

DOCTORS FIND PROMISING TREATMENT FOR EB

June 3rd, 2008

New York - We are delighted that investigators at the University of Minnesota, in collaboration with renowned EB researchers Drs. Angela Christiano and Jouni Uitto, are reporting promising progress toward treatment of Recessive Dystrophic Epidermolysis Bullosa (RDEB). We send our best wishes to the Liao family and the dedicated team of physicians and scientists who are working so hard to find a cure. We are proud to have been a part of the pre-clinical research leading up to this study, and will be sharing results with the EB community of physicians, families, and loyal supporters world-wide.

The investigation at the University of Minnesota is the culmination of several years of early pre-clinical work supported in part by **DebRA of America** that occurred in other EB research labs around the country. DebRA supported the development of a mouse model that replicates the features of human Recessive Dystrophic EB, including severe skin blistering at birth. This mouse model was created by investigators in the laboratory of Dr. Jouni Uitto at Thomas Jefferson University in Philadelphia, specifically for the purpose of pre-clinical trials of novel therapies, such as this one. Having introduced the concept of bone marrow transplantation as a treatment for EB, these mice were first tested in Christiano's lab at Columbia University, and later by Drs. Jakub Tolar and Bruce Blazar at the University of Minnesota, to demonstrate the feasibility of the bone marrow transfer for this type of EB. Once the mouse model showed success by this approach, Institutional approval for the first clinical trial for bone marrow transfer for EB was quickly obtained and performed at UMN under the direction of Dr. John Wagner.

With research support from **DebRA of America**, Drs. Christiano and Uitto have been working together on dystrophic EB for more than 15 years, since she was a post-doctoral fellow in his lab. Their accomplishments together include cloning of both human and mouse type VII collagen gene, and first demonstration of type VII collagen gene mutations in dystrophic forms of EB, and the development of DNA-based diagnosis for the disorder. Her laboratory at Columbia University performed the first genetic studies on the Liao family and determined the exact molecular basis for their individual case of RDEB. She and other EB researchers have studied several different gene-based therapy approaches for EB with **DebRA of America** support, before Christiano turned her attention to circulating stem cell transplantation. Additional research is already underway in which Christiano and Uitto will extend these studies in to junctional forms of EB, once again using a pre-clinical mouse model supported by **DebRA of America** research funds. Both Drs. Christiano and Uitto have been involved with the Scientific Advisory Board of **DebRA of America** for more than 15 years.

DebRA is the only national organization for the EB community that funds clinical support, educational programs and EB research for almost thirty years.

EB children are referred to as "Butterfly Children" due to the fact that the fragility of their skin is comparable to the wings of a butterfly.

DebRA of America embraces with optimism and caution this early investigation toward a cure. Mary Sprague, Executive Director, stated that “**DebRA of America** is deeply proud of its longstanding support of fundamental basic research in EB, and the way in which the support has facilitated this breakthrough. This is major achievement towards treatment, and potentially a cure, for this currently intractable disorder.”

###