

Modeling Robinow Syndrome in the fruit fly

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Background: Robinow syndrome (RS)

Rare developmental syndrome

- Affecting 1:500 000
- Craniofacial symptoms
- malformed limb bones



Zhang et al., 2022



White et al., 2015



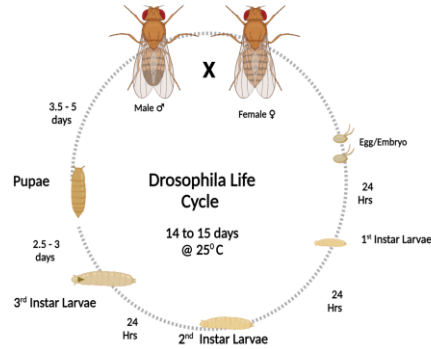
Zhang et al., 2022

Al Kaissi et al., 2020



Human gene, mode of inheritance AD = autosomal dominant AR = autosomal recessive	Function
WNT5A , AD	PCP/Wnt ligand
FZD2 , AD	Wnt co-receptor
DVL1 , AD	Wnt adaptor protein
DVL2 , AD	Wnt adaptor protein
DVL3 , AD	Wnt adaptor protein
ROR2 AR	PCP/Wnt co-receptor
NXN , AD	Stabilizer of DVL

Background: Why the fly?

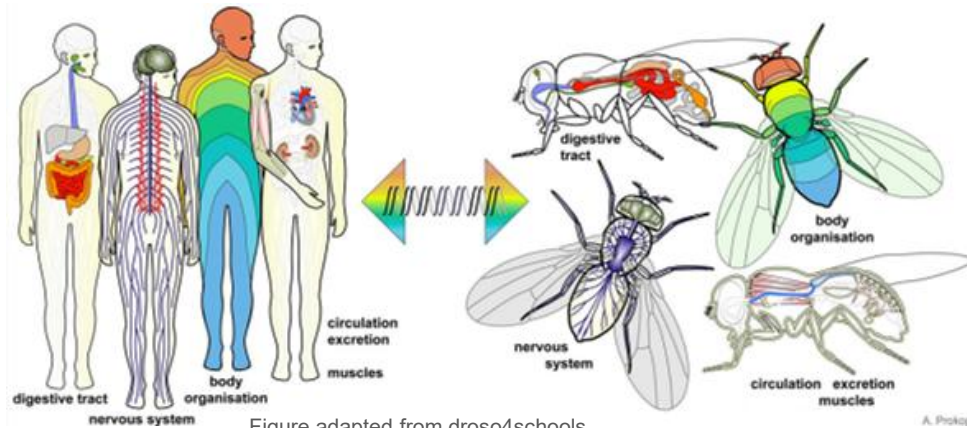


We share many organ systems

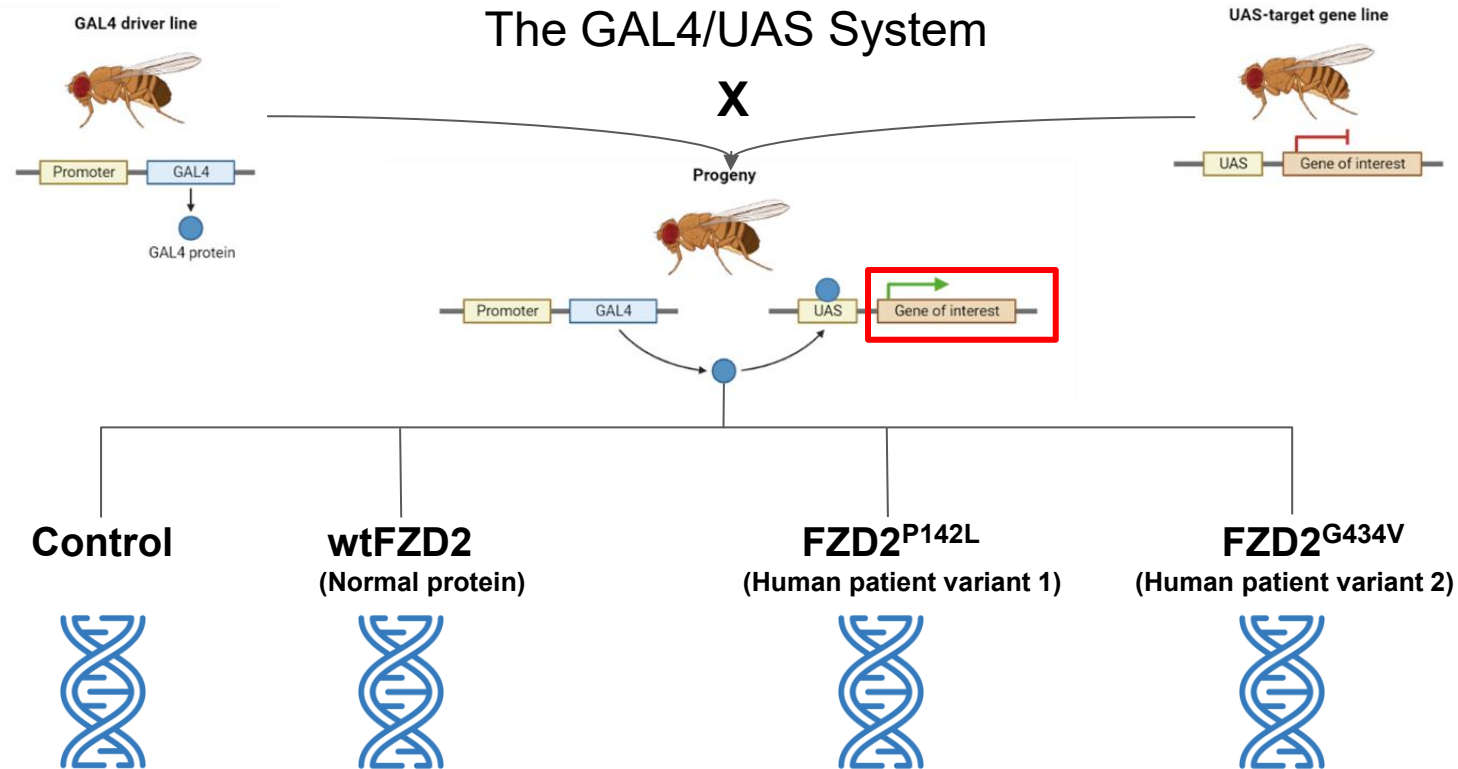
Short Life cycle

Easy to manipulate genetically

We share 75% of our disease causing genes

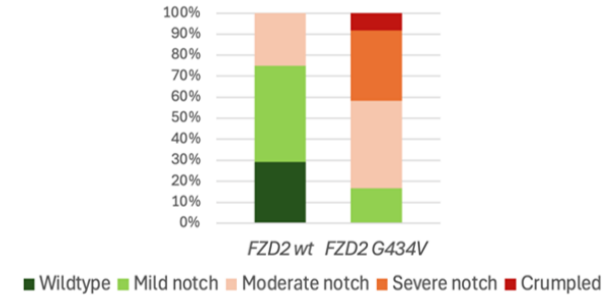
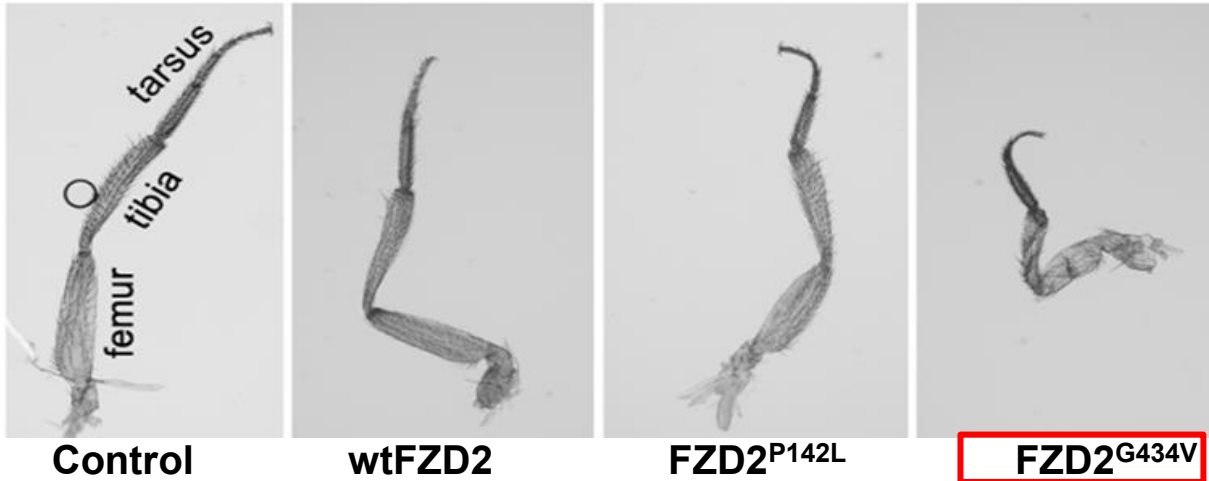


Methods: Genetic tools

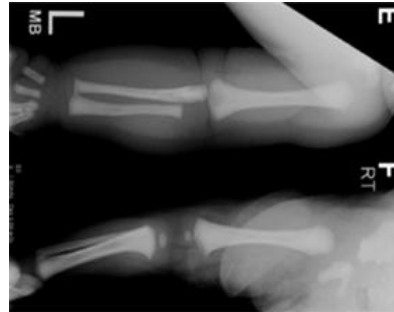


Goal: Generate fruit fly strains that express the human patient specific FZD2 variants to assess their effects compared to the normal protein

Results/Conclusion



Kecler-Pietrzyk., 2017



Zhang et al., 2022

Thank you



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