Scalp Erosion

Extract Letter from member Chrissy Lees-Jones to the Doctor

We receive "The Educator" newsletter (article below) and read your article in it about skin erosion. It is very interesting about your new treatment for transplantation.

Our daughter Tess was born with ED (Haywells syndrome) it's one of the rarer syndromes as you probably know.

Tess was affected by skin erosion on her head from 10 days old. It became very infected and eroded around the whole of her scalp.

There seems to not be much awareness in Britain about skin erosion, so after visiting 8 hospitals and over 70 dermatologists all of whom had very opposing ideas as to how to treat Tess' desperately affected scalp, I made the decision to treat it and bathe it twice a day in salt water morning and night. Over the course of 5 years it healed and is now strong and able to be open to the sun.

It was a very difficult time in our lives and Tess had to be brave, but I just wanted to let you know a bit about our story as I am concerned that many parents are not aware of the healing properties of salt water. It has also meant that the skin has come back stronger than if I had used steroid creams and also means that it looks so healthy.
This may be a possibility for other suffers of skin erosion.
My husband has the same syndrome, however he didn't have the skin erosion. He has given tissue samples so the professor at St Thomas' hospital in London can keep researching into the mutant P63 gene.

I wish you and your team and patients all the best for the future.

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Taking AEC and EEC Research to the Next Level

By Maranke Koster, Ph.D.

(Editor's Note: AEC is ankyloblepharon-ectodermal dysplasia-clefting syndrome, which was formerly known as Hay Wells syndrome or Rapp Hodgkin syndrome. EEC stands for ectrodactyly-ectodermal dysplasia-clefting syndrome.)

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Overview of Our Research

AEC and EEC are ectodermal dysplasias caused by mutations in the p63 gene. Although only AEC has historically been linked to the presence of skin erosions, data from the NFED's Ectodermal Dysplasias International Registry indicate that over 25% of EEC patients exhibited skin erosions at some point during their lives. Our overall goal is to develop a strategy for the treatment of skin erosions in AEC and EEC patients by generating healthy replacement skin that will not be immunologically rejected by the patients. To accomplish this goal, we are developing a strategy that utilizes the patients' own skin cells.

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These cells will be temporarily transformed into immortal stem cells, termed induced pluripotent stem cells (iPSC). After correcting the patients' p63 mutation using modern gene correction tools, the iPSC will be directed to develop into healthy skin cells. These cells are then used to generate full-thickness replacement skin for transplantation.

Since these cells originate from the patient receiving the transplant, immunological rejection of the skin grafts is unlikely. Although it will take some time to make this treatment option a reality, we have already made significant progress. This progress would have not been possible without the support of the NFED and willingness of its members to support our research by providing skin biopsies.

In addition, we plan to use the iPSC-derived skin cells (either with or without p63 mutation) to answer mechanistic questions about p63. Examples of questions we aim to answer are: Why do certain mutations in p63 cause AEC whereas others cause EEC? Why do some EEC mutations cause skin erosions and others do not? Are skin erosions in AEC patients similar to those that occur in a subset of EEC patients?

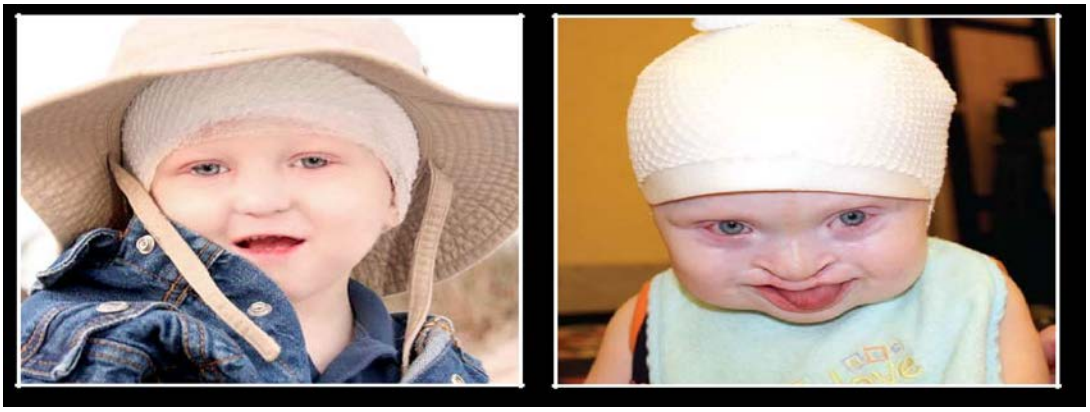
A better and more precise understanding of the role of mutant p63 proteins in both AEC and EEC may lead to the development of therapeutic options that are less invasive than the proposed cell therapy.

Experiments addressing both research objectives are performed simultaneously thereby increasing the possibility for developing novel therapies for skin erosions in AEC and EEC patients. Further, very similar approaches can be used to develop therapies for corneal lesions.

Upcoming Research Opportunities

Pending regulatory approval, we plan to obtain skin biopsies from patients affected by AEC or EEC (with or without a history of skin erosions) for our research studies. The overall objective of our research is to develop new treatment strategies for skin abnormalities that occur in patients with p63-related ectodermal dysplasias, such as AEC and EEC. We also want to better understand the differences and similarities between AEC and EEC.

For more information about the research, please contact Dr. Maranke Koster at Maranke.Koster@ucdenver. edu or 303-724-1640. For more information on participating in this research, please contact NFED Executive Director Mary Fete (mary@nfed.org.



AEC: James Burell is affected by AEC syndrome and experienced significant skin erosion on his scalp as a toddler.

EEC: Ethan Kranig is affected by EEC syndrome and had skin erosion on his scalp as a baby.

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