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## Summary

- Is there a difference in the marker of dystonia (movement disorder) between males and females?
- Expression of *Torsin* was quantified via qRT-PCR in female and male fruit flies.
- A 61.7-fold lower expression of *Torsin* was found in female relative to male fruit flies.

## Abstract

Dystonia is a common movement disorder more likely to affect women than men. This experiment quantified the sex-specific expression of *Torsin*, a dystonia-linked gene, in *Drosophila melanogaster* (fruit fly) brains under controlled conditions using qRT-PCR. Female fruit flies show 61.7-fold lower gene expression of *Torsin* relative to males, revealing a sex-specific pattern of mRNA expression.

## Introduction

### Hypothesis -

Due to the higher incidence of dystonia in women relative to men, *Torsin* expression will be higher in female relative to male fruit flies.

### Model Organism -

- Fruit flies can produce new generation of offspring within 12 days<sup>1</sup>.
- Are inexpensive to maintain, and easily differentiated by lifestage and sex.
- 75% of human disease genes are found in fruit flies<sup>1</sup>.



Figure 1. Female (left) and Male (right) *Drosophila melanogaster*<sup>2</sup>.

### Target Gene -

- *Torsin* in fruit flies encodes for an ATPase that regulates dopamine levels<sup>3</sup>.
- Mutations in the human counterpart of this gene are linked to dystonia, a neurological movement disorder<sup>4,5</sup>. This disorder is more common in women<sup>6</sup>.

## Methodology

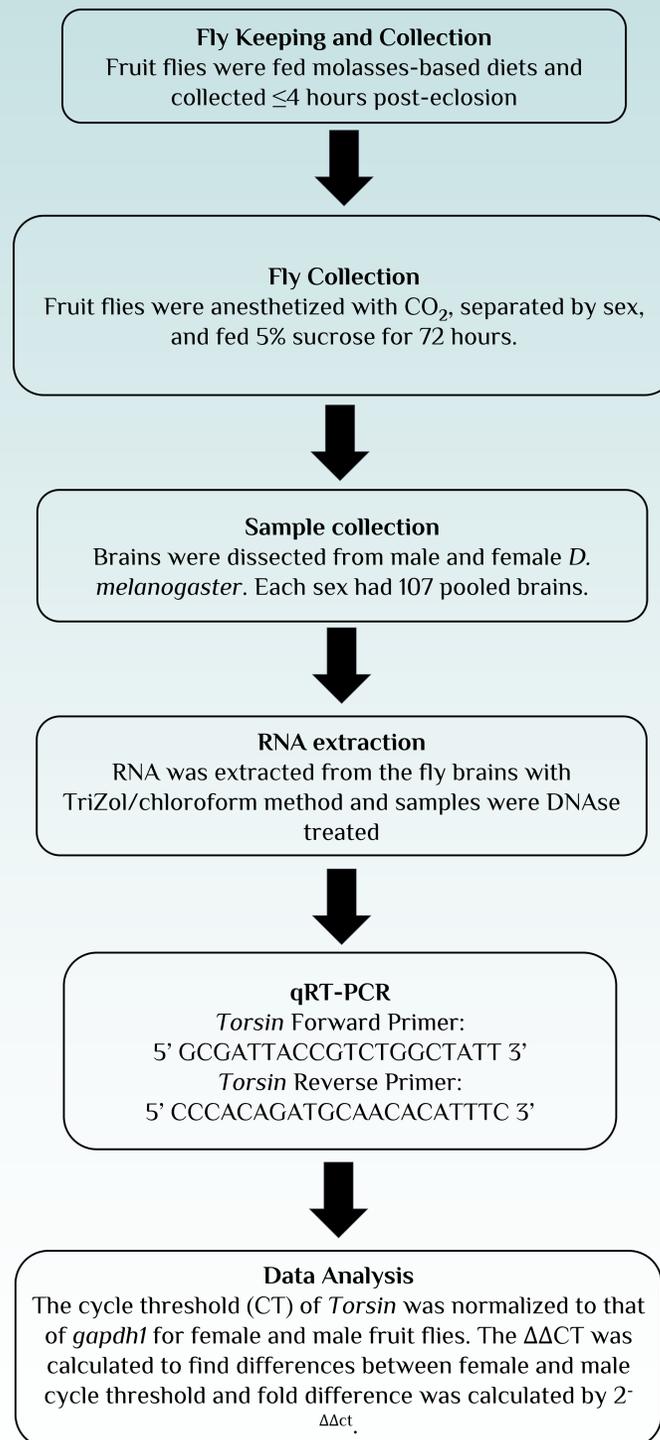


Figure 2. Flowchart of experimental design.

## Results

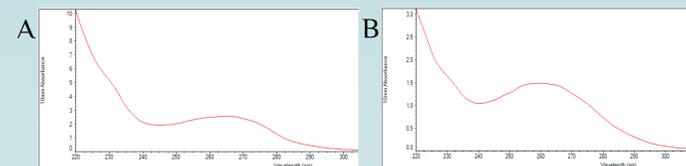


Figure 3. Total RNA yield and concentration for male and female fruit fly brain samples: (A) Total female RNA yield, (B) total male RNA yield, (C) and total RNA concentration of brain samples.

Sex	[RNA]	A260/A280
Male	97.6 ng/μl	1.97
Female	58.9 ng/μl	2.04

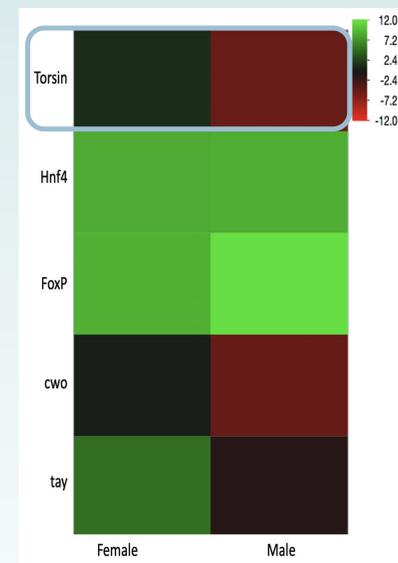


Figure 4. *Torsin* mRNA expression is 61.7-fold lower in females relative to males. Red indicates higher expression; green indicates lower expression. All genes normalized to reference gene *gapdh1*.

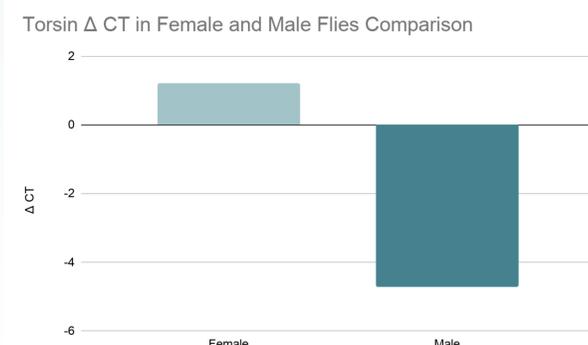


Figure 5. *Torsin*  $\Delta$ CT is lower in males relative to female fruit flies:  $\Delta$ CT values is 1.222 for female and -4.726 for males, with a more negative  $\Delta$ CT correlated to a higher mRNA level. Normalized to reference gene *gapdh1*.

## Discussion and Conclusion

- *Torsin* mRNA expression is 61.7-fold lower in female *D. melanogaster* relative to males.
- This contradicts the hypothesis; although dystonia affects women more than men, the results indicate *Torsin* is less expressed in female fruit flies compared to males.
- Sexually dimorphic expression of *Torsin* suggests sex-specific differences in dopamine level regulation.

This study helps determine baseline differences in *Torsin* mRNA expression between sexes that need to be accounted for when studying stress responses.

### Study Limitations

- Limited sample size and replicates.
- Only one developmental stage (post-eclosion) was studied.

### Future Directions

- In-depth genomic comparisons between human and *D. melanogaster* dystonia associated genes.
- *Torsin* gene expression analyzed across more life stages of *D. melanogaster*.
- Analyze baseline *Torsin* protein levels.



Figure 6. 3D-Structure of *Torsin-1A*<sup>7</sup>. Mutation at position 302/303 in *Torsin-1A* causes primary dystonia<sup>7,8</sup>

## References

