CHRD 2022: Abstract & Poster Submission Form

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

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

Role in the project
- ☑ Design
- ☑ Perform Experiments
- ☑ Analyze Data
- ☑ Write Abstract
- ☐

Title
The Bowen-Conradi Syndrome Protein EMG1 Contains an N-Terminal Intrinsically Disordered Region

Background

Bowen-Conradi Syndrome (BCS) is a rare genetic disorder that affects 1/355 live births in the Hutterite populations of the North American Prairies, presenting with severe developmental delay that leads to death in infancy. Specifically, BCS is a ribosome assembly disorder or ribosomopathy caused by a genetic variant in the EMG1 protein, an essential ribosome assembly protein and component of the SSU processome. We have identified a heavily post-translationally modified intrinsically disordered region (IDR) in the N-terminal region of EMG1.

Objective

Our objective is to validate the presence of this novel IDR and determine its function in ribosome assembly.

Methods

We used IDR predictors (Metapredict and PONDR) and protein structural analysis to identify and validate the novel IDR. We then investigated the function of this IDR by creating a series of Emg1 constructs of 5 amino acid truncations of the IDR region along with the IDR region alone in yeast over-expression plasmids. These plasmids were transformed into our yeast BCS model. Functional analysis of this IDR was done by growth monitoring, western blot, and ribosomal RNA analysis. Yeast two-hybrid (Y2H) and co-IP methods to investigate the role of the Emg1 IDR in mediating protein-protein interactions are ongoing.

Results

Analysis using disorder predictors reveal that Emg1 contains 11.9% disorder, with the disorder occurring at the N-terminal region of Emg1. Growth analysis reveal that the IDR is necessary for both cell growth and protein stability of Emg1. Ribosomal RNA analysis indicate that the Emg1 IDR may play an important role in ribosome processing and assembly.

Conclusion

Our results indicate that the N-terminal IDR of Emg1 is important for cell growth, Emg1 protein stability, as well as ribosome assembly and processing. In determining the structure, function, and regulation of Emg1 in ribosome assembly, the disease mechanism of BCS can be better understood.

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Authors

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