XLHED Treatment – From Set Back to Renewed Help

(reproduced with the kind permission of the NFED – Summer 2017)

Timmy, Drew and Liam are three of the babies who participated in the Newborn XLHED Clinical Trial from 2013-2015.

I am excited to share with you the latest news in our journey to develop a treatment for x-linked hypohidrotic ectodermal dysplasia (XLHED). EspeRare, a not-for-profit drug developer based in Switzerland, is picking up where Edimer Pharmaceuticals left off! EspeRare is exploring the possibility of re-launching the development of ER-004 (formerly known as EDI200) as a treatment for babies affected by XLHED. They are looking to start a phase II clinical study where ER-004 would be administered to babies "in utero".

This is incredible news for all of us! However, this relaunch is still at an early stage. EspeRare is conducting a study to ensure that its plan is feasible and suited to our community's needs. At this point, the main hurdles to overcome are developing both a regulatory and funding plan to take this project forward.
We are working closely with EspeRare during this key phase and doing all we can to help make it a success. We are thrilled with this development but understand it's early in the process!

What is ER004?

ER004 is a synthetic form of Ectodysplasin A (EDA), the protein missing in XLHED during development. Through the years you have heard us call it APO200 when Apoxis was developing it, then EDI200 when Edimer was involved and now it's called ER-004. It is the first and only therapy developed for XLHED. This is cutting edge research and a highly innovative therapy.

Edimer's first study successfully demonstrated the therapy to be well tolerated. In their follow-up study, the XLHED Newborn Clinical Trial, the treatment did not correct the XLHED symptoms in the newborn boys who received it, as we had hoped. At that point, Edimer stopped the development and closed its operations. All of us in the ectodermal dysplasias community were disheartened by the setback. But not for long.

Supporting a normal lifestyle

Last December, we shared the promising news that Prof. Holm Schneider in Germany had treated three babies prenatally with ER-004. It's still early to fully understand how well the treatment has worked. However, to date, the babies are able to sweat normally and have not had any respiratory-linked hospitalizations. They may have other positive outcomes yet to be assessed. Isn't that amazing! It looks like giving the replacement protein during pregnancy versus to newborn appears to markedly improve its effectiveness.

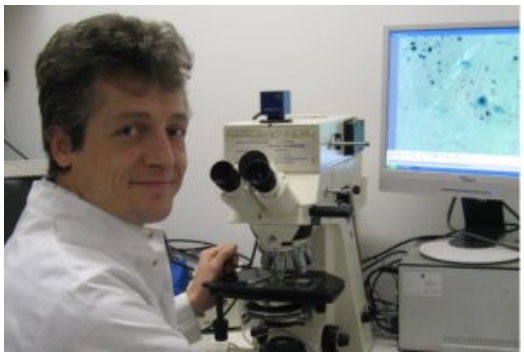
Maarten and Linus are sweating normally after receiving ER-004 in utero.

EspeRare's Plan

EspeRare plans to build on Prof. Schneider's early success and administer ER-004 in utero to boys affected by XLHED in a clinical trial to assess if this therapy works. First, EspeRare has started a dialogue with the regulatory authorities to understand if they will allow this ground-breaking clinical trial. If the regulatory path and funding are secured, then the trial could start in 2018 in the European Union (EU) with Prof. Schneider as principal investigator. They plan to expand the study to the United States after interim positive results.

The primary objective of this trial is to assess ER-004's ability to restore the babies' ability to sweat. ER-004 would be administered through two injections a few weeks apart during the third trimester of pregnancy. This means that this could be a single course of treatment that could have life-long effects – if successful!

But, First Things First - The Players:



While EspeRare is new to the world of XLHED research, they are talking and working with people and organizations like the NFED. EspeRare has been working with Neil Kirby, former Edimer CEO, on transitioning this project. They have identified Prof. Schneider, who was a part of the XLHED Newborn Clinical Trial and treated the three babies, to be the principal investigator. Also on board is Pascal Schneider, who was one of the researchers who engineered ER-004 (called APO200 at the time) in the lab with NFED funding!

Prof. Holm Schneider will be a principal investigator.

EspeRare needs to understand if the XLHED community is interested in such a therapy. This is where you come in. Soon, we will ask families affected by XLHED to complete a survey for EspeRare. Watch for those details to be coming very soon! We will need you to act quickly.

We hope you share our renewed hope in creating a treatment for our families with XLHED. We still have a long way to go and a lot of work to do. We are committed to advancing this important work and hope you are, too.

Onward!

Together, we can make this happen!

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