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
- ☑ New graduate student on the project

Presenter Status:

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

Title

The Ribosome Assembly Disorder Bowen-Conradi Syndrome Includes Changes in Protein Translation

Courtney Geer, Derrek Harris, Joel Hard, and J. Michael Charette

Background

Bowen-Conradi Syndrome (BCS) is a ribosome assembly disorder (or ribosomopathy) present in the Hutterite population. BCS presents with severe developmental delay, a failure to thrive, and death in infancy. BCS is due to a D86G variant in the pseudouridine methyltransferase and SSU processome protein EMG1. This protein is responsible for both ribosome assembly and methylation of a pseudouridine in the mRNA decoding P site of the small subunit of the ribosome. This 18S rRNA pseudouridine is post-transcriptionally hypermodified and critical to the translational fidelity of the ribosome in mRNA decoding. As with other ribosomopathies (such as Diamond-Blackfan anemia and Treacher-Collins syndrome), the consequences of ribosome mis-assembly on translational fidelity and alterations to the proteome has been under-investigated.

Objective

Here, we investigate suspected translation defects in BCS.

Methods

Using a yeast model system of BCS, we quantified translation by assessing total protein and using the translation terminating antibiotic. The translation of new proteins was monitored by total protein staining of SDS-PAGE blots and by western blot analysis using an anti-puromycin antibody.

Results

Total protein quantification and the puromycin translation assay reveals that BCS cells are translationally compromised in a number of differ ways. This includes a decrease in overall translational capacity, along with a likely change in translational preferences.

Conclusion

Our results reveal that BCS cells are translationally compromised. This is consistent with recent results suggesting that the rRNA target site of Emg1 is hypomodified in all cancers. Ongoing investigations will further characterize the nature of the translational defect (mis-incorporation, frame-shifting, stop codon read-through).

Authors

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

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