

SSHA PKU Backgrounder

Celine and Braden

Celine Beaulieu and her three-and-a-half-year-old son Braden Tonkin live in Thunder Bay. Braden was born with a rare genetic disorder called PKU; but thanks to ONE® (Ontario Network for e-Health) Network, a high-speed, high-capacity network provided by Smart Systems for Health Agency (SSHA), Braden and Celine have easy access to consultation, education and support from experts at the Hospital for Sick Children in Toronto. This is accomplished through a video-conferencing service provided by the Ontario Telemedicine Network (OTN), which uses ONE Network.

What is PKU?

PKU, or Phenylketonuria, is a recessive genetic disorder characterized by a deficiency of an enzyme in the liver that breaks down the protein in the food we eat. Protein that is not broken down remains in the bloodstream, where it can do serious damage. Left untreated, this condition can cause problems with brain development, leading to progressive mental retardation and seizures.

The incidence of PKU is about one in 15,000 births and there is only one other known case of PKU in Thunder Bay, a city of 100,000. Babies are tested for PKU as part of a standard battery of tests – referred to as metabolic screening – given to every newborn in Ontario within the first 24 to 28 hours after birth.

Although there is no cure, PKU is fortunately one of the few genetic diseases that can be controlled by diet; however, early detection is critical because any damage done is irreversible. With a life-long diet low in protein, an affected newborn can grow up with normal brain development. Supplementary formulas are used for infant PKU patients to provide the amino acids and other necessary nutrients that would otherwise be lacking in a protein-free diet. Supplements continue in other forms as the child grows up.

How OTN and ONE Network Help

Because PKU is so rare, the skills required to treat this disorder are not readily available across Ontario, so Braden is a patient at the Hospital for Sick Children in Toronto. There, a physician, nurse and dietitian provide the consultation, education and support that Braden and Celine need to keep his condition in check. Braden is kept on a protein-free diet supplemented by a protein-altered formula that he drinks three times a day.

Rather than having to make the long trek to Toronto though, Celine and Braden have already made 30 'virtual visits' to see their health care providers, using OTN video-conferencing from Thunder Bay Regional Health Sciences Centre. Through the video-conferences, which took place every two weeks initially and now about three to four times per year, all parties can review Braden's progress and discuss any changes to his formula that may be required based on recent blood test results. The video link allows the clinicians to see how Braden is doing.

"It seems simple, but there's been quite a bit to learn, and managing his diet properly is very complicated, especially when he was younger and we had to keep changing his formula because he was growing so quickly," says Celine. "The video-conferences not only save us from having to travel to Toronto, they give everyone a good opportunity to keep up with everything that has occurred and allow me to get additional tips on how to manage Braden's diet."