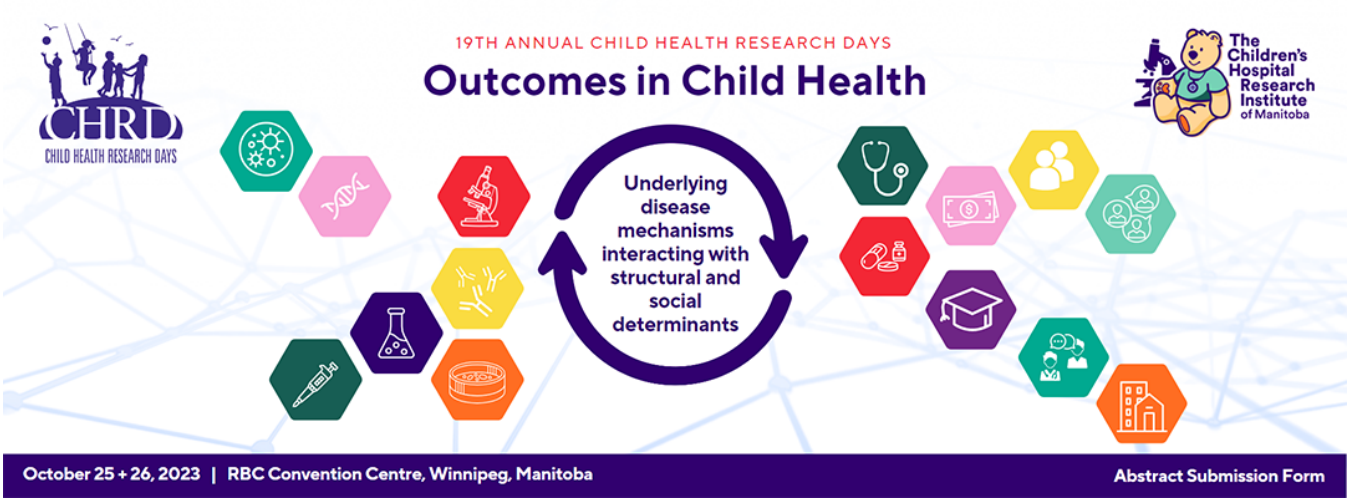

CHRD 2023: Abstract Submission Form

Submitter Name
Harsimran Kaur

Presenter Name
Harsimran Kaur

Presenter Status
Undergraduate Students

Research Category
Basic Science

Role in the project
Perform Experiments
Analyze Data
Write Abstract

Title
Characterizing the neuronal role of histone acetyltransferases KAT6A and KAT6B using Drosophila melanogaster

Background
Dominant variants in either histone acetylation genes, KAT6A or KAT6B, cause neurodevelopmental conditions (Arboleda-Tham syndrome and SBBYSS syndrome) with overlapping features. Children show developmental delay, motor and speech impairment, and some develop seizures.

Objective
We will study these disorders by examining the function of the single Drosophila ortholog, enok, in neurons.

Methods
We will study the effects of developmental and adult-specific neuronal knockdown of enok using the UAS-GAL4 system in flies. This bipartite system allows for the tissue-specific knockdown of our gene of interest, enok. We drove enok-RNAi using ubiquitous (Act-GAL4) and neuronal GAL4s (elav-GAL4, nSyb-GAL4) and examined lethality, lifespan, climbing, and sensitivity to seizures. Moreover, we will generate transgenic flies that express KAT6A and KAT6B using site-directed mutagenesis.

Results
The neuronal knockdown of enok showed significant phenotypes like seizures and climbing defects with elav-GAL4. With another pan-neuronal driver, nSyb-GAL4, the lifespan of the flies was diminished, and

some RNAi lines caused lethality. We successfully generated 18 KAT6A and KAT6B variants.

Conclusion
The function of enok in neurons is essential for the proper development of flies and knockdown can cause detrimental deficits. Future studies will examine how KAT6A and KAT6B disease variants function in vivo.

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