CHRD 2024: Abstract Submission Form

Presenter Name
Harsimran Kaur

Presenter Status
Non-Trainee

Role in the project
Design
Perform Experiments
Analyze Data
Write Abstract

Research Category
Basic Science

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. We propose to study these disorders by examining the role of the single Drosophila ortholog, enok, in neurons.

Objective
We will study the effects of developmental and adult-specific neuronal knockdown of enok using the UAS-GAL4 system in flies. Moreover, we will generate transgenic flies that express KAT6A and KAT6B and disease variants and assess functional outcomes in vivo.

Methods
We drove enok-RNAi using ubiquitous (Act-GAL4) and neuronal GAL4s (elav-GAL4, nSyb-GAL4) and examined lethality, lifespan, climbing, and seizure behaviour. The KAT6A/B variants were made using site-directed mutagenesis.

Results
The neuronal knockdown of enok showed significant phenotypes like seizures and climbing defects with elav-GAL4. With nSyb-GAL4, the lifespan of the flies was diminished, and some RNAi lines caused lethality. We successfully generated 19 KAT6A/B variants.

Conclusion
The function of enok in neurons is essential for the proper development of flies and knockdown can cause detrimental deficits.

Do you have a table/figure to upload?
No

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