Children's National Medical Center – Rare Disease Institute

Telehealth Stories from Providers

Dr. Marshall Summar, Division Chief, Genetics and Metabolism

Director, Rare Disease Institute

MSummar@childrensnational.org

Children's National Medical Center has been a leader in using telehealth to care for rare disease families across the country. Below are real stories from providers at Children's National about how telehealth has positively impacted the care and treatment of families living with rare disease.

1. I have had many patients who, despite the pandemic, live in rural areas over 2 hours away from any genetics specialist. With the aid of telemedicine, I was able to see these patients within 1 day of referral and talk parents through the rather scary process of having a child with an abnormal newborn screen. With telemedicine, I was able to see these families quickly and in the comfort of their own home. Many of these families have reverberated to me the ease that telemedicine had brought to them at such a difficult and scary time. Coming into a busy and bustling hospital far from home with your precious newborn child can be intimidating and scary even in normal times. I recently had a patient whose family did not have access to a car and urgently needed to be seen due to an abnormal newborn screen. Between figuring out childcare for their older children, arranging a ride to the clinic, and taking time off from work, it would have been days before this family could be seen in person. With the ease of telemedicine, I was able to see this family that very day with the help of a Spanish interpreter on zoom right there with us. This quick turnaround resulted in a quicker diagnosis. Equally as important, the use of telemedicine to ensure this family's care was prioritized in a timely fashion resulted in a trusting patient-provider relationship.

2. Our patient is a three-year old with Cardiofaciocutaneous syndrome, a rare genetic condition that causes learning problems, low muscle tone, heart defects, feeding issues, and other complications. His mother lives several hours from the hospital, and her husband was away in the military. She had an adult mother at home that needed help and other children to take care of on her own. During the pandemic, she worried about timely access to medical care and COVID, since her son had immune problems. Due to our ability to evaluate her child via telemedicine, we were able to treat an infection (asking our multidisciplinary team and dermatology for guidance), dehydration, help target services, and provide counseling. Her mother attributes our visits to help keep her child out of the hospital and her family safe.

https://wjla.com/news/coronavirus/mom-talks-challenges-of-caring-for-son-with-rare-disease-during-covid-19

3. Another patient is a 13 year old African-American patient who played football until he began to have episodes of undiagnosed rhabdomyolysis, muscle breakdown. He had a prolonged hospitalization prior to COVID. During the pandemic, he developed another episode. Our team had meanwhile developed and published a state of the art protocol for treating rhabdomyolysis. We were able to do several telemedicine visits, get genetic testing for him which showed a potential etiology, and when he had an acute crisis, do a same-day telemedicine visit and get immediate labs from a local laboratory near his house. Based on elevated CK (a measure of muscle breakdown), we advised him to start fluids, and dexamethasone, a drug that we had recently used with success. He was able to complete our regimen, go to a nearby lab to get follow-up testing, and stay out of the hospital all together by following our out-patient plan.

4. A family that lived several hours from the hospital had a mother with four children at home, all with diagnoses of autism or delay. She herself had severe anxiety. She had multiple transportation issues that kept her from accessing medical care. In one telemedicine visit, we were able to evaluate her and her four children. She had previously had a test for a condition called Fragile X, and her results showed she had the condition, but she had never been counseled. We confirmed the diagnosis in her and explained that her children likely had it as well. We were able to arrange testing close to home through her pediatrician, and get the testing she needed and confirm the diagnosis in her children who ranged in age from infants to teenagers. Due to the testing, she has been able to get proper supports in place and treatment for her own condition. In addition, she has a twin sister who also has the diagnosis, and we were able to recommend that her sister and family members get testing. For over 15 years, the diagnosis in this family was missed, and in one telemedicine visit, as she herself put it “One telemedicine visit changed our lives.”

5. I have a sweet family affected by an ultra-rare leukodystrophy who lives on the southern edge of Va. Mom had a baby who was diagnosed with the disease prenatally. She was anxious to bring the baby for an extended clinic visit, as infections/inflammation causes disease progression. We were able to see her remotely, order confirmatory genetic testing sent to the home, and order the baby's feed-and-bundle MRI to be performed when convenient for the family. The family was exceedingly grateful to not have to leave their home with a newborn during COVID19 to drive hours to our institution for a visit that was completely able to be telemed.

6. Patient with POTS/dysautonomia, possible Mast cell activation, Migraine headaches, Hypermobile EDS, AMPS, IBS-mixed, Generalized anxiety with Panic and associated somatization, and Physical deconditioning. She has not walked since November, discontinued school secondary to pain and migraines. Unable to attend outpatient visits - sensitive to light and cannot ambulate. She has been seen inpatient by other services for evaluation. Telemed visit was mostly history provided by parents. Based on history and complexity WES was recommended with mt DNA. Mitochondria was denied so we started with WES only Consent via

telemed - she was unable to participate but called with her verbal consent. Testing kit was sent to the home

7. Couple of years ago had a case of a thirtysomething-year-old woman with very advanced colon cancer from rural southeast MS. Was getting intensive chemo and in last few months of life. She had 3 sons. Astute doc wanted her to see genetics but she couldn't travel so we saw her by TH in Hburg, tested her quickly and cascaded her sons (age 5-15), 2 of whom were positive for her APC mutation- got them in for surveillance and care before she passed. All done remotely and with TH visits. I think that made a huge difference for her- knowing that her sons were caught early.