NEW HOPE FOR BABIES WITH
KRABBE’S DISEASE

by Eve Krakow

A recent study shows that a cord-blood transplant performed soon after birth can not only save the child’s life, but also significantly limit the progression of the disease.

Krabbe’s disease is a very rare neurological disorder resulting from an enzyme deficiency. Symptoms appear in the first few months of life, when the infants do not progress as they should. Affected infants deteriorate rapidly and usually die before they reach the age of 18 months.

In the study, 11 infants were diagnosed prenatally or shortly after birth and underwent the transplantation of umbilical-cord blood within the first few weeks of life. These were cases where the parents had already had a child with Krabbe’s disease, so they knew they were carriers. Fourteen more infants who were diagnosed between four and nine months of age, once symptoms appeared, also received transplants.

The 11 children who received the transplants as newborns (before the appearance of symptoms) have all survived and made phenomenal progress. “We changed the nature of the disease,” said Dr. Martin Champagne, medical director of the hematopoietic stem cell transplant program at the Sainte-Justine University Hospital Center in Montreal. “After the transplantation, we looked at the children’s ability to acquire developmental landmarks. Many still had mild to severe impairments in gross motor function, but they had age-appropriate cognitive function and language skills.”

Champagne said there had been some previous attempts with bone marrow transplants, but that it usually takes too long to find a match: 70% of children lack a suitable donor in their family. As well, donors who are family members are likely to be carriers of the disease, and researchers fear that stem cells from carriers would not be as effective in regenerating the missing enzyme. Cord blood, however, is readily available, and donors do not have to be related.

Micki Gartzke’s daughter died of Krabbe’s disease at two years of age. Gartzke is now the Director of Education and Awareness for Hunter’s Hope, a U.S. foundation that seeks to increase public awareness about Krabbe’s disease and improve early detection and treatment.

“One of the children in this study was born about 10 days before my daughter,” she said. “He came from a case index family, so he was diagnosed early and received the treatment. He goes to school now. My daughter has been buried in her grave for quite some time. So what this study means is a chance for children to have a life, a future.”

Gartzke has met most of the 11 children who received the transplants as newborns. “Some can run around, others need assistance with mobility and speech, but they’re all there, they’re all smiling, and they all know what’s going on.”

Unfortunately, of the children who received the transplants later because of the late diagnosis, some have died, while others are living with extreme difficulties. This is why Hunter’s Hope is working with researchers and health officials to have Krabbe’s disease included in universal newborn screening.

Dr. Champagne says the study could lead to treatment for other diseases, too. “The role of stem-cell transplants in many genetic diseases is not well established. This can be seen as a model to consider intervention very early on, before the onset of symptoms.”