

ABSTRACT SUBMISSION FORM

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CHR D 2020: Abstract Submission Form

Submitter Name

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Title

The Intrinsically Disordered Region of the Bowen-Conradi Syndrome Protein Emg1 and Its Function in Ribosome Assembly

Background

Bowen-Conradi Syndrome (BCS) is a rare genetic disorder affecting 1/355 live births in the Hutterite population of the Canadian prairies. This ribosome assembly disorder or ribosomopathy presents a severe developmental delay with death in infancy. BCS is due to a genetic variant in the Emg1 protein, an essential ribosome assembly protein and SSU processome component. We have identified an N-terminal heavily post-translationally modified intrinsically disordered region (IDR) in Emg1 which we believe is involved in mediating protein-protein interactions with another ribosome assembly and SSU processome component Utp2/Nop14.

Objective

Our aim is to validate the presence of this novel IDR and to determine its function in the role of Emg1 in ribosome assembly.

Methods

We used IDR predictors to identify the novel IDR which we validated by amino acid compositional bias and protein structural analysis. We then created a series of Emg1 constructs consisting of 5 amino acid truncations of the IDR region along with the IDR region alone. These were transformed into our yeast BCS model for functional characterization along with a yeast two-hybrid analysis of protein-protein interactions.

Results

Analysis of the amino acid composition of the IDR identified an increased number of charged amino acids, as expected. Structural analysis based on available cryoEM structures of the SSU processome identified the IDR as mediating protein-protein interactions with an IDR region in Utp2, one of the few IDR-IDR mediated protein-protein interactions described. Experimental validation of the function of the IDR is

ongoing.

Conclusion

These results will allow us to determine the function of Emg1 in ribosome assembly which is required in order to better understand the disease mechanism of BCS.

Theme:

Basic Science

Do you have a table/figure to upload?

No

Are you willing to participate in Goodbear's Den?

Yes

Presenter Status:

Undergraduate Students

What was your role in the project?

performed experiments, analyzed data, helped write abstract

Authors

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