Missense Mutation in CSNK1D, CSNK1E, ARNTL, and Clock Genetics and The Variations Among Them

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Abstract

Within the realm of biomedical sciences, coding patterns and relationships have been a big advancement in technology. Being able to map miscodings and misfires has been a very pertinent exercise when it comes to gene expression and potential gene therapies. The circadian rhythm is a large mechanism that has been known to regulate chemical processes in the body. The research being conducted looks at specific genes that can affect the period of the circadian rhythm. This clock is an important facet in cell regulation and chemical emission. Out of the numerous amino acid sequences a missense can cause substantial changes in physiological functionality and other types of debilitation. The future of gene technology is here and now. With the exploration of genetic mapping, scientists and researchers can uncover comprehensive patterns that lead to the solution of systemic dysfunction. The applications of the science could lead to the prevention of impairing genetic sequences potentially In Vivo and In Vitro.

Research Questions

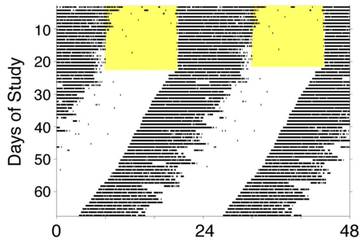
How does genetic mutation affect activity?
What type of mutations are most common?
What patterns can we infer based on mutations in mice?

Methods

Mice Husbandry
CRISPR amino acid insertion/deletion

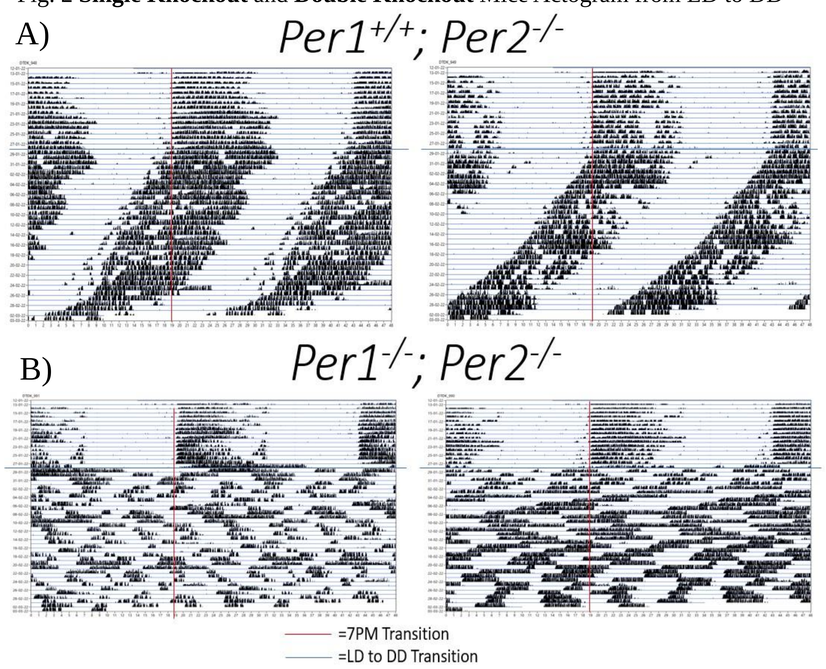
Results

Fig. 1 **Wild-Type** Mice Actogram from LD to DD



This is the normal phenotypical expression of behavior in wild-type mice. The normal circadian rhythm puts them on a clock that regulates on a sub-24-hour clock

Fig. 2 **Single Knockout** and **Double Knockout** Mice Actogram from LD to DD

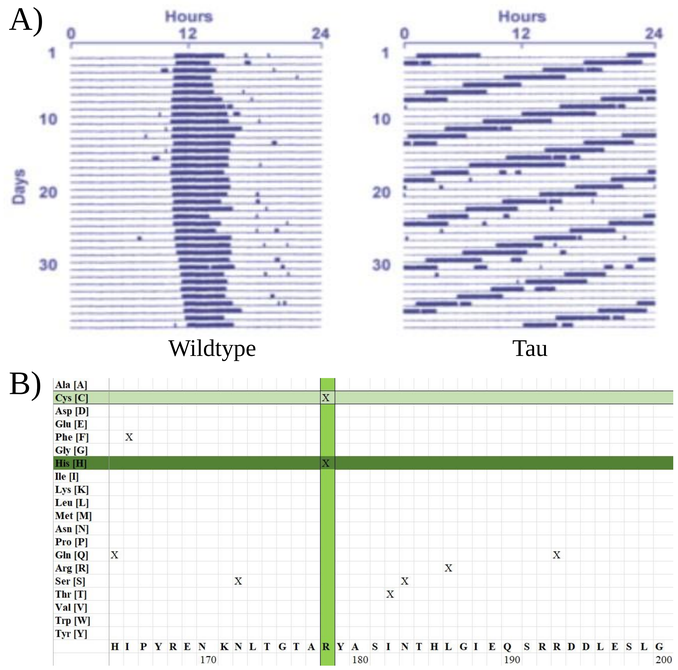


A) The Single Knockout mice are an example of the magnitude of phenotypical change in the mice with single-gene mutation
B) The Double Knockout mice are an example of a larger magnitude of change in phenotype, leading to arrhythmic activity

Summary

1. Mutations are inevitable and occur often in nature causing changes in circadian rhythmic activity
2. Certain genes are associated specifically with rendering altercations in sleep/activity in mice
3. Articulating gene correspondence has future implications for gene therapy and other associated techniques

Fig. 3 **Wildtype** Hamster vs. **Tau** Hamster



A) The R178 or Tau mutation has been proven to alter the circadian rhythm in hamsters. This mutation is seen in mice and now humans. It is a single-gene mutation proven to cause an atypical phenotype.
B) The amino acid that codon 178 synthesizes is Arginine [R], and has a reported missense of Cysteine [C] or Histidine [H]. These single-gene mutations are what cause the phenotypical changes and lead to alterations in the circadian rhythm.

Future Considerations

- Gene-specific therapy
  - Insomnia
  - Sleep Apnea
- Analysis of genetic disruptions that can diagnose disorders such as schizophrenia
- Small Molecules for Circadian rhythm modulation in Casein Kinase can lead to control of circadian rhythm