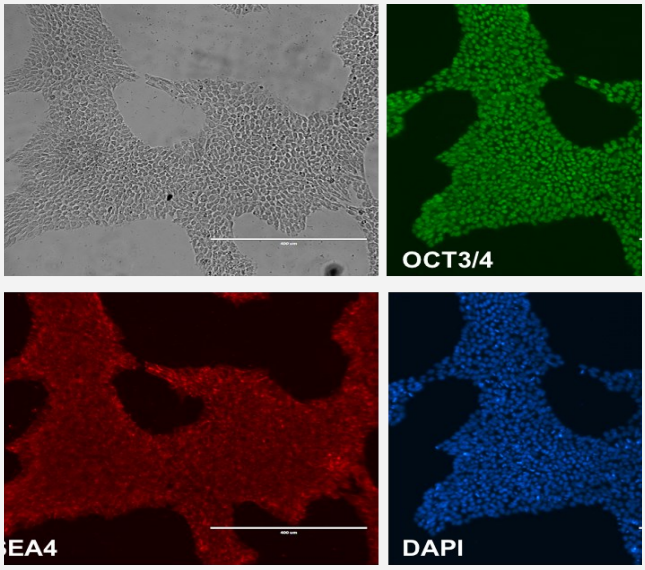
LCHAD Retinopathy Research Update

For the Scully Peterson Family Fund for LCHAD Retinopathy Research

OHSU

We are continuing to make great progress toward creating a cell model of LCHAD-deficient retina. The initial step to create an LCHAD-deficient stem cell was recently completed. Using patient skin cells that had the common LCHAD genetic mutation, we reprogrammed the cells to become stem cells. These new stem cells have LCHAD deficiency like the skin cells from which they were derived.

In order to verify the new cells have LCHAD deficiency and are stem cells, we genotyped the new cells and stained some of the cells for stem cell-specific markers. The cells did have the common LCHAD genetic mutation indicating they were in-fact LCHAD-deficient. The green staining is for a stem cell factor OCT3/4, and the red staining is for a stem cell factor SSEA4. The blue staining indicates the nucleus of the cells. Our results show that we now have LCHAD-deficient stem cells and can begin the process of programming the LCHAD-deficient stem cells to become retinal cells.



Expression of stem cell markers in LCHAD-deficient cells. *LCHAD-deficient stem cells show positive staining of OCT3/4 and SSEA4, markers that are only seen in stem cells. DAPI is a stain that binds to DNA, allowing us to mark the nucleus.*