



# Impact

Your donations at work



“Despite the temptation to attribute the symptoms to normal childhood allergies, Joseph’s mother knew in her heart that it was more.”

Joseph received his bone marrow transplant on January 5. Twenty-eight days after his transplant, Joseph was released to the Ronald McDonald House to await a therapeutic boost of stem cells. “This was specifically designed for osteopetrosis patients,” said Dr. Orchard. “They get a ‘boost’ of donor stem cells on day 42 to help with engraftment.”

Kim said the most difficult part of the experience was watching Joseph laboring to breathe with the post-transplant mucositis, which lasted about two weeks, and the high fever at five weeks after the transplant. Thankfully, however, the transplant seems to have preserved some of Joseph’s eyesight, which was a big concern of the physicians and his parents.

In April, Joseph was well enough for the family to go home to New York. They will return to the University of Minnesota for regular check-ups for several years. Now passionate about the importance of research, the family donated some of Joseph’s blood and a skin biopsy for future childhood cancer research.

As they continue on this journey, Pat’s dreams for his son are simple. “I hope he gets to live a happy, healthy, normal life,” he said. And what about that froggy blanket? “That blanket’s not going anywhere until Joseph’s 30,” he laughed.



## Joseph’s story

Osteopetrosis is a big word for such a little guy, and yet Joseph, now just a little more than 1 year old, has had that word attached to him since he was 8 months old. Osteopetrosis is a rare inherited disorder in which the bones become dense and harden because the body doesn’t shed old bone tissue.

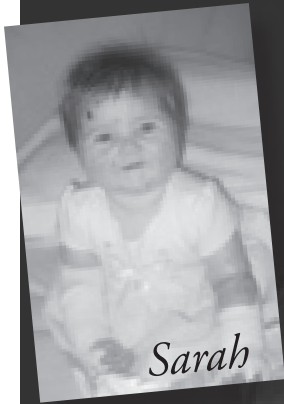
In severe cases, patients can experience stunted growth and deformity, in addition to increased fractures and bone infections. Sometimes osteopetrosis can cause an enlarged spleen, kidney problems, blindness, anemia and bone marrow failure. Without a bone marrow transplant to stop the progression of osteopetrosis, most children will not live to the age of 10.

For Joseph, gastrointestinal issues were the first sign that something was wrong, according to his mother Kim. Despite the temptation to attribute the symptoms to normal childhood allergies, she knew in her heart that it was more. Treatment after treatment failed to relieve Joseph’s symptoms. It was a neurologist in their hometown of Rochester, NY, who finally made the official diagnosis, which devastated the family.

“Our reaction was shock,” said Joseph’s father Pat. “It was very emotional. We immediately got on the Internet and began researching osteopetrosis, its outcome and its treatment. We kept hoping the diagnosis was wrong.”

Joseph, along with his favorite froggy blanket, was admitted to the University of Minnesota Amplatz Children’s Hospital on December 26, 2009, and began receiving inpatient chemotherapy, under the care of Dr. Paul Orchard. “Joseph was a trooper through the whole thing,” wrote his parents on their CaringBridge website. “He still remains a very happy baby.”

## Mooreland Family



### Sarah's story

By **Lonni Mooreland**

Imagine the anticipation and hope an expectant couple has for the future when they are about to welcome a new baby into their family. When we welcomed our third child, Sarah Rose, into our family, all the hope we had faded quickly when we were told what we could expect for her life — a life of constant pain, constantly wearing bandages, painful daily bandage

A disease where Collagen type VII proteins, the anchors for our skin, aren't present, so the slightest friction or rubbing can cause blisters or even the skin to tear off.

We were devastated, and even more, by that fact that there wasn't a cure and there wasn't any hope of a cure. I had days where I wondered how was I as the mother to this sweet little girl going to function when my baby's normal baby movements or things like bathing

We were really excited for this hope of a better life for our Sarah. We moved our family temporarily to Minnesota for the procedure to be done. After nine days of chemotherapy, Sarah was ready for the bone marrow transplant. The morning of her transplant, Sarah Rose died. It was the worst thing a parent can experience. We had so much hope for her, then lost her right at the peak of it all.

Since Sarah's death we have made a lot of life changes. Our family has moved from California to Minnesota in hopes of helping the doctors here. When we initially met with Dr. Wagner, we felt very strongly that he had a gift and knew he was offering something no other doctor had offered us — hope.

Why would we return to the place where our sweet little 9-month-old Sarah died? Why do we support these doctors after the outcome that we had? We feel incredibly passionate about what they offer. They have experience; they have positive results, passion, and incredible drive and desire to help children rare genetic diseases.

“I found hope one day online when I heard of a trial being done at the University of Minnesota.”

— Lonni Mooreland

changes that are time-consuming and painful for her. She would probably lose the ability to walk; her hands would eventually web together; eating could cause the lining of her mouth to tear away and be potentially lethal if blisters formed in her esophagus; and she would most likely die in her teens of an aggressive skin cancer.

This is what children born with Epidermolysis Bullosa can expect.

or the loving touch from one of her siblings too young to understand could cause a painful blister or the loss of a large amount of her skin.

I found hope one day online when I heard of a trial being done at the University of Minnesota. Dr. John Wagner was doing trials through Bone Marrow Transplant. He found that through donor-matched marrow, new healthy skin was beginning to regenerate.



## The facts about Epidermolysis Bullosa

Epidermolysis Bullosa (EB) is a group of genetic skin conditions characterized by fragile skin and easy blistering. The skin is so fragile in people with EB that even minor rubbing may cause blistering. At times, the person with EB may not be aware of rubbing or injuring the skin even though blisters develop.

In some forms, the blisters heal with scarring which can result in permanent change to the skin, for example fingers may fuse, reducing movement. In severe EB, blisters are not confined to the outer skin. They may develop inside the body, in such places as the linings of the mouth and esophagus.

There is currently no cure for EB. However, researchers at the University of Minnesota were the first to use a Blood and Marrow Transplant (BMT) to treat this disease. Led by Dr. John Wagner and Dr. Jakub Tolar, doctors in Minnesota were the first to demonstrate that Collagen type VII proteins can be given to patients via bone marrow transplant. The early results in patients show an improvement in their skin and the presence of Collagen type VII in their epidermis. Prior to BMT, the only treatment options being used were external — trying to manage skin care with topical ointments, bandages and other precautions.

At this point, the University of Minnesota is the first and only place in the world performing BMT on EB patients, and the number of EB patients and families coming to Minnesota for this groundbreaking treatment continues to grow.



## From grief to action

After experiencing the devastating loss of their daughter, Sarah, the Moorelands felt compelled to turn their grief into something positive. They founded PUCK, Pioneering Unique Cures for Kids (the family chose the name PUCK as a nod to Minnesota's hockey culture). PUCK is a dedicated fundraising effort to raise awareness about rare genetic diseases and to provide a voice for families facing similar diagnoses.

### PUCK's Mission

- To raise awareness of rare, genetic diseases
- To create a supportive resource for families and children with rare diseases
- To raise money for research on rare diseases

While Sarah's outcome was not as they had hoped, the Moorelands are dedicated to supporting the doctors and researchers at the University of Minnesota for their diligence in finding a cure for these debilitating diseases.



**To learn more, visit [puckfund.org](http://puckfund.org) or find it on Facebook by searching "PUCK".**



Jakub Tolar, M.D.

## The future of treating genetic diseases

Doctors at the University of Minnesota are world experts at treating different types of genetic diseases, like osteopetrosis and EB.

In most of these conditions, bone marrow transplantation can often arrest the course of the disease, but it is not without risk. "Ninety percent of my work with these children is fixing the damage I've done with chemotherapy and a BMT," said Dr. Jakub Tolar, a leader in the treatment of rare, genetic diseases.

Physicians want better treatments than the ones currently available. Dr. Tolar and his team are pioneering the use of a patient's own skin cells to create immature stem cells that could, in theory, be transplanted back into the body. The goal is to provide better outcomes with fewer risks and improve the quality of life for patients facing these diseases.

## Keeping up with our researchers



John Wagner, M.D.

### \$8.6 million grant for cell research

The University of Minnesota was awarded the grant to fund the development of cell-based therapies from laboratory research to clinical trials.

The University received a five-year funding contract totaling \$8.6 million from the National Heart, Lung and Blood Institute. The program's mission is to speed the research and development of new treatments by providing assistance to researchers nationwide in an effort to accelerate the U.S. Food and Drug Administration study approval process.

**John Wagner, M.D.**, Children's Cancer Research Fund's chief medical advisor, said the focus will be on two broad subjects: the study of immune cell populations, like natural killer cells that fight off diseases such as leukemia; and the advancement of stem cells.

### Improving therapies for brain tumors

Malignant brain tumors are very difficult to treat and are almost always fatal. New drugs have been developed that specifically target proteins that lead to brain tumor growth. These drugs, called "molecularly-targeted" therapy, have been extensively tested in clinical trials with great hope.

Past treatments have attempted to target brain tumor growth, but have yet to produce positive outcomes, even when these new drugs were used in conjunction with surgical removal of the tumor. A central question that has been essentially ignored, is; "Why are these new drugs failing, when they target the tumor biology directly?" Researchers at the University of Minnesota are attempting to answer that important question, which will undoubtedly lead to improved treatments for malignant brain tumors.

The purpose of this research project is to explore why drug therapies do not treat cancer cells that are located far away from the tumor site, and examine mechanisms that will help deliver more of the new molecularly-targeted drugs to invasive tumor cells. This research has resulted in more than \$1.8 million in new grants based on promising findings made from the Children's Cancer Research Fund's original \$100,000 investment given in 2009.



## Connect families

Lifesaving treatment is not always in your backyard. Often, families travel long distances to receive the best care available, and stay for many months at a time. As a result, parents may split time between home and the hospital, incurring significant travel costs. Care Flights connect families while a child is receiving treatment for cancer or other rare illnesses.

As a Delta Air Lines SkyWish Charity partner, Children's Cancer Research Fund relies on individuals like you to donate your extra miles in support of families in need.

Your miles can make a world of difference. **To donate your extra Delta miles, visit [ChildrensCancer.org/Delta](http://ChildrensCancer.org/Delta).**

