair out of her eyes or pulling up her covers when she gets cold night. What most keeps me hopeful and going is my faith in God and His plan and knowing that someday she will be perfect and healthy. Trying all I can do to keep her healthy and happy with weekly PT, OT and speech therapy gives me peace of mind as well.

Camilla has participated in two drug studies. She participated in the clinical trial for Daybue and continues to take the medication today. We have noticed some small but significant improvements while on the medication. Daybue is the first ever medication approved to treat Rett Syndrome and I feel like every person debilitated by Rett syndrome should have the opportunity to have access to Daybue, as there are many who have benefited from taking the drug.