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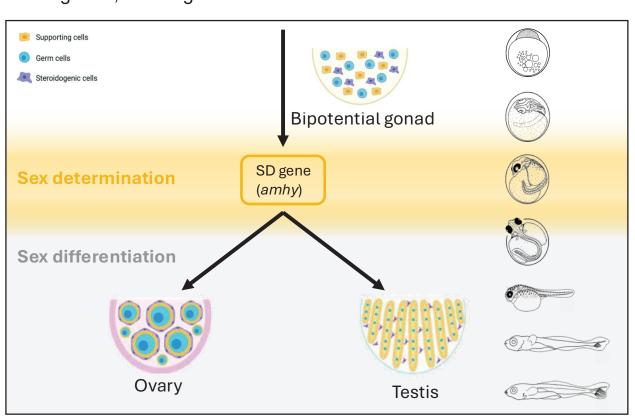
Burtchaell Award
Ph.D. Student, Genetics
Second Year ARCS Scholar



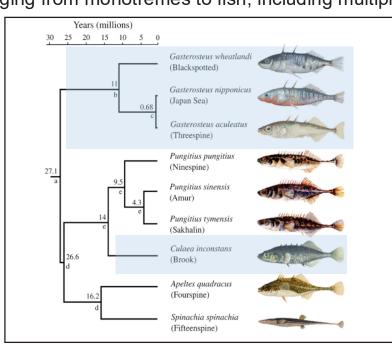
Understanding how regulation of a sex determining gene impacts development

Reproductive health and fertility of an individual requires development of differentiated male or female gonads. During genetic sex determination, a sex determining gene on a sex chromosome initiates sex-specific development. Disruptions to sex determination and differentiation lead to disorders of sex development (DSD) in humans. I study how sex determining genes are regulated to act in the correct time and place for normal development.

During genetic sex determination, a sex determining gene on a sex chromosome acts on an undifferentiated gonad, initiating either a male or female network of sex differentiation genes.



Amh is the most common sex determining gene in vertebrates. It has convergently evolved this role in species ranging from monotremes to fish, including multiple stickleback species.

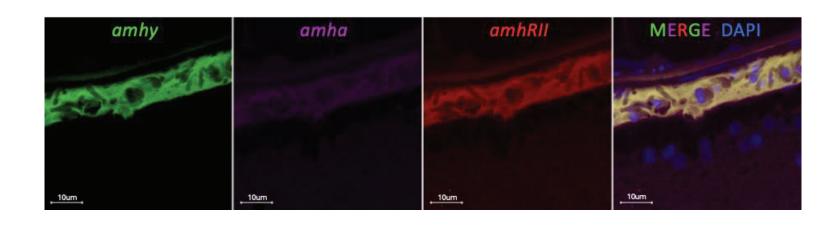


- To repeatedly evolve this role, amhy must repeatedly
- 1. effect the bipotential gonad during a narrow window of development.

2. integrate into a conserved network of sex differentiation genes.

How does amhy effect the bipotential gonad during a narrow window of development?

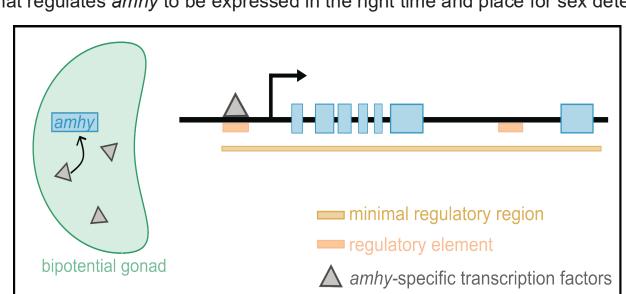
I am characterizing when and where *amhy* is expressed in development of the threespine stickleback. I see expression of *amhy* in the somatic cells of the bipotential gonad.



In humans, misexpression of the sex determining gene can lead to differences of sex development (DSD) which has health and fertility consequences.

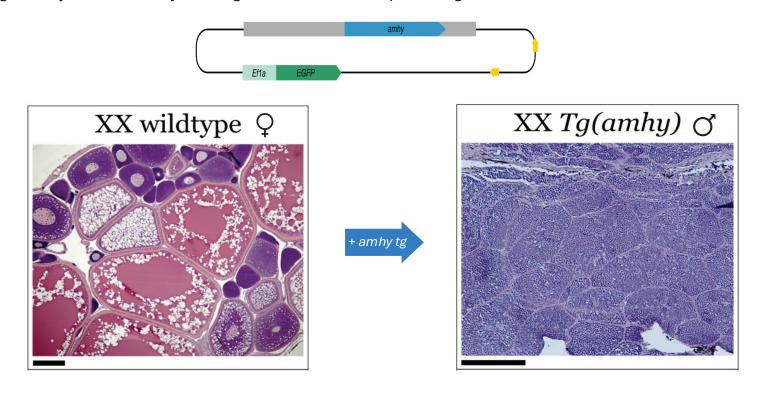
How does amhy effect the bipotential gonad during a narrow window of development?

Timing and location of gene expression are controlled by regulatory elements surrounding a gene. What regulates *amhy* to be expressed in the right time and place for sex determination?



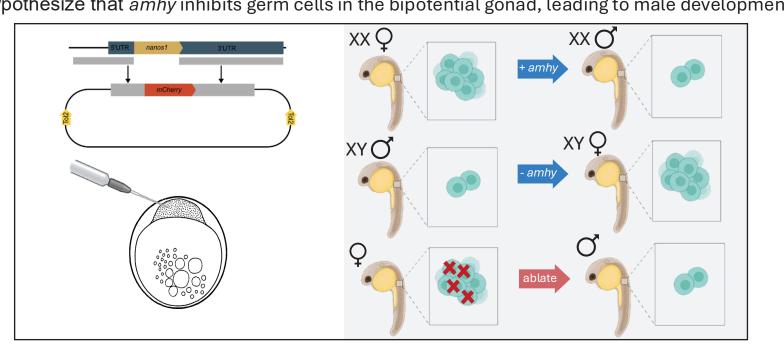
In humans, some DSD are caused by mutations in regulatory elements of the sex determining gene. Many DSD still have unknown genetic causes. Exploring regulation of other sex determining genes may elucidate other genetic causes.

To elucidate the regulatory elements that control *amhy*, I use transgenics to modify the regulatory region of *amhy*. I measure whether an injected transgene contained necessary regulatory elements by seeing if XX fish develop male gonads.



How does *amhy* integrate into a conserved network of sex differentiation genes and induce male development?

I hypothesize that *amhy* inhibits germ cells in the bipotential gonad, leading to male development.



Investigating how sex determining genes activate the sex differentiation pathway can help us understand how alterations in the sex differentiation pathway cause DSD in humans.