Sarah's Story

Sarah is a smart 6th grader who loves arts and crafts and spending time on her computer. She was born on September 27, 1998, after a normal pregnancy. Life has been nothing short of difficult for Sarah, and she has experienced more pain in her 12 years than most of us ever will our entire lives. Immediately after birth, doctors knew something wasn't right when the tissue lining her mouth came out after it was suctioned. Shortly after that, they noticed that all the handling caused her skin to slough off her body. She was immediately transferred to a hospital with a high-level NICU where she was kept in isolation because of the risk of infection. A few days later, a skin biopsy and genetic tests revealed Sarah was born with Epidermolysis Bullosa (EB), in the Recessive Dystrophic form (RDEB), a life-threatening skin blistering disorder. RDEB affects about one in every one million live births. It is impartial to race, ethnicity or sex. There is no cure.

In EB the skin is missing the critical proteins responsible for the glue, or anchoring fibrils, that adhere the skin to the body. Without these anchoring fibrils the skin blisters or sloughs off from the slightest friction. Scarring leads to major disfigurements, including webbing of fingers and toes and severe contractures that greatly impair mobility. Blisters in the mouth, throat and esophagus lead to eating difficulties. The accumulated scar tissue in the esophagus leads to strictures (narrowing) requiring surgery to stretch the esophagus so the child can swallow again. Nutritional deficiencies lead to a number of complications, such as anemia and poor wound healing. Metastatic squamous cell cancer of the skin is the most common cause of death and is often developed between age 15 and 35. This skin cancer can occur anywhere on the skin and is not due to sun exposure, rather to the constant regeneration of cells required to heal the chronic wounds. Sepsis (infection) is another major cause of death.

From the very beginning, one of the biggest challenges of living with EB was finding doctors experienced in treating this rare disorder. Over the years, her ever-growing team of specialists has extended into Cincinnati, where we meet with highly skilled specialists who have extensive experience with the multiple aspects of EB. We travel to the EB Center at Cincinnati Children's Hospital Medical Center several times a year for treatments and evaluations.

For the first 9 years of her life, Sarah fared very well compared to other children afflicted with this terrible illness. However, a little over two years ago her condition started to decline and she experienced widespread blistering and drastic weight loss. To date, Sarah has had seven esophageal dilatations, a blood transfusion and iron infusion to treat her anemia and most recently surgery for a feeding tube to help her get the 2,200 calories per day that her body requires. She has been hospitalized for dehydration and sepsis. EB does not just scar Sarah's skin. Emotional scars, such as anxiety, depression and self-esteem issues are no strangers to her.

Daily treatments for Sarah involve painful baths for which she is pre-medicated with strong medicines, like Methadone, Dilaudid and Ativan. These medicines just about take the edge off the pain associated with soaking in water containing the vinegar or bleach recommended for infection control. The entire process of removing bandages, bathing and re-bandaging takes approximately 5 hours. At school, she is accompanied by a nurse who monitors her vitals and tends to her medical needs.

It has been a difficult 12 years and unless a large stride is made in the treatment of EB, it will continue to be so. We are fortunate to have the love and support of many people who genuinely care about us. She tops many prayer lists and we are very grateful for her prayer warriors. It brings me great joy to know that we can share laughs together and I see her in a new light when we are able to discuss important topics in a mature way. While being Sarah's mother comes with much heartache and uncertainties, I am blessed that this experience has allowed me to fully appreciate the many things that so many people take for granted…a walk on the beach, a summertime outing or the simple luxury of traveling light.