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An evolutionary geneticist looks at how small genetic changes can have big evolutionary effects.

Vertebrates are diverse in both form and function. What genetic alterations underlie this diversity? Evolutionary modifications in brain size or limb length, for example, involve changes in the complex developmental processes that give rise to these structures. Work in fish elegantly illustrates how a single genetic change can have profound effects on morphological evolution.

David Kingsley at Stanford University in California and his colleagues focused on threespine sticklebacks (Gasterosteus aculeatus), which exist in both marine and freshwater ecosystems (Y. F. Chan et al. Science 327, 302-305; 2010). Most bear pelvic spines on their underside; however, several freshwater populations have lost these structures.

In previous work, the authors suggested that the gene Pitx1 is involved in pelvic reduction in these fish. Now they show that deletion of a sequence that activates Pitx1 expression in the pelvis is directly responsible for the loss of pelvic spines in sticklebacks. In a simple but powerful experiment, they demonstrate that introducing the Pitx1 gene to these pelvic-reduced fish, under the control of the intact Pitx1 regulatory element, is sufficient to restore pelvic spines.

The Pitx1 regulatory deletion has occurred independently in at least nine stickleback populations. This may be because the Pitx1 regulatory element is in a particularly fragile region of the genome that is prone to deletion. Moreover, population-genetic evidence suggests that this recurrent loss of Pitx1 pelvic expression confers a strong fitness advantage — possibly because insects that prey on sticklebacks can grab onto pelvic spines.

This study illustrates the power of laboratory genetics in understanding evolutionary mechanisms, and by doing so provides a conceptual basis for future functional studies of the evolutionary process.

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