CHRD 2021: Abstract & Poster Submission Form

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Research Category:

- ◉ Basic Science
- ○ Clinical
- ○ Community Health / Policy

What was your role in the project?

- ☑ Design
- ☑ Perform Experiments
- ☑ Analyze Data
- ☐ Write Abstract
- ☐

Presenter Status:

- ◉ Undergraduate Students
- ○ Masters Student
- ○ PhD Student
- ○ Post-Doctoral Fellows
- ○ Residents
- ○ Non-Trainee

Title

Does Bowen-Conradi Syndrome Include Metabolic Rewiring?

Background

Bowen-Conradi Syndrome (BCS) is a rare genetic disorder in the Hutterite. It is due to a sequence variant in the ribosome assembly protein and SSU processome component EMG1. This ribosome assembly disorder or ribosomopathy presents with severe developmental delay and death in infancy. A recently completed proteomic analysis of our yeast BCS model surprisingly identifies mitochondrial proteins as the most differentially expressed relative to wild-type cells. This suggests that metabolic and mitochondrial rewiring may be occurring in BCS, in line with recent findings in other ribosomopathies.

Objective

Our aim is to further explore if BCS includes metabolic rewiring as part of the disease pathogenesis.

Methods

Using our yeast BCS model, we are assessing metabolic function by monitoring growth under different stressor conditions and metabolic substrates.

Results

Growth analysis following heat shock finds that BCS model cells are sensitive to high temperatures, suggesting a dysregulation to the proteasome in protein homeostasis. Reduced growth after peroxide exposure reveals that BCS cells are sensitive to reactive oxygen stress. This may suggest a change in oxidative phosphorylation.

Conclusion

Our preliminary results find that BCS model cell display proteotoxic and reactive oxygen species stress. These suggest a likely metabolic and mitochondrial rewiring in BCS. Further experiments will assess ATP production along with the ability to grow on different metabolic substrates.

Authors

- For each author, please click "**[+] Add Item**" and provide the author's information

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