

CHOP Research Institute Summer Scholars Program



Marni Falk, M.D.
Human Genetics

Translational Research: Research focused on bringing concepts in laboratory research directly into medical practice (may or may not involve human subjects)

Genetic Causes, Metabolic Consequences, and Novel Therapies for Mitochondrial Respiratory Chain Diseases

My research lab investigates the genetic causes, metabolic consequences, and novel therapies for mitochondrial respiratory chain diseases. We utilize human patient cells, as well as simple model animals including *C. elegans* (nematodes), *D. rerio* (zebrafish), and mice, with the goal of translating our findings back to the benefit of patients in clinical treatment trials.

- **Potential summer research project:**
 - (1) Validating novel human disease genes identified by next generation sequencing;
 - (2) Testing the efficacy and toxicity of new therapies in existing cell and animal models of mitochondrial disease in our group; and
 - (3) Developing new methods for evaluating the range of biological dysfunction that occurs in mitochondrial disease.
- **Students will learn the following techniques:**
 - (1) Zebrafish studies of mitochondrial disease mechanisms and therapies;
 - (2) *C. elegans* studies of mitochondrial disease therapeutic modeling; and
 - (3) Human cell studies of mitochondrial disease gene validation, mechanism, and therapies.

Please click [here](#) to read more about Dr. Falk