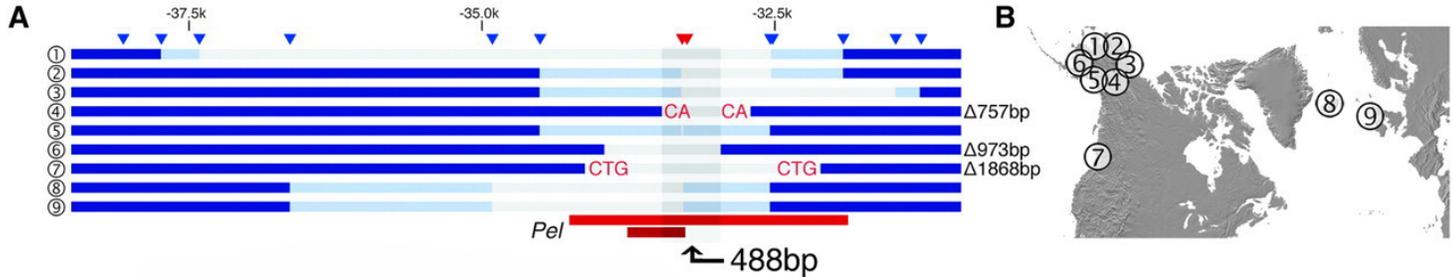




Pelvic Evolution in Sticklebacks



Caption: The results of SNP genotyping in nine stickleback fish populations with reduced or absent pelvises (source locations shown in Figure B). Triangles indicate SNP markers that were used to identify the approximate location of deletion mutations. Dark blue bars indicate that the SNP markers on both sides of that section were present, thus that section of DNA was present. Light blue bars indicate that only one of the two SNP markers on either side of the section was present, meaning that the deletion started somewhere in that section. Light gray horizontal bars indicate that both SNP markers on each side of that section were missing, meaning that the section of DNA was wholly deleted. Three sequences, 4, 6, and 7, were sequenced completely and the lengths of the deletions are indicated to the right. Red bars indicate the location of two noncoding regions (*Pel*-2.5-kb and *Pel*-501-bp), based on DNA constructs from marine fish, which are able to drive gene expression specifically in the pelvis. The gray-shaded vertical rectangle indicates the 488-bp region where all the deletions overlap, which includes SNP markers indicated in red.

BACKGROUND INFORMATION

Species can undergo major changes in body morphology as they adapt to their environments, but the molecular mechanisms responsible for these changes are not always known. A classic example of a major evolutionary change in morphology is the modification, and sometimes even complete loss, of limbs and fins. Pelvic hind limbs have been lost in many species, including whales, manatees, and some amphibians, reptiles, and fish. Most members of a fish species called the threespine stickleback (*Gasterosteus aculeatus*) are marine fish that have a pelvis which supports prominent serrated spines that protrude from the underside, deterring predatory fish from eating them. But over two dozen geographically isolated freshwater stickleback populations either partially or completely lack the pelvis. One reason that pelvis and spine loss may occur is because some freshwater populations live in low calcium environments where building a pelvis may be metabolically costly, and are preyed upon by insects that can grasp onto spines, rather than by fish that can be deterred by the spines.

To determine the genetic mutations responsible for stickleback pelvic reduction and loss, and to explain how these changes arose separately in different populations, scientists looked for genetic changes in a chromosome region that controls most of the differences in pelvis size when marine and freshwater sticklebacks are crossed with each other in the laboratory. The key chromosome region contains a gene called *pituitary homeobox transcription factor 1* (*Pitx1*), which is required for both pituitary and hind limb development. Sequencing and expression studies have shown that the *Pitx1* gene of freshwater fish encodes a normal protein, but it fails to be expressed in the developing pelvic region, suggesting a possible regulatory change. The scientists conducted SNP (single-nucleotide polymorphism) genotyping of nine different stickleback populations with reduced pelvises to find genetic mutations that may be responsible for altered *Pitx1* expression in freshwater fish. SNP genotyping identified a variety of deletion mutations in a noncoding region of the genome called *Pel*, located upstream of the *Pitx1* gene. *Pel* normally enhances *Pitx1* expression during pelvic fin development but fails to do so when mutations render it nonfunctional. The above figure shows the locations of the deletion mutations.