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RTD mouse investigation by Jackson Laboratory (JAX).

The Cure RTD Foundation has awarded a $34,596 collaborative research grant to Jackson Laboratories (JAX - www.jax.org) for RTD Type 2 mice studies.

Using gene editing technology, mice carrying genetic mutations that are the human equivalent to the SLC52A2 gene mutations causing RTD type 2 ("RTD Mice") have been successfully created. Early versions of these RTD mice died before birth. However, successful RTD mice could be born alive for study by providing mothers with high-dose riboflavin (FMN) during pregnancy and after trials of different SLC52A2 gene mutation combinations and mice breeds.

These RTD mice have been created primarily to demonstrate the safety and effectiveness of RTD gene therapy, which is a necessary step before human clinical trials can begin. The gene therapy project is being done in collaboration with Dr. Steven Gray, PhD, at the University of Texas (UT) Southwestern in Texas. These RTD mice will also be used to investigate the mechanisms causing RTD and develop other novel therapeutic strategies for RTD.

Before beginning gene therapy trials, it is necessary to understand these RTD Mice symptoms and survival rates. This project tracked groups of RTD mice receiving no treatment or riboflavin treatment similar to what a person with RTD would receive. At the end of the study, riboflavin levels (RF, FMN, and FAD) in the mice's blood and brain were measured.

Optic atrophy causing vision loss is one of the most common early symptoms in people with RTD Type 2. Given this information, 18 RTD mice also had their eyes dissected to look for signs of eye disease.

We are pleased to announce that this project has already been completed. The results of this study were encouraging and indicated gene therapy trials on these RTD mice should proceed.

About Jackson Laboratories

Founded in 1929, The Jackson Laboratory (JAX) is a nonprofit biomedical research institution that seeks to understand the human organism — in all its biological and genomic complexity — by using the mouse as a model. JAX is the world's source for more than 8,000 strains of genetically defined mice and is the home of the Mouse Genome Informatics database. Their research breakthroughs have helped form the foundation of modern medicine. Organ and bone marrow transplants, stem cell therapies, and in vitro fertilization all have a foundation in JAX research.