CHRD 2024: Abstract Submission Form

Presenter Name
Katelyn Panchoo

Presenter Status
Undergraduate Students

Role in the project
Design
Perform Experiments
Analyze Data
Write Abstract

Research Category
Basic Science

Title
Investigating ELFN1 Variants in Relation to Neurodevelopmental Disorders

Background
Extracellular leucine-rich repeat and fibronectin type 3 domain containing 1, ELFN1, is an important synaptic adhesion molecule (SAM) that aids in neuronal communication. Previous studies have found that ELFN1 modulates the activity of a particular class of receptors known as group III metabotropic glutamate receptors (mGluRs), broadening our understanding of SAM function as more than structural components. Mouse studies have also found that downregulation of mGluRs at the synapse can result in excess amounts of glutamate being released, leading to symptoms of neurodevelopmental disorders such as hyperactivity, compulsivity, and anxiety. Interestingly, these symptoms have been found in children with different mutations of ELFN1. My research investigates an intracellular (ELFN1-ΔCT) and extracellular mutation (ΔNT-ELFN1).

Objective
The purpose of my experiment is to examine whether the expression of these variants is altered, affecting their interaction with mGluRs and leading to these neurodevelopmental disorders.

Methods
My experiments were done through western blot analysis, which separates proteins based on molecular weight or size via gel electrophoresis. After transferring to a solid support matrix, the western blot was imaged and densitometric analysis was conducted to quantify expression levels.

Results
Through quantitative analysis, we found ELFN1-ΔCT expressed significantly lower than ΔNT-ELFN1 and was trending towards a significant difference from wild-type ELFN1.

Conclusion
Lowered expression of ELFN1 may indicate a reduced number of synaptic ELFN1/mGluR interactions, resulting in the excess release of glutamate and behaviours found in neurodevelopmental disorders. Gaining a better understanding of the mechanistic consequences could help us identify better therapeutic strategies for patients harboring these mutations.

Do you have a table/figure to upload?
Yes

Authors

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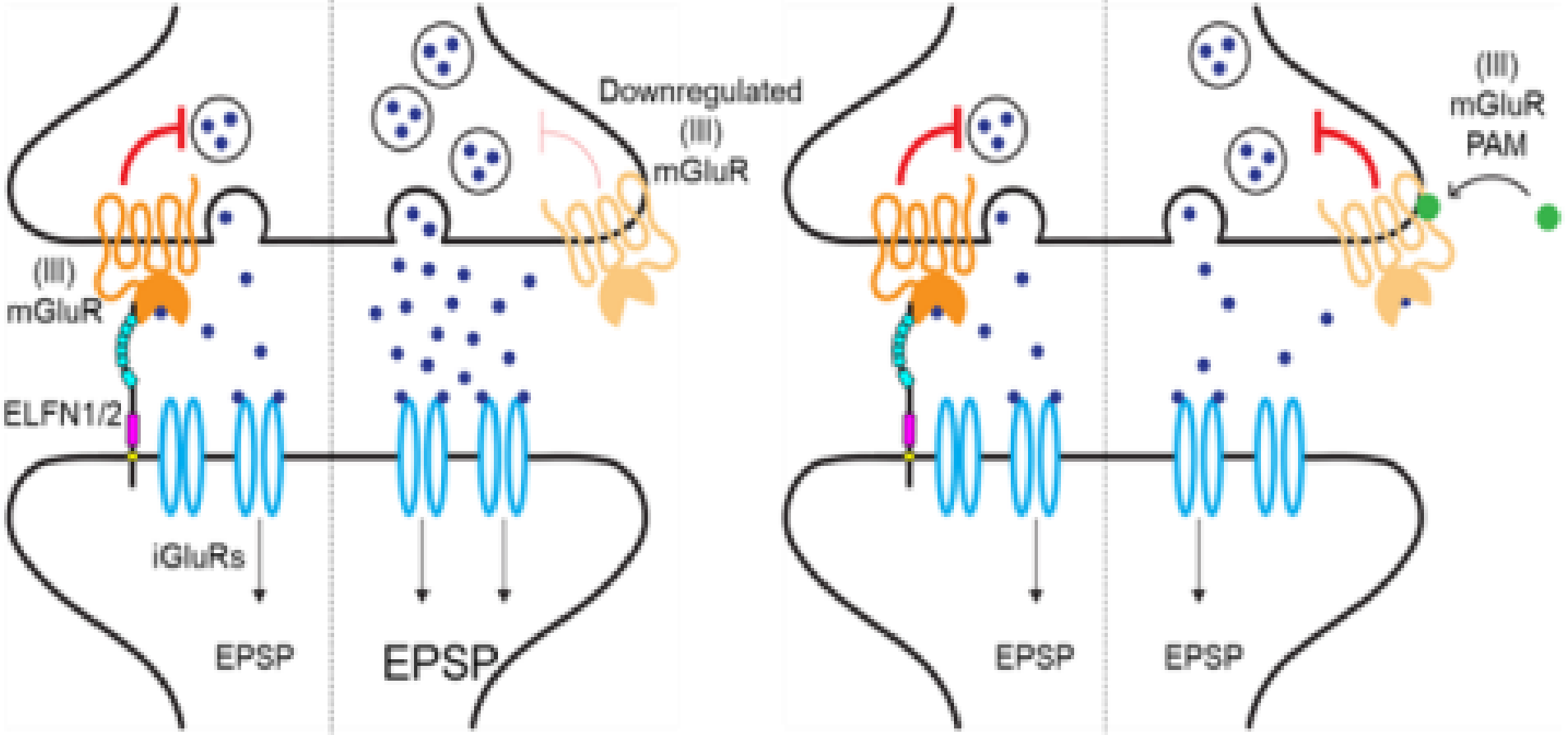
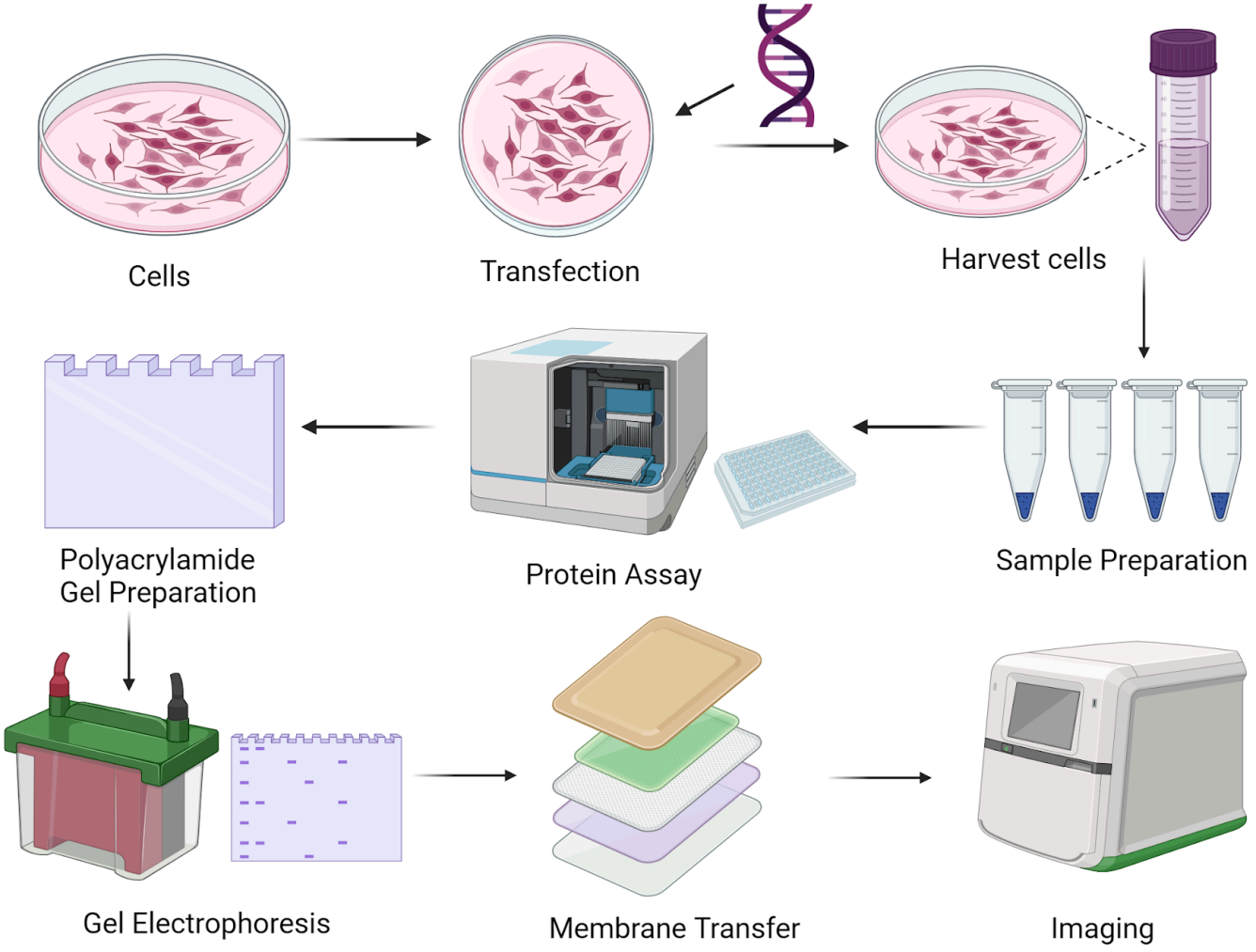

Downregulated
(III)
mGluR
(III)
mGluR
PAM
(III)
mGluR
ELFN1/2
iGluRs
EPSP
EPSP
EPSP
EPSP
• Normal behavior
• Hyperactivity
• Compulsivity
• Anxiety
• Normal behavior
• Normal behavior


Cells
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Membrane Transfer
Imaging