

Clinical Trial for Prenatal treatment of boys affected by X-linked Hypohidrotic Ectodermal Dysplasia



Information for Families

The EspeRare Foundation and their partner, Pierre Fabre Medicament, are developing a drug for X-linked hypohidrotic ectodermal dysplasia (XLHED). The drug is called ER004 and this is the same drug that was previously developed by Edimer and was known at the time as EDI200.

Prof Schneider, a German paediatrician actively involved in ER004's clinical development, showed that while ER004 does not work in newborn babies, ER004 shows promise when given before birth. In particular, sweat function was shown to be improved in six XLHED-affected boys treated before birth <https://www.nejm.org/doi/full/10.1056/NEJMoa1714322>. It is now EspeRare and Pierre Fabre's intention to confirm these results in a robust clinical trial.

Who may participate?

Knowing that you are a carrier of XLHED and that you are pregnant with a boy is sufficient information to consider taking part in the study. Women who suspect they are carriers of XLHED should have a genetic test to determine their XLHED carrier status. For information regarding genetic testing, please speak with your family doctor or clinical geneticist.

Confirmation of the XLHED status of your unborn son will determine whether you can enter the study or not. Women pregnant with boys in the next 24 months may be eligible to participate in the study.

We also need participants who are XLHED-affected male blood relatives of the pregnant women to form a "comparator group". This group will not be treated with ER004. Comparison of data collected from the treated babies with data from the untreated subjects will allow us to measure if ER004 has worked.

What will happen in the study?

Women pregnant with XLHED-affected male fetuses taking part in the study will:

- Receive 3 doses of the ER004 drug, before birth through a series of 3 injections into the amniotic fluid: At pregnancy weeks 25, 28-29 and 31-32

When the child is born, women will:

- Be required to attend a number of visits at the treatment site, until their child is 5 years old, to evaluate long-term safety and efficacy effects of the given drug.

Male relative of the pregnant woman, XLHED-affected taking part in the study will:

- Not be treated and be required to attend a single visit at the Cardiff investigational site.

For More Information

Pregnant women must be enrolled in the clinical trial during the second trimester, between pregnancy weeks 19 and 24. Therefore, it is important that families consider this information as early as possible. For more information about the clinical trial visit: www.clinicaltrials.gov (search for clinical trial EDELIFE) or contact the following:



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